

## Chapter 7.1

# Principles of Inheritance and Variation

The term genetics (Gk. Genesis – descent) was coined by Bateson (1906). Genetics is the study of principles and mechanism of heredity and variations. The resemblance amongst offspring is never 100% (except in monozygotic twins) due to reshuffling of chromosomes and their genes.

Table : 7.1-1

Father of genetics (classical genetics)	Mendel
Father of modern genetics / Animal genetics	Bateson
Father of experimental genetics / <i>Drosophila</i> genetics	Morgan
Father of human & physiological genetics	Garrod
Father of quantitative inheritance	Kolreuter
Father of <i>Neurospora</i> genetics	Dodge
Father of genetic Engineering	Paul Berg

## Heredity

Heredity is the study of transmission of genetic characters and variations from one generation to the next. Heredity involves the transfer of chromosomes from parents to offspring or one individual to another. Therefore, chromosome is the base of heredity. The physical basis of heredity are genes while chemical basis of heredity is DNA.

### Pre-Mendelian view points

**Vapour theory (Pythagoras)** : Different body parts produce minute particles.

**Fluid theory** : Empedocles, proposed that each body part produces a fluid. The fluid of different body parts of the two parents mixes up and is used in the formation of embryo.

**Preformation theory** : Malpighi believed that homunculus or miniature individual is present in sperm or egg. Antony Von Leeuwenhoek was first to observe human sperm.

**Particulate theory** : Maupertuis proposed that the body of each parent gives rise to minute particles. These particles unite together to form the daughter individual.

**Encasement theory** : Charles Bonnet and his supporters presumed that every female contains within her body miniature prototypes of all the creatures which would descend from her, one generation within the other, somewhat like a series of chinese boxes. This was named as encasement theory.

**Theory of epigenesis** : Wolff proposed that the germ cells contain definite but undifferentiated substances, which after fertilization, become organised into various complex body organs that form the adult. This idea was referred to as epigenesis.

**Pangenesis theory** : Proposed by Charles Darwin (1868) according to this theory every cell, tissue and organ of animal body produces minute invisible bodies, called gemmules or pangenes. They can produce offsprings.

**Weismann theory of germplasm** : August Weismann (1889) suggested the theory of continuity of germplasm. He described reproductive cells as germplasm and rest of the body as somatoplasm.

Pre-Mendelian theories of inheritance are also called theories of blending inheritance.

### Evidences against blending theory

Under this concept, the progeny of a black and white animal would be uniformly grey. The further progeny from crossing the hybrids among themselves would be grey, for the black and white hereditary material, once blended, could never be separated again. Pattern of inheritance shown by atavism also speaks against blending theory. The traits of sex do not blend in unisexual organisms.

### Basic features of inheritance

- Traits have two alternative forms.
- Traits are represented in the individual by distinct particles which do not blend or change.

(iii) Traits may remain unexpected for one or more generations and reappear later unchanged.

(iv) Traits may remain together in one generation and separate in a later generation.

(v) One alternative of a trait may express more often than the other.

### Variations

Variations are differences found in morphological, physiological and cytological behaviouristic traits of individuals belonging to same species, race and family. They appear in offspring or siblings due to :

- ☐ Reshuffling of genes/chromosomes by chance separation of chromosomes
- ☐ Crossing over
- ☐ Chance combination of chromosomes during meiosis and fertilization.

### Types of variations

(1) **Somatic variations** : These variations influence the somatic or body cells. They appear after birth and are, also called acquired characters, modifications or acquired variations. Somatic variations are non-inheritable and usually disappear with the death of the individual. They are formed due to three reasons i.e., environmental factors, use and disuse of organs, and conscious efforts.

(2) **Germinal variations** : They are inheritable variations formed mostly in germinal cells which are either already present in the ancestors or develop a new due to mutations. Germinal variations are of two types :

(i) **Continuous variations** : They are fluctuating variations and also called recombinations because they are formed due to recombination of alleles as found in sexual reproduction. Darwin (1859) based his theory of evolution on continuous variations.

(ii) **Discontinuous variations** : They are mutations, which are ultimate source of organic variations. Discontinuous variations are caused by chromosomal aberrations, change in chromosome number and gene mutations. In pea seed coat colour changes grey to white is an example of spontaneous mutation.

### Importance of variations

- (1) Variations continue to pile up forming new species with time.
- (2) They are essential in the struggle for existence.
- (3) Adaptability is due to variations.
- (4) Variations allow breeders to improve races of plants and animals.
- (5) Discontinuous variations introduce new traits.
- (6) Inbreeding between closely related organisms reduces variation.

### Important terms used in inheritance studies

**Gene** : (Mendel called them factor) In modern sense an inherited factor that determines a biological character of an organism is called gene (functional unit of hereditary material).

**Allelomorphs or alleles (Bateson 1902)** : Alleles indicates alternative forms of the same gene. e.g., Tall TT and dwarf tt are alternation forms of the same gene etc.

**Gene locus** : The specific place on a chromosome where a gene is located.

**Wild and mutant alleles** : An original allele, dominant in expression and wide spread in the population is called wild allele. An allele formed by a mutation in the wild allele, recessive in expression and less common in the population is termed as mutant allele.

**Homozygous (Bateson and Saunders, 1902)** : Both the genes of a character are identical is said to be homozygous or genetically pure for that character. It gives rise to offspring having the same character on self-breeding e.g., TT (Homozygous dominant) or tt (Homozygous recessive).

**Heterozygous (Bateson and Saunders, 1902)** : Both the genes of a character are unlike is said to be heterozygous or hybrid. Such organisms do not breed true on self fertilization e.g., Tt.

If we know the number of heterozygous pairs we can predict the following :

Number of types of gametes =  $2^n$

Number of  $F_2$  phenotype =  $2^n$

(Where  $n$  is the number of heterozygous pairs).

Number of  $F_2$  genotype =  $3^n$

**Genotype (Johannsen 1909)** : The genotype is the genetic constitution of an organism. TT, Tt and tt are the genotypes of the organism with reference to these particular pairs of alleles.

**Phenotype** : External feature of organisms, colour and behaviour etc.

**Pure line** : Generations of homozygous individuals which produce offsprings of only one type i.e., they breed true for their phenotype and genotype.

**Monohybrid, dihybrid and polyhybrid** : When only one allelic pair is considered in cross breeding, it is called monohybrid cross. Similarly when two allelic pairs are used for crossing, it is called dihybrid cross and when more than two allelic pairs in a cross are used it is called polyhybrid cross.

**Reciprocal cross** : The reciprocal crosses involve two crosses concerning the same characteristics, but with reversed sexes.

**Genome** : Total set of genes (DNA instructions) in the haploid set of chromosomes and inherited as unit from parents to offspring is called genome.

**Gene pool** : All the genotypes of all organisms in a population form the gene pool.

**$F_1$  Generation** :  $F_1$  or first filial (filus-son, filia-daughter; Bateson, 1905) generation is the generation of hybrids produced from a cross between the genetically different individuals called parents.

**F<sub>2</sub> Generation (Bateson, 1905)** : F<sub>2</sub> or second filial generation is the generation of individuals which arises as a result of inbreeding or interbreeding amongst individuals of F<sub>1</sub> generation.

**Punnet square** : It is a checker-board used to show the result of a cross between two organisms, it was devised by geneticist, R.C. Punnet (1927). It depicts both genotypes and phenotypes of the progeny.

**Back cross** : It is a cross which is performed between hybrid and one of its parents. In plant breeding, back cross is performed a few times in order to increase the traits of that parent.

**Test cross** : It is a cross to know whether an individual is homozygous or heterozygous for dominant character. The individual is crossed with recessive parent. The ratio will be 50% dominant and 50% recessive in case of hybrid or heterozygous individual. In case of double heterozygote (e.g., RrYy) crossed with recessive (rryy) the ratio will be 1:1:1:1. Test cross helps to find out genotype of parents.

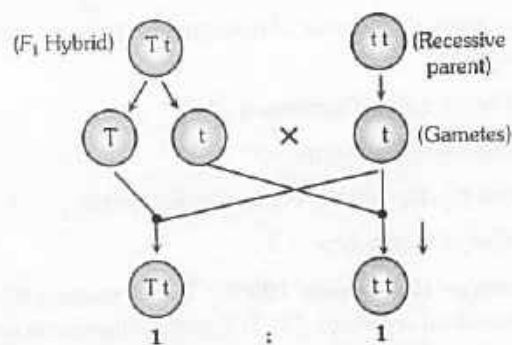


Fig : 7.1-1 Showing test cross

**Self cross/selfing** : It is the process of fertilization with pollen or male gametes of the same individual.

**Observed Vs expected results** : Experimental results confirm to the ones expected through the theory of probability if the size of the sample is small but they tend to approach the latter if the sample size is large.

**Hybrid** : The organism produced after crossing of two genetically different individuals is called hybrid.

**Heredity and variations in sexual and asexual reproduction**

**Sexual reproduction** : Variations are common in animals and plants which reproduce by sexual means. The reason for this is that the sexual reproduction is biparental, involves meiosis and fertilization, and the offspring receives some traits from father and some from mother.

**Asexual reproduction** : Those organisms which reproduce by asexual means e.g., bacteria, amoeba, euglena, rose etc. The asexual reproduction is monoparental, involves mitosis and the organism produced by it, inherits all the traits of its single parent. With the result, it is almost a carbon copy of the parent and is known as ramet. A group of ramets is called a clone.

## Mendelian period

Gregor Johann Mendel first "geneticist", also known as father of genetics was born on July 22, in 1822 in Silisian, a village in Heizendorf (Austria). In 1843, he joined Augustinian monastery at Brunn (then in Austria, now Brno Czechoslovakia). In 1856, Mendel got interested in breeding of Garden pea (*Pisum sativum*). He selected pure breeding varieties or pure lines of pea. Breeding experiments were performed between 1859 – 1864. The results were read out in two meetings of Natural History Society of Brunn in 1865 and published in 1866 in "Proceedings of Brunn Natural History Society" under the topic "Experiments in Plant Hybridization". Mendel died in 1884 without getting any recognition during his lifetime.

**Rediscovery of Mendel's work** : In 1900, Hugo de Vries of Holland, Carl Correns of Germany and Erich von Tshermak of Austria came to the same findings as were got by Mendel. Hugo de Vries found the paper of Mendel and got it reprinted in 'Flora' in 1901. Correns converted two of the generalisations of Mendel into two laws of heredity. These are law of segregation and law of independent assortment.

## Reasons for Mendel's success

**Method of working** : He maintained the statistical records of all the experiments and analysed them. He selected genetically pure (pure breed line) and purity was tested by self-crossing the progeny for several generations.

**Selection of material** : Mendel selected garden pea as his experimental material because it has the following advantages :

- (1) It was an annual plant.
- (2) Its short life-cycle made it possible to study several generations within a short period.
- (3) Has perfect bisexual flowers containing both male and female parts.
- (4) The flowers are predominantly self-pollinating because of self-fertilization, plants are homozygous.
- (5) It is easy to get pure lines for several generations.
- (6) It is easy to cross because pollens from one plant can be introduced to the stigma of another plant by removing anthers (emasculation) and bagging.
- (7) He studied seven pairs of characters which were present on four different pairs of chromosomes.

**Selection of traits** : Mendel selected seven pairs of contrasting characters as listed in the table. Luckily all were related as dominant and recessive.

Table : 7.1-2 Seven pairs of contrasting characters in pea plant

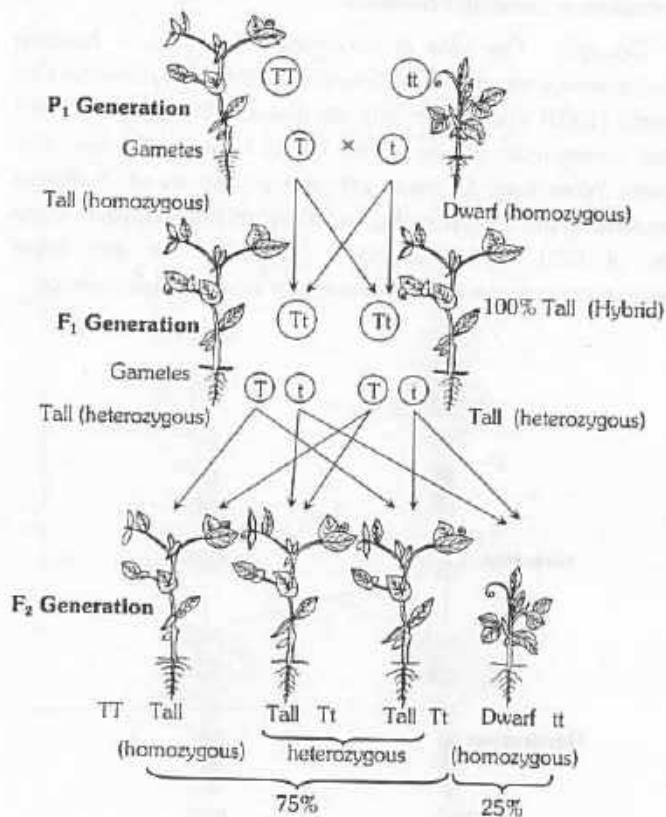
S. No.	Character	Dominant	Recessive
(1)	Stem length	Tall	Dwarf
(2)	Flower colour	Violet	White
(3)	Flower position	Axial	Terminal
(4)	Pod shape	Inflated	Constricted
(5)	Pod colour	Green	Yellow
(6)	Seed shape	Round	Wrinkled
(7)	Seed colour	Yellow	Green



**Mendel's experiments**

**Monohybrid cross :** Experiments with garden pea for single pair of contrasting characters.

Mendel crossed pure tall and dwarf plants. The plants belonged to  $F_1$  generation all tall hybrid were self-pollinated. The plants of  $F_2$  generation were both tall and dwarf, in approximate 3:1 ratio phenotypically and 1:2:1 genotypically.



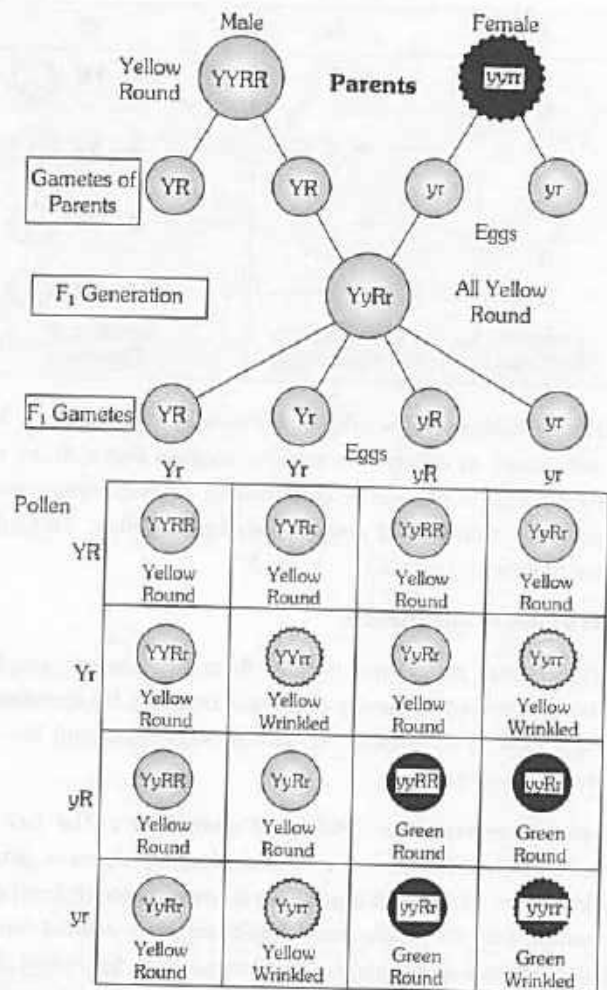
**Fig : 7.1-2 Mendel's monohybrid crosses between tall and dwarf pea plants**

**Mendel's explanation :** Mendel explained above results by presuming that tallness and dwarfness are determined by a pair of contrasting factors or determiners (now these are called genes). A plant is tall because it possesses determiners for tallness (represented by  $T$ ) and a plant is dwarf because it has determiners for dwarfness (represented by  $t$ ). These determiners occur in pairs and are received one from either parent. On the basis of this behaviour the tallness is described as dominant character and dwarfness as recessive (law of dominance). The determiners are never contaminated. When gametes are formed, these unit factors segregate so that each gamete gets only one of the two alternative factors. When  $F_1$  hybrids ( $Tt$ ) are self pollinated the two entities separate out and unite independently producing tall and dwarf plants (law of segregation). Monohybrid test cross ratio is 1:1.

**Dihybrid cross** (Crosses involving two pairs of contrasting traits).

Later on Mendel conducted experiments to study the segregation and transmission of two pairs of contrasting traits at a time. Mendel found that a cross between round yellow and wrinkled green seeds ( $P_1$ ) produced only round and yellow seeds in  $F_1$  generation, but in  $F_2$  four types of combinations were observed. These are :

- Round yellow 9 Parental combinations
- Round green 3 Non-parental combinations
- Wrinkled yellow 3 Non-parental combination
- Wrinkled green 1 Parental combination



**Fig : 7.1-3 Mendel's dihybrid cross between pea plants having yellow round seeds and green wrinkled seeds**

Thus the offsprings of  $F_2$  generation were produced in the ratio of 9 : 3 : 3 : 1 phenotypically and 1 : 2 : 2 : 4 : 1 : 2 : 1 : 2 : 1 genotypically. This ratio is called dihybrid ratio.

**Mendel's explanation :** Mendel explained the results by assuming that the round and yellow characters are dominant over wrinkled and green so that all the  $F_1$  offsprings are round yellow. In  $F_2$ -generation since all the four characters were assorted out independent of the others, he said that a pair of alternating or contrasting characters behave independently of the other pair i.e., seed colour is independent of seed coat.



Therefore, at the time of gamete formation genes for round or wrinkled character of seed coat assorted out independently of the yellow or green colour of the seed. As a result four types of gametes with two old and two new combinations i.e., YR, Yr yR, yr are formed from the  $F_1$  hybrid. These four types of gametes on random mating produce four types of offsprings in the ratio of 9:3:3:1 in  $F_2$  generation (Law of Independent Assortment). Dihybrid test cross ratio is 1 : 1 : 1 : 1.

**Table : 7.1-3 Forked-line method showing formation of four types of gametes from a  $F_1$  – dihybrid for seed colour and seed shape**

A	B	C
Y	R	YR (1)
Y	r	Yr (2)
y	R	yR (3)
y	r	yr (4)
Factor for Seed Colour	Factor for Seed Shape	Genotype of Gametes

**Trihybrid cross :** The offsprings shows 27 : 9 : 9 : 9 : 3 : 3 : 3 : 1 ratio found in trihybrid cross. This suggests that a di, tri, or polyhybrid cross is actually a combination of respectively two, three or more monohybrid crosses operating together. Trihybrid test cross ratio is 1 : 1 : 1 : 1 : 1 : 1 : 1 : 1.

### Mendel's laws of inheritance

Mendel's laws are still true because these take place in sexually reproducing organisms or parents are of pure breeding. He enunciated two major laws of inheritance i.e., law of segregation and law of independent assortment.

**Law of segregation (Purity of gametes) :** The law of segregation states that when a pair of contrasting factors or genes or allelomorphs are brought together in a heterozygote (hybrid) the two members of the allelic pair remain together without being contaminated and when gametes are formed from the hybrid, the two separate out from each other and only one enters each gamete as seen in monohybrid and dihybrid cross. That is why the law of segregation is also described as law of purity of gametes.

**Law of independent assortment :** If the inheritance of more than one pair of characters (two pairs or more) is studied simultaneously, the factors or genes for each pair of characters assort out independently of the other pairs. Mendel formulated this law from the results of a dihybrid cross.

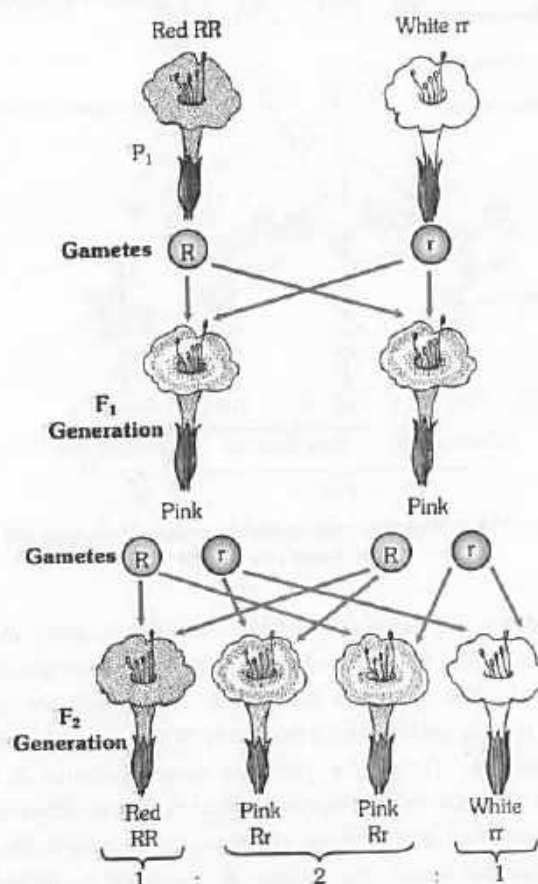
### Interaction of genes

Genes interaction is the influence of alleles and non-alleles on the normal phenotypic expression of genes. It is of two types :

(1) **Inter-allelic or intra-genic gene interaction :** In this case two alleles (located on the same gene locus on two homologous chromosomes) of gene interact in such a fashion to produce phenotypic expression e.g., co-dominance, multiple alleles.

(i) **Incomplete dominance or Blending inheritance (1: 2:1 ratio) :** After Mendel, several cases were recorded where  $F_1$  hybrids were not related to either of the parents but exhibited a blending of characters of two parents. This is called incomplete dominance or blending inheritance.

Example : First case of incomplete dominance or blending inheritance was reported in 4-O'clock plant, (*Mirabilis jalapa*) by Carl Correns (1903) when plants with red flowers (RR) are crossed with plants having white flowers (rr) the hybrid  $F_1$  plants (Rr) bear pink flowers. When these  $F_1$  plants with pink flowers are self pollinated they develop red (RR), pink (Rr) and white (rr) flowered plants in the ratio of 1:2:1 ( $F_2$  generation). Snapdragon or dog flower (*Antirrhinum majus*) is another example of incomplete dominance.



**Fig : 7.1-4 Incomplete dominance of flower colour in *Mirabilis jalapa***

(ii) **Codominance (1:2:1 ratio) :** In codominance, both the genes of an allelomorph pair express themselves equally in  $F_1$  hybrids. 1:2:1 ratio both genotypically as well as phenotypically in  $F_2$  generation.

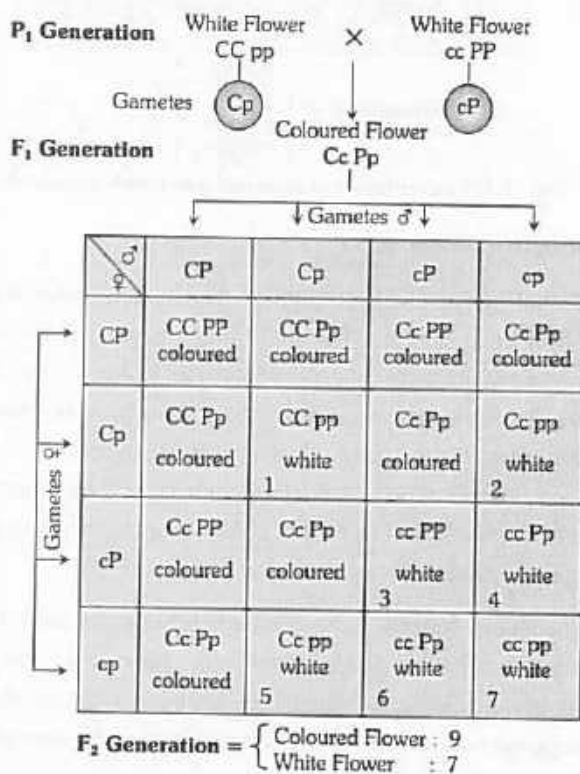
Example : Codominance of coat colour in cattle, Codominance in andalusian fowl and Codominance of blood alleles in man.

**Table : 7.1-4 Differences between incomplete dominance and codominance**

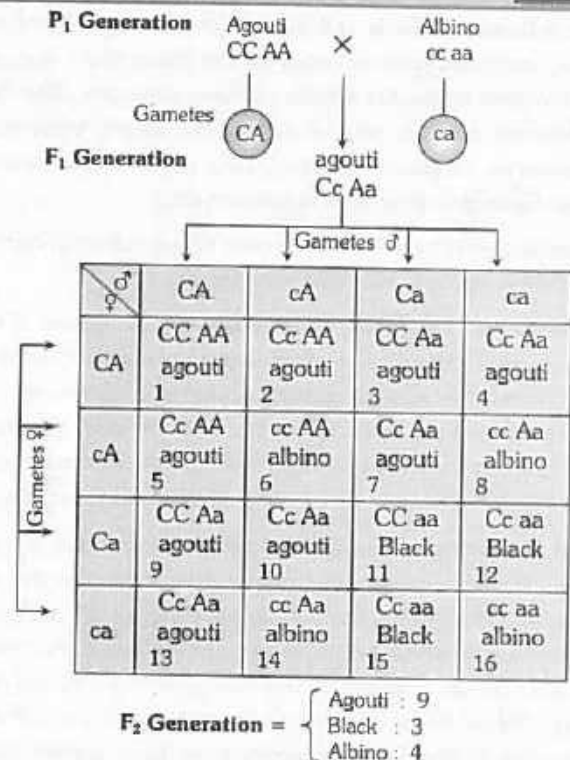
Incomplete dominance	Codominance
Effect of one of the two alleles is more conspicuous.	The effect of both the alleles is equally conspicuous.
It produces a fine mixture of the expression of two alleles.	There is no mixing of the effect of the two alleles.
The effect in hybrid is intermediate of the expression of the two alleles.	Both the alleles produce their effect independently, e.g., $I^A$ and $I^B$ , $Hb^S$ and $Hb^A$ .

(2) **Non-allelic or inter-genic gene interaction** : Here two or more independent genes present on same or different chromosomes, interact to produce a new expression e.g., epistasis, complementary genes, supplementary genes, duplicate genes, inhibitory genes, lethal genes etc.

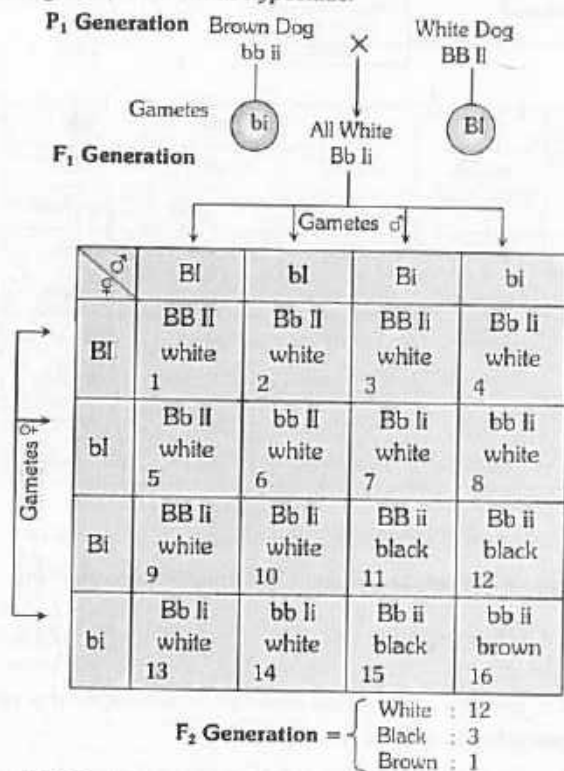
(i) **Complementary genes (9 : 7 ratio)** : The complementary genes are two pairs of nonallelic dominant genes (i.e., present on separate gene loci), which interact to produce only one phenotypic trait, but neither of them if present alone produces the phenotypic trait in the absence of other.

**Fig : 7.1-5** The results of an experiment to show the operation of complimentary genes in the production of flower colour in sweet pea (*Lathyrus*)

(ii) **Supplementary genes (9 : 3 : 4 ratio)** : Supplementary genes are two independent pairs of dominant genes which interact in such a way that one dominant gene will produce its effect whether the other is present or not. The second dominant when added changes the expression of the first one but only in the presence of first one. In rats and guinea pigs coat colour is governed by two dominant genes.

**Fig : 7.1-6** Interaction of supplementary genes in mice for coat colour

(iii) **Epistasis (Inhibiting genes)** : Epistasis is the interaction between nonallelic genes (Present on separate loci) in which one gene masks, inhibits or suppresses the expression of other gene. The gene that suppresses the other gene is known as inhibiting or epistatic factor and the one, which is prevented from exhibiting itself, is known as hypostatic.

**Fig : 7.1-7** Interaction of inhibiting genes in dog for coat colour showing dominant epistasis

**Dominant epistasis (12:3:1 or 13:3 ratio) :** In dominant epistasis out of two pairs of genes the dominant allele, (i.e., gene A) of one gene masks the activity of other allelic pair (Bb). Since the dominant epistatic gene A exerts its epistatic influence by suppressing the expression of gene B or b, it is known as dominant epistasis. Example – Dominant epistasis in dogs

Similar phenomena have been seen in fruit colour in cucurbita as summer squash and coat colour in chickens.

**Recessive epistasis (9:3:4 ratio) :** Epistasis due to recessive gene is known as recessive epistasis, i.e., out of the two pairs of genes, the recessive epistatic gene masks the activity of the dominant gene of the other gene locus. The dominant A expresses itself only when the epistatic locus C also has the dominant gene if the epistatic locus has recessive gene c, gene A fails to express.

(iv) **Duplicate genes (15:1 ratio) :** Sometimes two pairs of genes located on different chromosomes determine the same phenotype. These genes are said to be duplicate of each other. The dominant triangular fruit shape of *Capsella bursa pastoris* (shepherd's purse) is determined by two pairs of genes, say A and B. If any of these genes is present in dominant form, the fruit shape is triangular. In double recessive forms the fruits are top shaped and thus we get a 15 (triangular) : 1 (top shaped) ratio in  $F_2$  generation.

Example : Coat colour of mice.

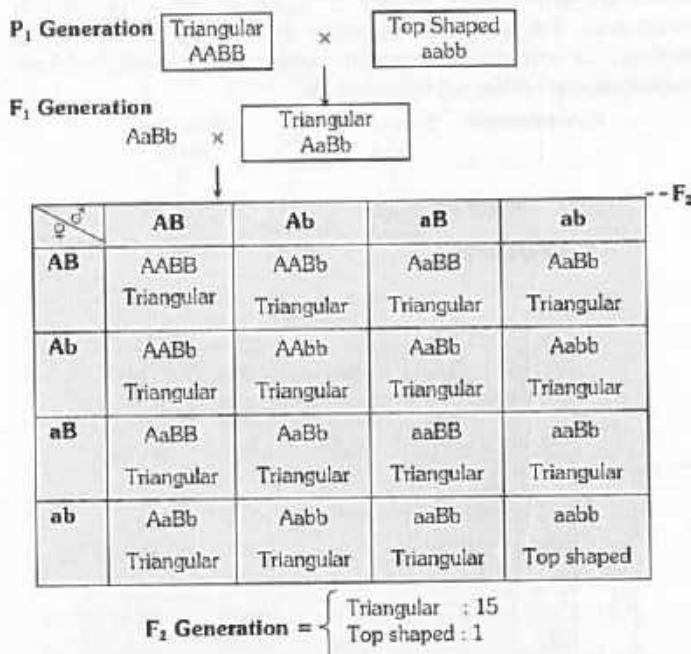


Fig : 7.1-8 Duplicate genes in *Capsella bursa pastoris*

(v) **Collaborator genes :** In collaboration two gene pairs, which are present on separate loci but influence the same trait, interact to produce some totally new trait or phenotype that neither of the genes by itself could produce.

Example : Inheritance of combs in poultry, where two genes control the development of comb.

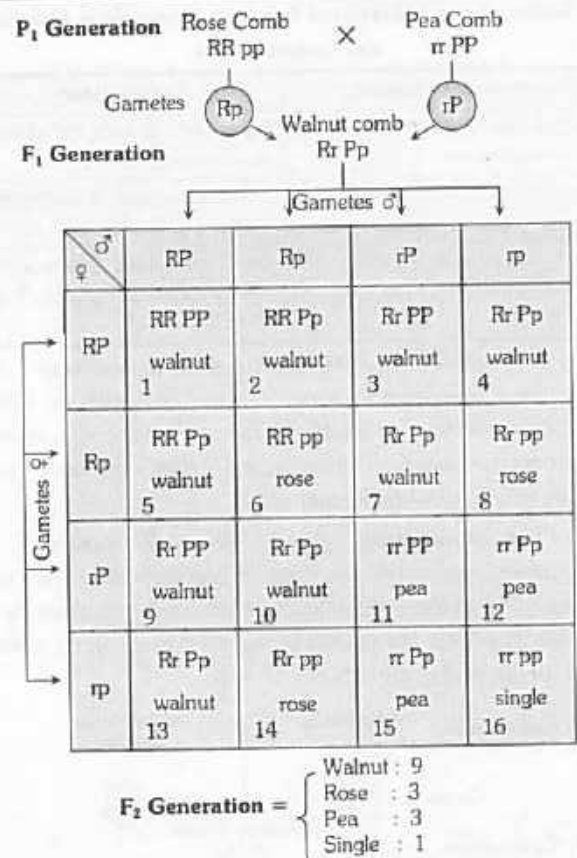


Fig : 7.1-9 Inheritance of rose and pea comb in poultry

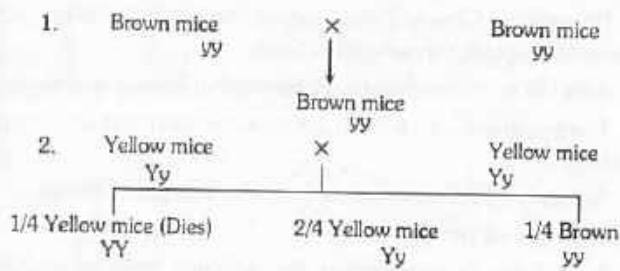
### Pleiotropic effect of genes

**Lethal genes :** Lethal factor were first of all reported in mice body by French geneticist 'Cuenot'. Certain genes are known to control the manifestation of some phenotypic trait as well as affect the viability of the organism. Some other genes have no effect on the appearance of the organism but affect the viability alone. These genes are known as lethals or semilethals depending upon their influence. Lethal factors in case of plants were reported first of all in snapdragons (*Antirrhinum majus*) by E. Baur (1907).

**Dominant lethals :** The dominant lethal genes are lethal in homozygous condition and produce some defective or abnormal phenotypes in heterozygous condition. Their most serious effect in heterozygous may also cause death. Following are the examples of dominant lethal genes.

Example – Yellow lethal in mice : A well known example of such lethals is from mice, given by Cuenot. He found that the yellow mice never breed true. Whenever the yellow mice were crossed with yellow mice, always yellow and brown were obtained in the ratio of 2:1. A cross between brown and brown mice always produced brown offsprings and a cross between brown and yellow produced yellow and brown in equal proportions. Yellow mice never present homozygous condition.



**A. Monohybrid Crosses****B. Test Cross**

In 1917, Stiegler concluded that yellow mice are heterozygous. The homozygous yellow ( $1/4^{\text{th}}$  of the total offsprings) dies in the embryonic condition. When these unborn ones are added to the 2:1 ratio of yellow and brown, these form typical 3:1 ratio. Cuenot suggested that gene  $Y$  has a multiple effect. It controls yellow body colour and has a dominant effect. It affects viability and acts as a recessive lethal. Other examples are inheritance of sickle cell anaemia in man, Brachyphalangy, Huntington's chorea in man.

**Recessive lethals :** The recessive lethals produce lethal effect only in homozygous condition. Their heterozygotes are normal. Therefore, recessive lethals remain unnoticed in the population but are established in the population because female are carrier for lethal gene. These are detected only when two heterozygous persons get married. Example : Tay Sach's lethal

**Qualitative Inheritance :** Qualitative inheritance or monogenic inheritance is that type of inheritance in which one dominant allele influences the complete trait, so that two such allele do not change the phenotype. Here dominant allele is monogene.

**Quantitative/Polygenic inheritance :** Quantitative inheritance or polygenic inheritance can be defined as, two or more different pairs of alleles which have cumulative effect and govern quantitative characters. The quantitative inheritance is due to incomplete dominance.

Polygenic/quantitative inheritance produces a number of phenotypes in  $F_2$  generation. 1 : 2 : 1 (3 phenotypes) in case of a pair of alleles, 1 : 4 : 6 : 4 : 1 (5 phenotypes) in case of two pairs.

Examples : Ear size in maize, White spotting in mice, Grain colour in wheat.

**Cytoplasmic / Extrachromosomal inheritance**

The fact that nucleus contains the units of inheritance was proposed by Oscar Hertwig in 1870. The mechanism was clearly understood with the development of Mendel's laws of inheritance. Further researchers proposed that cytoplasm also contains the hereditary material. The evidence for cytoplasmic inheritance was first presented by Correns in *Mirabilis Jalapa* and by Baur in *Pelargonium zonale* in 1908. The cytoplasm in such cases contain self perpetuating hereditary particles formed of DNA. These may be mitochondria, plastids or foreign organism, etc. The total self

duplicating hereditary material of cytoplasm is called **plasmon** and the cytoplasmic units of inheritance are described as plasmagenes.

**Criteria for cytoplasmic inheritance :** The cases of cytoplasmic inheritance are found to exhibit maternal influence. The reason is very simple. Very little cytoplasm is contained in the sperm cell of an animal. Most of the cytoplasm is contributed to the zygote by the ovum or egg. Hence if there are hereditary units in the cytoplasm, these will be transmitted to the offsprings through the egg. The offspring, therefore will exhibit maternal influence. This could be explained further by following example :

- (i) Maternal influence on shell coiling in snail.
- (ii) Inheritance of sigma particles in *Drosophila*.
- (iii) Breast tumour in mice.
- (iv) Plastid inheritance in *Mirabilis* (4 O' clock plant).
- (v) Plastid inheritance in *Oenothera*.
- (vi) Male sterility in plants – e.g. maize.
- (vii) Inheritance of kappa particles in *Paramecium*.
- (viii) Mitochondrial genetics – *Saccharomyces cerevisiae*, *Neurospora crassa*, *Aspergillus nidulens*.

**Linkage**

**Introduction :** "When genes are closely present they link together in a group and transmitted as a single unit this phenomenon is called linkage". It was reported in *Drosophilla* by T.H. Morgan in 1910.

**Theories of linkage**

**Sutton's hypothesis of linkage (1903) :** The number of groups of genes are equivalent to the number of chromosomes.

**Morgan's hypothesis of linkage (1910) :** It was given by T. H. Morgan. According to him the genes of homologous parents enter in the same gamete and tend to remain together, which is opposite in heterozygous parents. Linked group are located on the same chromosome and distance between linked group of gene limits the grade of linkage.

**Coupling and repulsion hypothesis :** Proposed by Bateson and Punnett (1906) states that dominant alleles tend to remain together as well with recessive alleles, called gametic coupling. If dominant and recessive alleles are present in different parents they tend to remain separate and called repulsion. When  $BBLL$  and  $bbll$  are crossed, the  $F_1$  is  $BbLl$  and the test cross of it will show progeny in 7 : 1 : 1 : 7 ratio i.e.,  $BbLl$  :  $Bbll$  :  $bbLl$  :  $bbll$  (coupling) when  $BbLl$  is crossed with  $bbll$  the  $F_1$  is  $BbLl$  or the test cross progeny will show 1 : 7 : 7 : 1 ratio i.e.,  $BbLl$  :  $Bbll$  :  $bbLl$  :  $bbll$  (repulsion). Coupled and repulsed genes are known as linked genes. Linkage has coupling phase and repulsion phase. In coupling phase both the linked genes have their dominant alleles in one chromosome and recessive alleles in other chromosomes. The heterozygotes with such constitution is called *cis* heterozygote. *Cis*-arrangement is an original arrangement which form two types of gametes as  $(AB)$  and  $(ab)$ . In Human X-chromosomes carry 102 genes and Y chromosome carries 10 genes only.

In repulsion phase the normal alleles as well as mutant alleles lie in opposite chromosomes of the homologous pair, such heterozygote is called as trans heterozygote. It is not original arrangement, caused due to crossing over, which form two types of gametes as (Ab) and (aB).

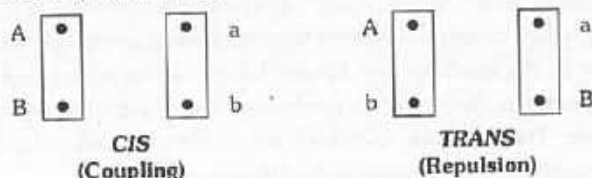


Fig : 7.1-10 CIS and TRANS-Arrangement of genes

**Chromosomal hypothesis of linkage :** It was given by Morgan and Castle. According to them linked genes are bound by chromosomal material and are transmitted as a whole.

### Types of linkage

Depending upon the absence or presence of nonparental or new combination of linked genes, linkage has been found to be complete or incomplete.

**Complete linkage (Morgan, 1919) :** Such cases in which linked genes are transmitted together to the offsprings only in their original or parental combination for two or more or several generations exhibit complete linkage. In such cases the linked genes do not separate to form the new or non-parental combinations. This phenomenon is very rare. Some characteristics in males of *Drosophila* are found to exhibit complete linkage.

**Incomplete linkage :** In majority of cases, the homologous chromosomes undergo breakage and reunion during gametogenesis. During reunion the broken pieces of the chromatids are exchanged, producing some nonparental or new combinations. Therefore, the linkage is rendered incomplete. The phenomenon of interchange of chromosome segments between two homologous chromosomes is called crossing over. Incomplete linkage is very common and has been studied in almost all the organisms. Hutchinson described incomplete linkage in maize seed.

### Linkage groups

All the genes which are linked with one another, form a linkage group. Since linked genes are present in the same chromosome, the number of linkage group in an animal or plant is equal to the haploid number of chromosomes present in its cells. e.g., in *Drosophila*  $n=4$ , hence linkage groups = 4. Similarly in *Pisum sativum*  $n = 7$ , hence linkage group = 7.

Number of linkage group in prokaryotes (bacteria, cyanobacteria or blue green algae and mycoplasma) is one. This hypothesis was given by Sutton and was proved by experiments on *Drosophila* by T.H. Morgan.

### Strength of linkage

The strength of linkage between any two pairs of linked genes of a chromosome depend upon the distance between them. Closely located genes show strong linkage, while genes widely located show weak linkages.

$$\text{Strength of linkage} \propto \frac{1}{\text{Distance between the gene}}$$

### Factor affected to linkage

**Distance :** Closely located genes show strong linkage while genes widely located show weak linkage.

**Age :** With increasing age the strength of linkage increases.

**Temperature :** Increasing temperature decreases the strength of linkage.

**X-rays :** X-rays treatment reduces the strength of linkage.

### Significance of linkage

- (i) It helps in maintaining the valuable traits of a newly developed variety.
- (ii) It helps locating genes on chromosome.
- (iii) It disallows the breeders to combine all the desirable traits in a single variety.

### Crossing over

The process by which exchange of chromosomal segment take place is called crossing over. Crossing over may be defined as "the recombination of linked genes" brought about as a result of interchange of corresponding parts between the chromatid of a homologous pair of chromosomes, so as to produce new combination of old genes. The term was given by Morgan and Castle. Janssen (1909) observed chiasmata during meiosis-I (Prophase). Morgan proposed that chiasmata lead to crossing over by breakage and reunion of homologous chromosomes. Crossing over results in new combination while non-cross over result in parental type, which leads to variations. Recon is the unit of recombination.

### Crossing over and chiasma

There are two views extended to explain the relationship between crossing over and chiasma formation. They are summarised here under :

**Chiasma type theory :** According to Janssen, 1909 the act of crossing over is followed by chiasma formation. He suggests that the crossing over takes place at the pachytene stage and the chiasma appear at diplotene.

**Classical theory :** According to Sharp, 1934, crossing over is the result of chiasma formation. According to this view, the chiasma are organised at pachytene and crossing over takes place at diplotene stage. On the basis of evidence available from molecular biology, that is untenable and hence rejected.

### Mechanism of crossing over

There are different views put forward to explain the mechanism of crossing over.

**Copy choice hypothesis :** According to Belling, 1928 the chromomeres represent the genes joined by interchromomeric regions. The chromomeres duplicate first and then the interchromomeric regions. The synthesis of these regions may occur in such a way that the chromomeres of the chromatid of a homologue get connected of the chromatid of the other homologue at a specific location. As a result, the adjacent chromatids of a pair of homologue are exchanged.

**Precocity hypothesis :** According to Darlington, the pairing of homologues occurs to avoid singleness of a chromosome. The pairing need of a chromosome could be nothing less than the replication of DNA. The crossing over takes place due to torsion on chromosome created by coiling of the two homologues around each other.

**Cross over value :** The percentage of crossing over varies in different materials. The frequency of crossing over is dependent upon the distance of two genes present on a chromatid.

**Coincidence :** Coincidence or coefficient of coincidence is inverse measure of interference and is expressed as the ratio between the actual number of double cross over and the expected number of such double cross. That is:

$$\text{Coincidence} = \frac{\text{Actual number of double cross over}}{\text{Expected number of double cross over}}$$

#### Factors controlling frequency of crossing over

Primarily, frequency of crossing over is dependent upon the distance between the linked genes, but a number of genetic, environmental and physiological factors also affect it. These are:

**Temperature :** High and low temperature increase the frequency of crossing over.

**X-ray :** Muller has discovered that exposure to X-ray and other radiations increases the frequency of crossing over.

**Age :** The frequency of crossing over decreases with increasing age in female *Drosophila*.

**Chemicals :** Certain chemicals which act as mutagens do affect the frequency of crossing over. Gene mutations may affect the frequency of crossing over. Some increase the frequency, whereas some may decrease it.

**Sex :** Crossing over in *Drosophila* males is negligible. Males of mammals also exhibit little crossing over. In silk-moth, crossing over does not occur in females.

**Chiasmata formation :** Chiasmata formation at one point discourages chiasmata formation and crossing over in the vicinity. This phenomenon is known as interference.

**Inversions :** Inversions of chromosome segments suppresses crossing over.

**Distance :** Distance between the linked genes is the major factor which controls the frequency of crossing over. The chances of crossing over between distantly placed genes are much more than between the genes located in close proximity.

Figure depicts that chance of crossing over between a and c are double as compared to the chances between a and b or b and c.

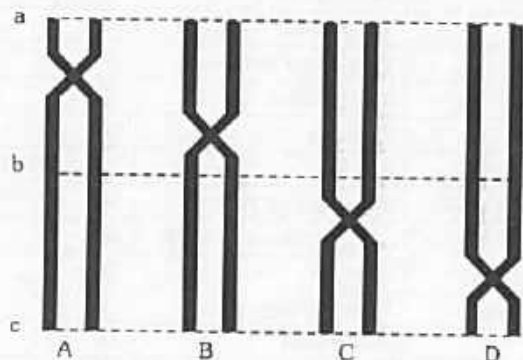


Fig : 7.1-11 Diagram showing possibilities of crossing over between genes at different distances

**Nutritional effect :** Crossing over frequencies are affected by concentration of metallic ions, such as calcium and magnesium.

**Genotypic effect :** Crossing over frequencies between the same two loci in different strains of the same species show variation because of numerous gene differences.

**Chromosome structure effect :** Changes in the order of genes on a chromosome produced by chromosomal aberrations usually act as cross over suppressors.

**Centromere effect :** Genes present close to the centromere region show reduced crossing over.

**Interference :** If there are two double crossovers, then one crossover tries to influence the other by suppressing it. This phenomenon is called as interference. Due to this phenomenon, the frequency of crossing over is always lower than the expected.

#### Significance of crossing over

This phenomenon is of great biological significance, which are as under:

(i) It gives evidence that the genes are linearly arranged on a chromosome. Thus, it throws light on the nature and working of the genes.

(ii) It provides an operational definition to a gene. It is deemed as the smallest heritable segment of a chromosome in the interior of which no crossing over takes place.

(iii) The crossing over is helpful in the chromosomal mapping. The percentage of crossing over is proportional to the distance between two genes.

(iv) It is the main cause of genetic variations. Its occurrence during the act of meiosis produces variations in the heritable characters of the gametes.

(v) This phenomenon has also found its utility in breeding and evolving new varieties. The linkage of undesirable characters can be broken by temperature treatment, using X-ray or chemicals. Thus, new recombinants can be prepared.

#### Chromosomal maps

A linkage or genetic chromosome map is a linear graphic representation of the sequence and relative distances of the various genes present in a chromosome. A chromosome map is also called a linkage map or genetic map.

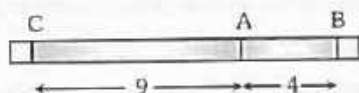
The percentage of crossing over between two genes is directly proportional to their distance. The unit of crossing over has been termed as by Haldane as centi Morgan (cM). One unit of map distance (cM) is therefore, equivalent to 1% crossing over. When chiasma is organised in between two gene loci, only 50% meiotic products shall be crossovers and 50% non-crossovers. Thus, the chiasma frequency is twice the frequency of cross over products i.e.,  $\text{chiasma \%} = 2 (\text{cross over \%})$  or  $\text{crossover \%} = 1/2 (\text{chiasma \%})$ .



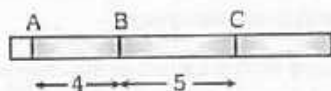
Accordingly, Sturtevant, 1911 prepared the first chromosomal map. Infact this map is a line representation of a chromosome where the location of genes has been plotted as points at specific distances. These distances are proportional to their crossing over percentage. Suppose there are three genes on a chromosome say, A B and C which could be arranged as A, B, C, A, C, B or B, A, C. Three point test cross confirms as to which gene is located in the centre. By determining the crossing over value between A and B, B and C as also between A and C, the linkage maps can be prepared. Broadly speaking, a chromosomal map can be prepared from the following results of crossing over between the genes A, B and C :

(i) 4% crossing over taking place between A and B. (ii) 9% crossing over taking place between A and C.

Hence the genes be located as above and there should be 13% crossing over between B and C and the genes may be arranged as under :



If there is 5% crossing over between B and C, the genes are arranged in the following manner and there should be 9% crossing over between A and C.



#### Uses of chromosomal map

- (i) Finding exact location of gene on chromosomes.
- (ii) Knowing recombination of various genes in a linkage group of chromosomes.
- (iii) Predicting result of dihybrid and trihybrid cross.

### 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.

#### 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.

#### Chromosomal theory of inheritance

It was proposed independently by Sutton and Boveri in 1902. The chromosome theory of inheritance proposes that chromosomes are vehicles of hereditary information and expression as Mendelian factors or genes.

#### Kinds of chromosomes

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

**Prokaryotic / 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.

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

#### Number of chromosomes

The number of chromosomes varies from two, the least number an organism can have, to a few hundred in different species. But chromosome number is fixed for a species. The least number of chromosomes are found in *Ascaris megalocephala* i.e., 2 ( $n = 2$  in *Mucor hiemalis* in plants) while in a radiolarian protist (*Aulocanitha*) has maximum number of chromosomes is 1600 (*Ophioglossum reticulatum*,  $2n = 1262$  in plants). The male of some roundworms and insects have one chromosome less than the females.

**Table : 7.1-5 Diploid number of chromosomes in some organisms**

Common name	Scientific name	Chromosomes
Amoeba	<i>Amoeba proteus</i>	500
Man	<i>Homo sapiens</i>	46
Gorilla	<i>Maccaca mulatta</i>	48
Pig	<i>Sas scrofa</i>	40
Sheep	<i>Ovis aries</i>	54
Cat	<i>Felis maniculata</i>	38
Dog	<i>Canis familiaris</i>	78
Rat	<i>Rattus rattus</i>	42
Rabbit	<i>Oryctolagus cuniculus</i>	44
Honey bee	<i>Apis mellifera</i>	32, 16
Mosquito	<i>Culex sp</i>	6
Grasshopper	<i>Gryllus</i>	23(M), 24(F)
Pink bread mould	<i>Neurospora crassa</i>	14
Baker's yeast	<i>Saccharomyces cerevisiae</i>	34
Broad bean	<i>Vicia faba</i>	12
Garden pea	<i>Pisum sativum</i>	14
Onion	<i>Allium cepa</i>	16
Maize	<i>Zea mays</i>	20
Potato	<i>Solanum tuberosum</i>	48

Cabbage	<i>Brassica oleracea</i>	18
Radish	<i>Raphanus sativum</i>	18
Compositae	<i>Haplopappus gracilis</i>	4
Adder's tongue fern	<i>Ophioglossum reticulatum</i>	1262
Jimson weed	<i>Datura stramonium</i>	24
Evening primrose	<i>Oenothera biennis</i>	14
Bread wheat	<i>Triticum aestivum</i>	42
Emmer wheat	<i>Triticum turgidum</i>	28
Tomato	<i>Lycopersicon esculentum</i>	24
Giant sequoia	<i>Sequoia sempervirens</i>	22

### Structure of chromosome

Different regions or structure recognized in chromosomes are as under

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

**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.

**Chromonemata** : They are coiled threads which form the bulk of chromosomes. A 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.

**Primary constriction** : A part of the chromosome is marked by a constriction. It is comparatively narrow than the remaining chromosome. It is known as primary constriction or centromere.

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

**Secondary constriction or nucleolar organizer** : Sometimes one or both the arms of a chromosome are marked by a constriction other than the primary 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).

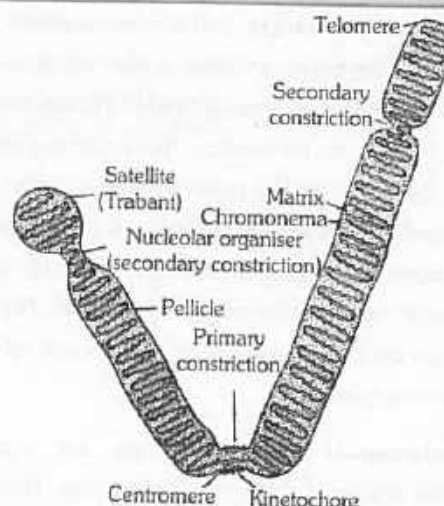


Fig : 7.1-12 Structure of chromosome

**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.

**Telomeres** : The tips of the chromosomes are rounded, 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.

**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.

### Molecular organisation of chromosome

Broadly speaking there are two types of models stating the relative position of DNA and proteins in the chromosomes.

(1) **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.

(2) **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.

(a) **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 (20Å). 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 is given by E.J. Dupraw (1965).

(b) **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 55Å height and 110Å 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 :

(i) **Nucleosomal histone** : These are small proteins responsible for coiling of 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.

(ii) **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.

(iii) **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.

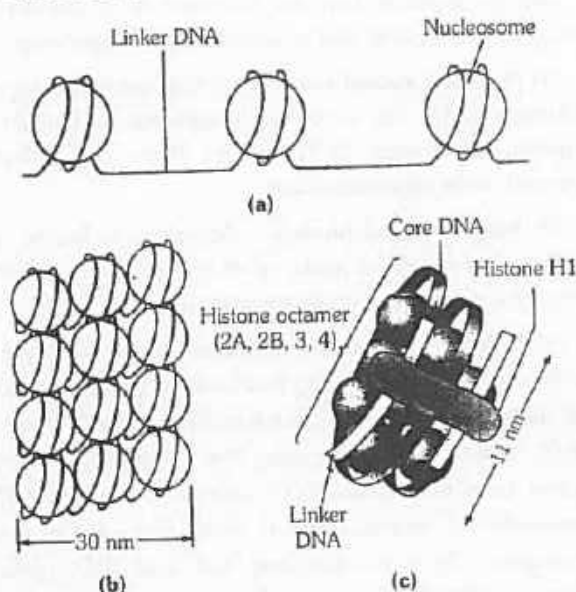


Fig : 7.1-13 Nucleosomes (a) 3 Nucleosomes (b) Nucleosomes coiled to form a solenoid (c) Basic structure of a nucleosome

(c) **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 300Å. The solenoid is further coiled to form a supersolenoid of 2000-4000Å 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 Finch and Klug 1976. Klug was awarded by nobel prize in 1982 for his work on chromosome.

(d) **Dangler-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.

### 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.

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. 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.

### 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  $\sim 10^7$ 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.



Table : 7.1-6 Differentiation of chromosomes by banding

Type of banding	Staining technique	Nature of bands
<b>Q (quinacrine) banding</b>	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 fluorescing Q bands which correspond to G-bands. DNA of Q/G bands contains more closely spaced SARs, giving tighter loops (Q loops).
<b>G (Giemsa) banding</b>	Chromosomes treated with alkaline solution and subjected to controlled trypsin digestion before staining with Giemsa, 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.
<b>R (reverse) banding</b>	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.
<b>T (telomeric) banding</b>	Prolonged heat treatment of chromosomes before staining with Giemsa or combination of dyes and fluorochromes.	T bands are a subset of R bands which are the most intensely staining. They are especially concentrated at the telomeres.
<b>C (centromere) banding</b>	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.

**Human karyotype and idiogram**

Tijo 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 23<sup>rd</sup> pair 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.

**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 microscope 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.

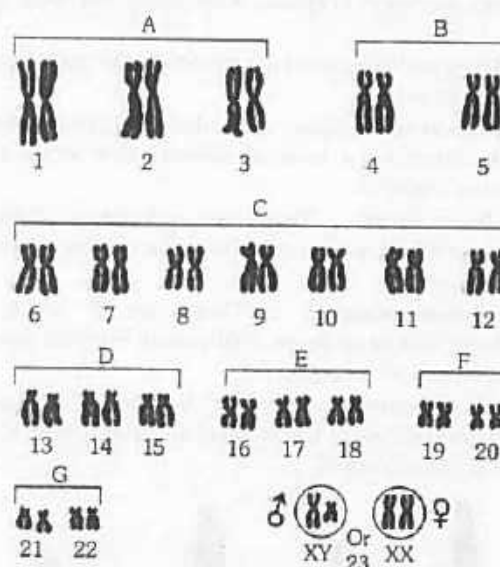


Fig : 7.1-14 Human karyotype

**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.

**Table : 7.1-7 Characteristics of the Chromosomes in 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

**Type of chromosomes**

(a) 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.

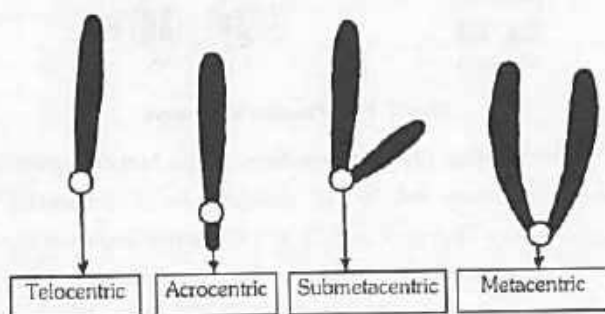
(b) Based on the location of centromere the chromosomes are categorised as follows :

(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.

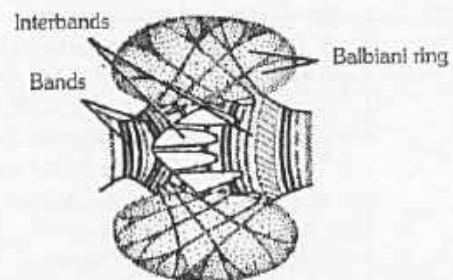
(4) **Metacentric** : These are V-shaped chromosomes in which centromere lies in the middle of chromosomes so that the two arms are almost equal.

**Fig : 7.1-15 Types of chromosomes****Special types of chromosomes**

**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 2000  $\mu\text{m}$ .

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 *Balbani rings*. Balbani rings produce a number of *m*-RNA, which may remain stored temporarily in the puffs. These also occur in Malpighian tubules, rectum, gut, foot pads, fat bodies, ovarian nurse cells etc.

**Fig : 7.1-16 Polytene chromosome showing balbiani ring**

**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. 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 5900  $\mu\text{m}$ . 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 5-100  $\mu\text{m}$ .

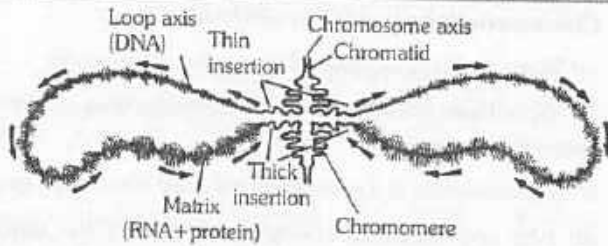


Fig : 7.1-17 A part of main axis with a pair of lateral loops of a lampbrush chromosome showing synthesis of RNA

**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.
- (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 synapse with them.
- (6) They are found more commonly in plants than in animals.

**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. Those of males have 9 chromosomes. 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.

**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.

**S and E-chromosomes :** S and E-chromosomes have been reported in insects in the family Cecidomyiidae (gall insects) and family Chironomidae (Diptera).

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 of gall insect 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.

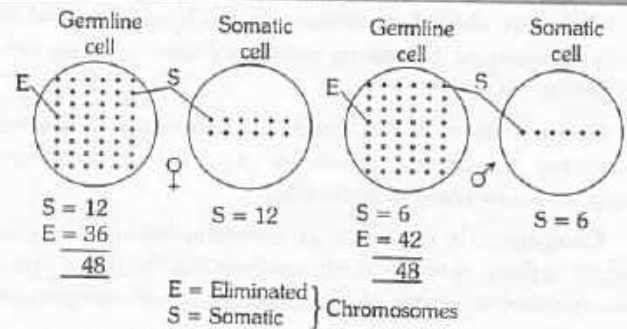


Fig : 7.1-18 Schematic representation of the S and E chromosomes of the gall insect *Miastor*

## Genes

Term 'gene' was given by Johannsen (1909) for any particle to which properties of Mendelian factor or determiner can be given. Thomas Hunt Morgan (1910) defined gene as 'any particle on the chromosome which can be separated from other particles by mutation or recombination is called a gene'. In general, gene is the basic unit of inheritance.

According to the recent information a gene is a segment of DNA which contains the information for one enzyme or one polypeptide chain coded in the language of nitrogenous bases or the nucleotides. The sequence of nucleotides in a DNA molecule representing one gene determines the sequence of amino acids in the polypeptide chain (the genetic code). The sequence of three nucleotides reads for one amino acid (codon). Khorana was awarded nobel prize for the synthesis of an artificial gene.

### Gene action

Gene act by producing enzymes. Each gene in an organism produces a specific enzyme, which controls a specific metabolic activity. It means each gene synthesizes a particular protein which acts as enzyme and brings about an appropriate change.

**One gene one enzyme theory :** This theory was given by Beadle and Tatum (1958), while they were working on red mould or *Neurospora* (ascomycetes fungus). Which is also called *Drosophila* of plant kingdom. Wild type *Neurospora* grows in a minimal medium (containing sucrose, some mineral salts and biotin). The asexual spores i.e. conidia were irradiated with x-rays or UV-rays (mutagenic agent) and these were crossed with wild type. After crossing sexual fruiting body is produced having asci and ascospores. The ascospores produced are of 2 types -

- (i) The ascospores, which are able to grow on minimal medium called 'prototrophs'.
- (ii) Which do not grow on minimal medium but grow on supplemented medium called 'auxotrophs'.

### Molecular structure of gene

Gene is chemically DNA but the length of DNA which constitutes a gene, is controversial 3 term i.e. cistron, muton and recon were given by Seymour Benzer to explain the relation between DNA length and gene.

**Cistron or functional gene or gene in real sense :** Benzer (1955) related gene to arm cistron or Cistron is that particular length of DNA which is capable of producing a protein molecule or polypeptide chain or enzyme molecule.



**Muton or unit of mutation :** Muton is that length of DNA which is capable of undergoing mutation. Muton is having one or two pairs of nucleotide.

**Recon :** Recon is that length of DNA which is capable of undergoing crossing over or capable of recombination. Recon is having one or two pairs of nucleotides.

**Complon :** It is the unit of complementation. It has been used to replace cistron. Certain enzymes are formed of two or more polypeptide chains whose active groups are complimentary to each other.

**Operon :** Operon is the combination of operator gene and sequence of structure genes which act together as a unit. Therefore it is composed of several genes. The effect of operator gene may be additive or suppressive.

**Replicon :** It is the unit of replication. Several replicons constitute a chromosome.

#### Some specific terms

**Transposons or Jumping genes :** The term 'transposon' was first given by Hedges and Jacob (1974) for those DNA segments which can join with other DNA segments completely unrelated and thus causing illegitimate pairing. These DNA segments are transposable and may be present on different place on main DNA. The transposons are thus also called Jumping genes. Hedges and Jacob reported them in bacteria. But actual discovery of these was made by Barbara Mc Clintock (1940) in maize and she named them as controlling elements or mobile genetic elements. For this work, she was awarded nobel prize in (1983).

**Retroposons :** The term was given by Rogers (1983) for DNA segments which are formed from RNA or which are formed by reverse transcription under the influence of reverse transcriptase enzyme or RNA dependent DNA polymerase enzyme. About 10% of DNA of genome in primates and rodents is of this type.

RNA  $\xrightarrow{\text{Reverse transcriptase}}$  DNA (Retroposon)

**Split genes or interrupted genes :** Certain genes were reported first in mammalian virus and then in eukaryotes by R. Roberts and P. Sharp in (1977) which break up into pieces or which are made of segments called exons and introns. These are called split genes or interrupted genes.

Split gene = Exons + Introns

If mRNA formed from split gene exons are present and not corresponding to introns. So in split genes, exons carry genetic information or informational pieces of split genes are exons.

**Pseudogenes or false genes :** DNA sequences present in multicellular organisms, which are useless to the organism and are considered to be defective copies of functional genes (cistrons) are called pseudogenes or false genes. These have been reported in *Drosophila*, mouse and human beings.

#### Multiple allelism

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.

#### Characteristics of multiple allelism

- (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.

**Examples of multiple allelism :** A well known example of a trait determined by multiple alleles is the blood groups in man and skin colour. Other example are eye colour in *Drosophila*, colour of wheat kernel, corolla length in *Nicotiana*, Coat colour in Cattle etc.

#### Blood groups in man

**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.

**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 the 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.

#### Types of blood groups

**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.

**Table : 7.1-8 Blood groups of man with antigen and antibodies**

Type of blood group	Antigen	Antibody	% in society
A	A	Anti-B or 'b'	23.5
B	B	Anti-A or 'a'	34.5
AB	A, B	Absent	7.5
O	None	'a' and 'b'	34.5

**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.

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.

#### 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<sup>-</sup> are universal donors as they lack both the antigens and Rh<sup>-</sup> person can donate to Rh<sup>+</sup> person as well as Rh<sup>-</sup> person but Rh<sup>+</sup> person cannot donate blood to Rh<sup>-</sup> 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.

#### 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.

#### 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<sup>a</sup>, gene for antigen B by I<sup>b</sup> and the gene for the absence of both antigens by I<sup>o</sup>. It is customary to use the letter I (Isohaemagglutininogen) as a basic symbol for the gene at a locus. Based on this, six genotypes are possible for four blood groups in human population.

**Table : 7.1-9 Genotype of blood groups in man**

Type of blood group	Genotype	Nature of gene
A	I <sup>a</sup> I <sup>a</sup>	Homozygous (Dominant)
A	I <sup>a</sup> I <sup>o</sup>	Heterozygous
B	I <sup>b</sup> I <sup>b</sup>	Homozygous (Dominant)
B	I <sup>b</sup> I <sup>o</sup>	Heterozygous
AB	I <sup>a</sup> I <sup>b</sup>	Codominant
O	I <sup>o</sup> I <sup>o</sup>	Homozygous (Recessive)

The alleles I<sup>a</sup> and I<sup>b</sup> 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<sup>a</sup> and I<sup>b</sup> are codominant and are dominant over the allele I<sup>o</sup> (I<sup>a</sup> = I<sup>b</sup> > I<sup>o</sup>). 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.

**Table : 7.1-10 Cross between parents heterozygous for blood group A and B**

		Male (Heterozygous for blood group A)	
		Gametes	
Female (Heterozygous for blood group B)	I <sup>a</sup> I <sup>b</sup>	I <sup>a</sup>	I <sup>b</sup>
	I <sup>a</sup> I <sup>b</sup>	I <sup>a</sup> I <sup>a</sup>	I <sup>a</sup> I <sup>b</sup>
	I <sup>a</sup> I <sup>b</sup>	Group AB	Group B
	I <sup>a</sup> I <sup>b</sup>	I <sup>a</sup> I <sup>o</sup>	I <sup>b</sup> I <sup>o</sup>
		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.

**Table : 7.1-11 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
O and O	I <sup>o</sup> I <sup>o</sup> × I <sup>o</sup> I <sup>o</sup>	O	A, B, AB
O and A	I <sup>o</sup> I <sup>o</sup> × I <sup>a</sup> I <sup>a</sup>	O, A	B, AB
A and A	I <sup>a</sup> I <sup>a</sup> × I <sup>a</sup> I <sup>a</sup>	O, A	B, AB
O and B	I <sup>o</sup> I <sup>o</sup> × I <sup>b</sup> I <sup>b</sup>	O, B	A, AB
B and B	I <sup>b</sup> I <sup>b</sup> × I <sup>b</sup> I <sup>b</sup>	O, B	A, AB
A and B	I <sup>a</sup> I <sup>a</sup> × I <sup>b</sup> I <sup>b</sup> I <sup>a</sup> I <sup>a</sup> × I <sup>b</sup> I <sup>o</sup> I <sup>a</sup> I <sup>o</sup> × I <sup>b</sup> I <sup>b</sup>	O, A, B, AB	None
O and AB	I <sup>o</sup> I <sup>o</sup> × I <sup>a</sup> I <sup>b</sup>	A, B	O, AB
A and AB	I <sup>a</sup> I <sup>a</sup> × I <sup>a</sup> I <sup>b</sup>	A, B, AB	O
B and AB	I <sup>b</sup> I <sup>b</sup> × I <sup>a</sup> I <sup>b</sup>	A, B, AB	O
AB and AB	I <sup>a</sup> I <sup>b</sup> × I <sup>a</sup> I <sup>b</sup>	A, B, AB	O

**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.

**Table : 7.1-12 Possibilities of blood groups of other parent on the basis of blood group of child and one parent being known**

Blood group of child (known)	Genotype of child (known)	Blood group of father or mother (known)	Blood group of other parent	
			Possible	Not possible
O	$I^o I^o$	O	A, B O, B O, A	AB
A	$I^a I^a, I^a I^o$	O, B	A, AB	O, B
B	$I^b I^b, I^b I^o$	O, A	B, AB	O, A
AB	$I^a I^b$	B	A, AB	O, B
		AB	A, B, AB	O

#### Rhesus or 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 recessive 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  $Rh^+$ .

**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 recipient develops Rh antibodies in its blood plasma. If  $Rh^+$  blood is transfused for the second time it causes agglutination and leads to the death of  $Rh^-$  person.

**Erythroblastosis foetalis :** This disease is related to the birth of a child with Rh factor. It causes the death of the foetus within the womb or just after birth. It was studied by Levine together with Landsteiner and Wiener.

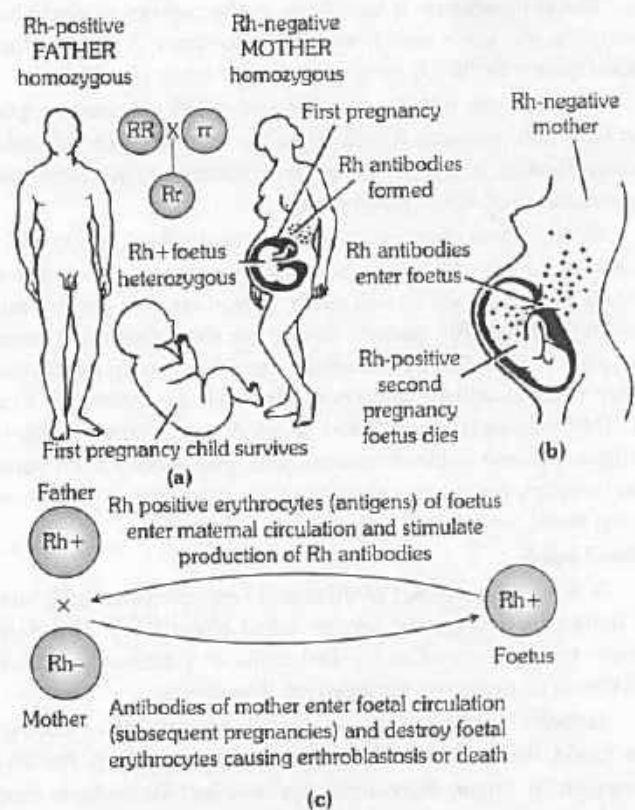
The father of Rh affected foetus is  $Rh^+$  and the mother is  $Rh^-$ . The child inherits the  $Rh^+$  trait from the father. A few  $Rh^+$  red blood corpuscles of foetus in the womb enter in the blood of the mother where they develop Rh antibodies. As mother's blood is  $Rh^-$  i.e. devoid of Rh antigen, it causes no harm to her. These Rh antibodies along with the mother's blood on reaching the foetal circulation cause clumping of foetal RBCs or agglutination reaction. The first child is somehow 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.

**Table : 7.1-13 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 : 7.1-19 Foetal death in the womb due to erythroblastosis foetalis**

**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.



**Inheritance of Rh factor :** Rh factor or Rh antigen is determined by a series of four pair of multiple alleles. They are denoted as  $R^1$ ,  $R^2$ ,  $R^0$ ,  $R^+$ ,  $r^1$ ,  $r^2$ ,  $r^+$  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<sup>-</sup> factor is inherited according to Mendelian principle.

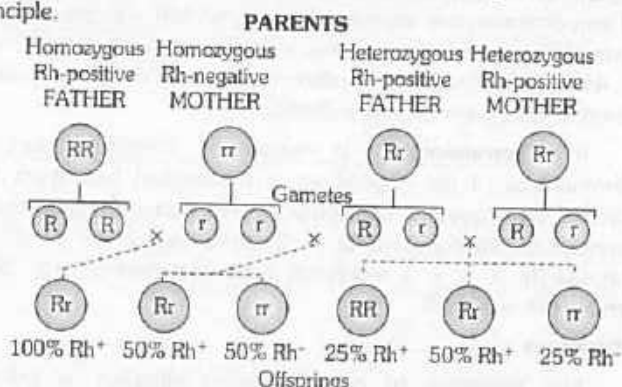


Fig : 7.1-20 Inheritance of Rh antigen

## Genetic Mutation

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 :

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

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

### Gene mutations

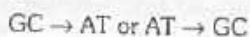
Gene or point mutations are stable changes in genes i.e. DNA chain. Many times a change in a gene or nucleotide pair does not produce detectable mutation. Thus the point or gene mutation mean the process by which new alleles of a gene are produced. The gene mutation are of following types :

**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 protons in the molecules.

**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 mRNA 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.



(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.

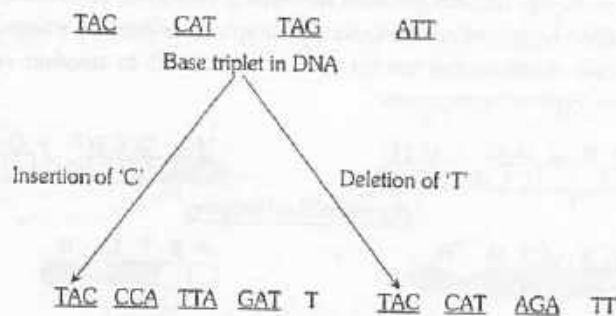


Fig : 7.1-21 Frame shift mutations

## 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.

### (1) Morphological aberrations of chromosomes

**Deletion or deficiency :** Sometimes a segment of chromosome break off and get lost. If a terminal segment of a chromosome is lost, it is called deficiency. Deficiency generally proves lethal or semilethal. If intercalary segment is lost it is termed deletion.



Deletion occurs during pairing in meiosis. For example in human babies deletion of a segment of chromosome number 5 causes a disease called *cri-du-chat* syndrome (the baby cries like a cat and is mentally retarded with small head).

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.

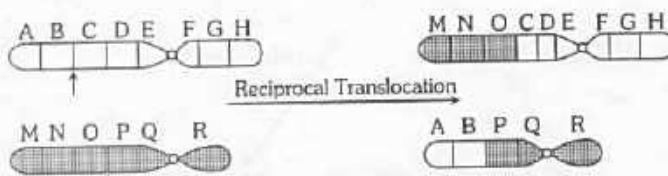
**Duplication :** In this mutation deleted chromosomal segment is attached to its normal homologous chromosome. Here a gene or many genes are repeated twice or more times in the same chromosome.



**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 the centre portion turned through  $180^\circ$ , the resulting gene order is A, D, C, B, E, F, G, H.



**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.



## (2) Numerical aberrations of chromosomes

**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. It can be divided into following types :

(i) **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  $4n$  or  $6n$  chromosomes the haploids will possess  $2n$  or  $3n$  chromosome whereas its monoploid will possess only one set ( $n$ ) of chromosome.

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

(a) **Autopolyploidy** : It is a type of polyploidy in which there is a numerical increase of the same genome, e.g., Autotriploid (AAA), autotetraploid (AAAA). e.g., Maize, Rice, Gram. Autopolyploidy induces *gigas* effect.

(b) **Allopolyploidy** : It has developed through hybridisation between two species followed by doubling of chromosomes (e.g., AABB). Allopolyploids function as new species. e.g., Wheat, American cotton, *Nicotiana glauca*. Two recently produced allopolyploids are *Raphanobrassica* and *Triticale*.

(c) **Autoallopolyploidy** : It is a type of allopolyploidy in which one genome is in more than diploid state. commonly autoallopolyploids are hexaploids (AAAABB), e.g., *Helianthus tuberosus*.

**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.

(i) **Monosomy** : Diploid organism that are missing one chromosome of a single pair with genomic formula  $2n - 1$ . Monosomics can form two kind of gametes, ( $n$ ) and ( $n - 1$ ). e.g., *Turner's syndrome* ( $44 + X$ ).

(ii) **Nullisomy** : An organism that has lost a chromosome pair is nullisomic. The result is usually lethal to diploids ( $2n - 2$ ).

(iii) **Trisomy** : Diploids which have extra chromosome represented by the chromosomal formula  $2n + 1$ . One of the pairs of chromosomes has an extra member, so that a trivalent may be formed during meiotic prophase. e.g., Down's syndrome ( $45 + XX$  or  $45 + XY$ ), Klinefelter's syndrome ( $44 + XXY$ ). All the possible trisomic have been studied in *Datura*.

(iv) **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  $2n + 2$  rather than  $2n + 1 + 1$ . The formula  $2n + 1 + 1$  represents a double trisomic. e.g., Super female ( $44 + XXXX$ ).

## Mutagens

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

(1) **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.

(i) **Ionizing (Particulate)** :  $\alpha$ -particles,  $\beta$ -rays, protons and neutrons.

(ii) **Ionizing (non particulate)** : X-rays,  $\gamma$ -rays and cosmic rays.

(iii) **Nonionizing** : Ultraviolet rays

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

(i) **Base analogues** : 5-bromodeoxyuridine (BrdU), 2-amino purine.

(ii) **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).

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

## 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.

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



**Huntington disease** : Huntington disease is caused by a dominant gene on chromosome 4. 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. The death comes ten to fifteen years after the onset of symptoms.

**Neurofibromatosis** : Also called "von Recklinghausen disease" caused by a dominant gene on chromosome 17. The affected individual may have ten spots on the skin which later may increase in size and number. Small benign tumours called neurofibromas may occur under the skin or in various organs.

**Tay-Sachs disease** : 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. 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.

**Cystic fibrosis** : The most common lethal genetic disease due to a recessive mutation on the chromosome 7. The body produces abnormal glycoprotein which interferes with salt metabolism. The mucus secreted by body becomes abnormally viscid and blocks passages in the lungs, liver and pancreas.

**Alzheimer's disease** : 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. Alzheimer's disease, a form of dementia, occurs in karyotypically normal individuals. The brain of Alzheimer's patients show a marked loss of neurons. 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  $\beta$  protein. The occurrence of Alzheimer's disease in people with Down's syndrome suggests that a gene or genes on chromosome 21 is involved. According to Bush (2003) Alzheimer's disease is caused by a copper and zinc build up in the brain.

**Marfan's syndrome** : Marfan's syndrome is due to dominant mutation resulting in the production of abnormal form of connective tissues and characteristic extreme looseness of joints. The long bones of body grow longer, fingers are very long called 'spider fingers' or arachnodactyly. The lenses in eyes become displaced.

**Albinism** : Albinism is an autosomal recessive mutation. An albino cannot synthesize melanin which provides black colouration to skin and hair. Albinism is due to tyrosinase deficiency. The enzyme tyrosinase normally converts the amino acid tyrosine to melanin through an intermediate product DOPA (dihydro phenyl alanine).

**Sickle-cell disease** : 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  $\beta$  chain of adult haemoglobin. The mutated gene  $Hb^S$  produces sickle-cell haemoglobin. The sixth amino acid in  $\beta$  chain of normal haemoglobin is glutamic acid. In sickle-cell haemoglobin this amino acid is replaced by valine. The children homozygous ( $Hb^S Hb^S$ ) produce rigid chains. When oxygen level of the blood drops below certain level, RBCs undergo sickling. Such cells do not transport oxygen efficiently; they are removed by spleen causing severe

anaemia. Individuals with the  $Hb^A Hb^A$  genotype are normal, those with the  $Hb^S Hb^S$  genotype have sickle-cell disease, and those with the  $Hb^A Hb^S$  genotypes have the sickle-cell trait. Two individuals with sickle-cell trait can produce children with all three phenotypes. Individuals of sickle-cell trait are immune to malaria.

**Thalassemia** : 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. Heterozygous persons show a milder disease, thalassemia minor or also called Cooley's anaemia. The persons suffering from thalassemia major are unable to produce  $\beta$  chain. Their haemoglobin contains  $\delta$  chains like that of foetus which is unable to carry out normal oxygen transporting function.

**Alkaptonuria** : Alkaptonuria was the first of the recessive human trait discovered in 1902 by Archibald Garrod, 'father of physiological genetics' or 'father of biochemical genetics'. Patients of alkaptonuria excrete large amounts of homogentistic acid in urine. Such urine turns black upon exposure to light. In normal person, homogentistic acid (alkapton) is oxidized by a liver enzyme homogentistic acid oxidase to maleyl acetoacetic acid.

**Phenylketonuria (PKU)** : Phenylketonuria was discovered by the Norwegian physician A. Folling in 1934; an autosomal recessive mutation of gene on chromosome 12. PKU results when there is a deficiency of liver enzyme phenylalanine hydroxylase that converts phenylalanine into tyrosine. There is a high level phenylalanine in their blood and tissue fluids. Increased phenylalanine in the blood interferes with brain development; muscles and cartilages of the legs may be defective and the patients cannot walk properly.

**Gaucher's disease** : 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. Non processing of lipids results in accumulation of fatty material in spleen, liver, bone marrow and brain. The swelling of these organs occurs and patients usually die by the age of 15 years.

**Galactosemia** : Galactosemia is inherited as an autosomal recessive, and the affected person is unable to convert galactose to glucose. Galactosemia is due to the deficiency of the enzyme Galactose Phosphate uridyl Transferase (GPT). Milk is toxic to galactosemic infants; child usually dies at three years of age.

**Taste blindness of PTC** : Taste blindness of PTC is a genetic trait, not a disease, discovered by Fox in 1932. PTC (phenyl thiocarbamide) is a compound of nitrogen, carbon and sulphur with sour taste. About 30% people lack the ability to taste PTC which is transmitted by a dominant gene T. The genotypes TT and Tt are tasters of PTC, while tt are non-tasters or taste blind persons.

**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'. 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.



**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.

#### Sex chromosome abnormalities

**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 oestrogen. About one out of every 5,000 female births results in Turner's syndrome.

**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.

**Super females or 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.

**Criminal's or Jacob'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.

#### Autosomal abnormalities

**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 21st 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  $2n + 1$  ( $21^{\text{st}}$ ) = 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.

**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.

**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.

#### 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 :

##### 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.

**XX-XY type or Lygaeus type :** This type of sex-determining mechanism was first studied in the milk weed bug, *Lygaeus turticus* by Wilson and Stevens. Therefore, it is called Lygaeus type. It is most common in plants and animals. e.g., In all mammals including man and among plants in *Melandrium album*, *M. rubrum*, *Elodea*, *Rumex angiocarpus*, *Populus*, *Salix*, *Smilax*, *Morus*, *Canabis* etc. These are two different patterns of sex determination in Lygaeus type.

(1) Female homogametic XX and male heterogametic XY e.g., *Drosophila*.

(2) Female heterogametic and male homogametic e.g., Fowl, Birds and some fishes.

**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. e.g., Grasshopper and plant kingdom in *Dioscorea sinata* and *Vallisneria spiralis*.

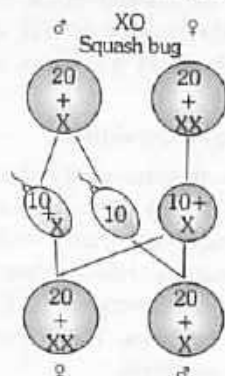


Fig : 7.1-22 Protector type of sex determination in Grasshopper

#### 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.

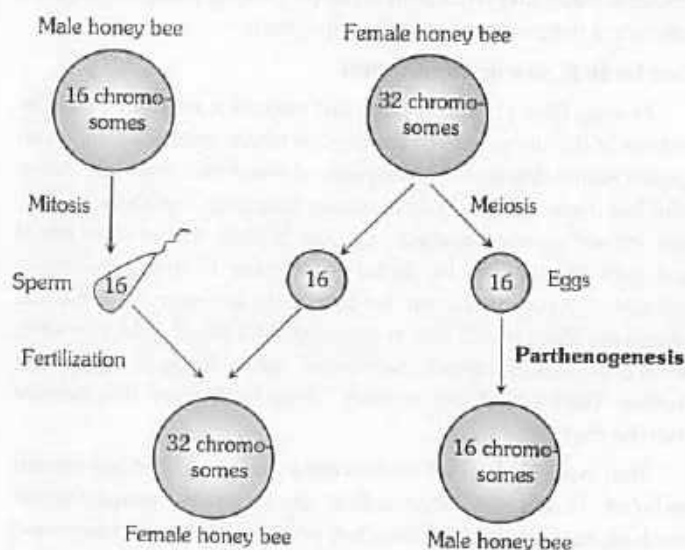


Fig : 7.1-23 Haploid-diploid mechanism of sex determination in honeybee

Table : 7.1-14 Different types of chromosomal mechanisms of sex-determination in animals

Organisms	Heterogametic sex	Gamete		Zygotes	
		Sperms	Eggs	F	M
<i>Drosophila</i> , man etc.	Male	X and Y	All X	XX	XY
Protector (Bug Grasshopper)	Male	X and O	XX	XX	XO
Birds, moths	Female	All X	X and Y	XY	XX
Fumea (a moth)	Female	All X	X and O	X	XX

#### 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.

**Intersexes in *Drosophila* and ratio theory of sex determination :** 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

**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 – Diploid females
- (3) AAXXY – Diploid females
- (4) AAAXX – Intersexes
- (5) AAAXXY – Intersexes
- (6) AAXY – Normal males
- (7) AAXXX – Super females
- (8) AAAXY – Super males

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

**Gynandromorphs in *Drosophila* 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. The occurrence of gynandromorphs clearly indicates that the number of X-chromosomes determines the sex of the individual. The term Gynandromorphism was introduced by Goldschmidt in 1915.

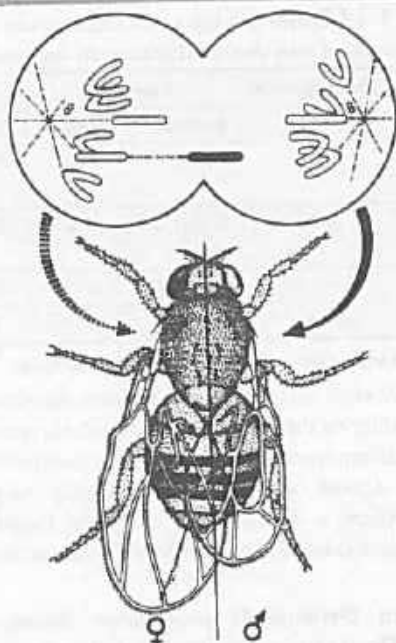


Fig : 7.1-24 Gynandromorph of *Drosophila* in which right half is male and left half is female

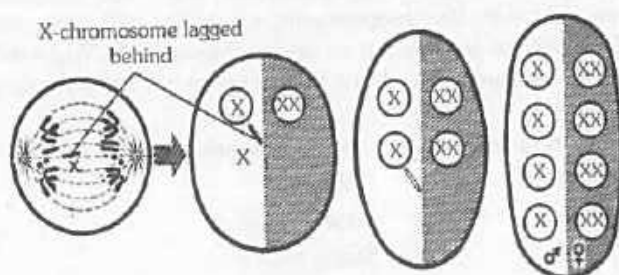


Fig : 7.1-25 Diagram to show origin of gynandromorphs

### Genic balance theory

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.

Table : 7.1-15 Ratio of X-chromosome to autosomes and the corresponding phenotype in *Drosophila*

Sex	Number of X-chromosomes	Number of autosomal set	Sex Index X/A ratio
Super female	XXX (3)	AA (2)	$3/2 = 1.5$
Normal female	XX (2)	AA (2)	$2/2 = 1.0$
Tetraploid	XXXX (4)	AAAA (4)	$4/4 = 1.0$
Tripled	XXX (3)	AAA (3)	$3/3 = 1.0$
Diploid	XX (2)	AA (2)	$2/2 = 1.0$
Haploid	X (1)	A (1)	$1/1 = 1.0$
Intersex	XX (2)	AAA (3)	$2/3 = 0.66$
Normal male	X (1)	AA (2)	$1/2 = 0.50$
Super male	X (1)	AAA (3)	$1/3 = 0.33$

**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.

### 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.

**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.

### Environmental theory of sex determination

In some animals, there is environmental determination of sex.

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.

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.

### 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 discoverer 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.



**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.

Table : 7.1-16

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

### 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 T. H. Morgan in 1910, while working on *Drosophila melanogaster*.

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.

### Types of sex linked inheritance

(1) **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 Criss-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*.

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

**Sex linked inheritance in man :** Colour blindness and Haemophilia are the two main sex linked or X-linked disease are found in man.

**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 deuteranopia. 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.

Table : 7.1-17 Inheritance of colour blindness

PARENTS				OFFSPRINGS			
Female		Male		Daughters		Sons	
Genotype	Phenotype	Genotype	Phenotype	Genotype	Phenotype	Genotype	Phenotype
XX	Normal	X <sup>c</sup> Y	Colourblind	XX <sup>c</sup>	Carrier	XY	Normal
XX <sup>c</sup>	Carrier	XY	Normal	(i) XX	Normal	XY	Normal
				(ii) XX <sup>c</sup>	Carrier	X <sup>c</sup> Y	Colourblind
XX <sup>c</sup>	Carrier	X <sup>c</sup> Y	Colourblind	(i) XX <sup>c</sup>	Carrier	XY	Normal
				(ii) X <sup>c</sup> X <sup>c</sup>	Colourblind	X <sup>c</sup> Y	Colourblind
X <sup>c</sup> X <sup>c</sup>	Colourblind	XY	Normal	X <sup>c</sup> X	Carrier	X <sup>c</sup> Y	Colourblind

**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 of necessary substrate, thromboplastin. It is of two types :

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

**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.

(2) **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.

(3) **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. e.g., Xeroderesia pigmentosa, Nephritis.

(4) **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. e.g., Baldness in man, Length of index finger.

(5) **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. e.g., Moustaches and beards in human males, breast in human females, milk secretion in human females.

### Pedigree analysis

A pedigree is a systematic listing (either as words or symbols) of the ancestors of a given individual or it may be the "family tree" for a number of individuals.

Pedigree analysis is carried out in order to word off possible disaster due to picking up of harmful genetic defects like dominant polydactyly (extra digits), syndactyly (joined digits) and brachydactyly (short digits), recessive haemophilia, deaf mutism, birth blindness, colour blindness, thalassemia, alkaptonuria, phenylketonuria, sickle cell anaemia attached ear lobes, tongue rolling etc.

**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.

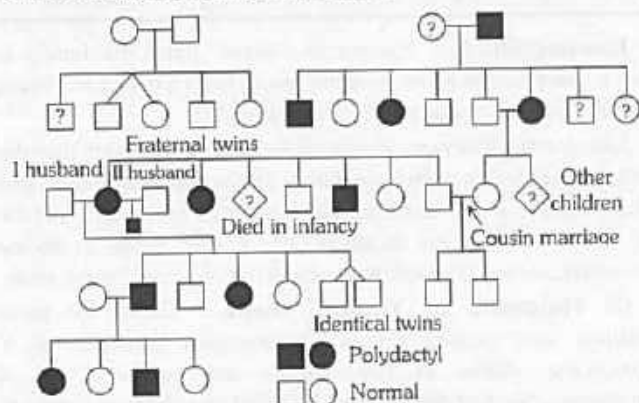


Fig : 7.1-26 Commonly used symbols in pedigree chart

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.

### Twins and I.Q.

**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.

(1) **Identical or monozygotic twins** : Identical twins are formed when one sperm fertilizes one egg to form a single zygote. They have the same genotype and phenotype and are of same sex. Differences if any, may be due to different environmental conditions.

(2) **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 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.

(3) **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.

**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.

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

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

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

Table : 7.1-18

I.Q.	Person	I.Q.	Person
0 - 24	Idiot	90 - 109	Average
25 - 49	Imbecile	110 - 119	Superior
50 - 69	Moron	120 - 139	Most superior
70 - 79	Dull	140 or more	Genius
80 - 89	Ordinary		

## Eugenics, Euthenics and Euphenics

### Eugenics

The term eugenics (Gr. Euegenes, 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. Eugenics can be divided into two types :

(1) **Positive eugenics** : In this approach of eugenics the future generations are improved by encouraging the inheritance of better traits.

(2) **Negative eugenics** : This is a negative aspect of improving mankind by restricting the transmission of poor and defective germplasm.

### 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.

### 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.

## Tips & Tricks

✍ In thalassemia, the  $\beta$  chain of haemoglobin is changed due to frame shift mutation as a result, bone marrow is not formed.

✍ Bateson coined the term Genetics, allele,  $F_1$ ,  $F_2$ , homozygous heterozygous and epistasis. He is also known as father of animal genetics.

✍ Johannsen coined the term genotype, phenotype, pure line.

✍ Mendel also observed that flower colour and colour of the seed coat may not assort independently.

✍ The genes for seed form in pea was present on chromosomes no. 7.

✍ Independent assortment is shown by the alleles present on different loci.

✍ Nilsson-Ehle (1908) was the first scientist to prove quantitative inheritance.

✍ Gene flow is spread of genes from one breeding population to another by migration.

✍ The genes, which enhance the effect of other gene, is also known as extender.

✍ Single copy genes : Represented only once in the whole genome.

✍ Multigenes : A group of nearly similar genes.

✍ Sutton and Winiweter (1900) expressed that number of chromosome is reduced to half in meiosis and doubled in fertilization.

✍ Sometimes two satellites are present in a chromosome these chromosome are called tandem SAT-Chromosomes.

✍ SAT Chromosomes are used as marker chromosomes.

✍ Genes modifying the effect of other gene called modifiers.

✍ Separation of a chromosome segment and its union to non-homologous chromosomes is called illegitimate crossing over.

✍ Study of phenotype to DNA sequence in gene come under forward genetics.

✍  $tt \times tt \rightarrow Tt$ , This type of inheritance is an example of de-novo mutation.

✍ One gene one enzyme theory was given by G. W. Beadle and E. L. Tatum they worked on *Neurospora crassa* (pink bread mould). Which is replaced by one gene one polypeptide theory was given by Yanofsky et al. (1965) utilizing bacterium *E. coli*.

✍ Two types of genes;

(1) Constitutive genes : It constantly express themselves e.g. enzymes of glycolysis, which are also known as house keeping gene, which lacks TATA boxes.

(2) Non constitutive genes : They express themselves only when needed, known as luxury genes Example- Inducible and Repressible genes.

✍ Morgan is called father of experimental genetics.

✍ Bateson is called father of modern genetics.

✍ **Heteropyknosis** : Darkly staining property of chromatin.

✍ Satellite is also called trabant.

✍ The frequency of an allele in an isolated population is due to genetic drift.

✍ 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.

✍ Inheritance of A, B, AB and O blood types in man was discovered by Bernstein in 1925.

✍ 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.





## Ordinary Thinking

## Objective Questions

## Mendelism

- Mendelism is related with [BVP 2003]
  - Heredity in living beings
  - Meiosis during sexual reproduction
  - Mutation in living organisms
  - None of the above
- The branch of botany dealing with heredity and variation is called [MP PMT 1998]
  - Geobotany
  - Sericulture
  - Genetics
  - Evolution
- Term 'genetics' was given by [CPMT 1994, 97; MP PMT 2007]
  - Mendel
  - Morgen
  - Bateson
  - Boveri
- The first great "geneticist" was [CBSE PMT 1991]
 

Or

Who is considered as father of genetics [NCERT]

  - Engler
  - Mendel
  - Schwann
  - Miller
- Mendel was born in [MP PMT 1999]
  - 17<sup>th</sup> century
  - 18<sup>th</sup> century
  - 19<sup>th</sup> century
  - 8<sup>th</sup> century
- Mendel was the native of [CPMT 1993]
  - France
  - Sweden
  - India
  - Austria
- Organism with two different allele is [Odisha JEE 2008]
  - Heterozygous and homozygous
  - Heterozygous for the allele
  - Homozygous for the allele
  - None of these
- In the first step of Monohybrid cross experiment, Mendel selected pea plants which were [MHCET 2015]
  - Pure tall as male and pure dwarf as female
  - Pure tall as female and pure dwarf as male
  - Heterozygous tall as male and pure dwarf as female
  - Heterozygous tall as female and pure dwarf as male
- Which one of the following cannot be explained on the basis of Mendel's Law of Dominance [CBSE PMT (Pre.) 2010]
  - Factor occur in pairs
  - The discrete unit controlling a particular character is called a factor
  - Out of one pair of factor one is dominant, and the other recessive
  - Alleles do not show any blending and both the characters recover as such in  $F_2$  generation
- A man having the genotype EEFfGgHH can produce P number of genetically different sperms, and a woman of genotype liLLMmNn can generate Q number of genetically different eggs. Determine the value of P and Q [WB JEE 2012]
  - $P=4, Q=4$
  - $P=4, Q=8$
  - $P=8, Q=4$
  - $P=8, Q=8$
- How many types of gametes will be produced by an individual having genotype AaBbcc [MHCET 2015]
  - Four
  - Three
  - Two
  - One
- In 1900 A.D. three biologists independently discovered Mendel's principles. They are [RPMT 1997; MP PMT 2002]
  - De Vries, Correns and Tschermak
  - Sutton, Morgan and Bridges
  - Avery, McLeod and McCarthy
  - Bateson, Punnett and Bridges
- When a dihybrid cross is fit into a punnett square with 16 boxes, the maximum number of different phenotypes available are [Kerala PMT 2008]
  - 8
  - 4
  - 2
  - 16
  - 12
- In a monohybrid cross between two heterozygous individuals, the number of pure homozygous individuals obtained in  $F_1$  generation is [Odisha JEE 2012]
  - 2
  - 4
  - 6
  - 8
- In Mendel's experiment how many different kinds of seeds are produced from a short plant with wrinkled seeds (ttrr) [Odisha JEE 2009]
  - 9
  - 4
  - 2
  - 1
- In garden pea, yellow colour of cotyledons is dominant over green and round shape of seed is dominant over wrinkled. When a plant with yellow and round seeds is crossed with a plant having yellow and wrinkled seeds, the progeny showed segregation for all the four characters. The probability of obtaining green round seeds in the progeny of the cross is [EAMCET 2009]
  - $\frac{1}{4}$
  - $\frac{1}{8}$
  - $\frac{1}{16}$
  - $\frac{3}{16}$
- Two pea plants were subjected cross pollination. Of the 183 plants produced in the next generation, 94 plants were found to be tall and 89 plants were found to be dwarf. The genotypes of the two parental plants are likely to be [KCET 2006]
  - TT and tt
  - Tt and Tt
  - Tt and tt
  - TT and TT
- A homozygous sweet pea plant with blue flowers (RR) and long pollen ( $R_0R_0$ ) is crossed with a homozygous plant having red flowers (rr) and round pollen ( $r_0r_0$ ). The resultant  $F_1$  hybrid is test crossed. Which of the following genotype does not appear in its progeny [EAMCET 2009]
  - $Rrr_0$
  - $RrRr_0$
  - $Rrr_0r_0$
  - $rrR_0r_0$
- Ratio of progeny when a red coloured heterozygote is crossed with a white coloured plant in which red colour is dominant in white colour [DPMT 2006]
  - 3 : 1
  - 1 : 1
  - 1 : 2 : 1
  - 9 : 3 : 3 : 1

20. A true breeding plant producing red flower is crossed with a pure plant producing white flower. Allele for red colour of flower is dominant. After selfing the plants of first filial generation, the proportion of plants producing white flower in the progeny would be [KCET 2009]
- (a)  $\frac{3}{4}$  (b)  $\frac{1}{4}$   
(c)  $\frac{1}{3}$  (d)  $\frac{1}{2}$
21. Which one of the following represents a test cross [Kerala PMT 2009]
- (a)  $Ww \times WW$  (b)  $Ww \times Ww$   
(c)  $Ww \times ww$  (d)  $WW \times WW$   
(e)  $ww \times ww$
22. How many type of genotypes are formed in  $F_2$  progeny obtained from self-pollination of a dihybrid  $F_1$  [MP PMT 2001, 06]
- (a) 6 (b) 3  
(c) 9 (d) 4
23. How many types of gametes may be produced by genotype  $D/d : E/e : F/f$  [RPMT 2006]
- Or
- How many types of gametes will be produced by individuals having genotype  $AaBbCc$  [NCERT; AIIMS 2004]
- (a) 27 (b) 8  
(c) 3 (d) 6
24. In his classic experiments on pea plants, Mendel did not use [AIPMT 2015]
- (a) Pod length (b) Seed shape  
(c) Flower position (d) Seed colour
25. Mendel is famous for his work on [CPMT 1994]
- (a) *Pisum* (b) *Drosophila*  
(c) *Neurospora* (d) *Oenothera*
26. Which of the following Mendel has selected for his experiment [Bihar MDAT 1995]
- (a) Garden pea (b) Pigeon pea  
(c) Sweet pea (d) Moong  
(e) None of these
27. How many different kinds of gametes will be produced by a plant having the genotype  $AABbCC$  [NCERT; CBSE PMT 2006]
- (a) Nine (b) Two  
(c) Three (d) Four
28. Mendel choose pea plants because [MP PMT 2003; BVP 2003]
- (a) They were cheap  
(b) They were having seven pairs of contrasting characters  
(c) They were easily available  
(d) Of great economic importance
29. In a population of 1000 individuals 360 belong to genotype  $AA$ , 480 to  $Aa$  and the remaining 160 to  $aa$ . Based on this data, the frequency of allele  $A$  in the population is [CBSE PMT 2014; GUJCET 2014]
- (a) 0.6 (b) 0.7  
(c) 0.4 (d) 0.5
30. Test cross in plants or in *Drosophila* involves crossing [CBSE PMT 2006; WB JEE 2010; CBSE PMT (Mains) 2011]
- (a) Crossing the  $F_1$  hybrid with a double recessive genotype  
(b) Crossing between two genotypes with dominant trait  
(c) Crossing between two genotypes with recessive trait  
(d) Crossing between two  $F_1$  hybrids
31. What is the correct sequence of the following events
1. Formation of the chromosome theory of heredity
  2. Experiments which proved that DNA is the hereditary material
  3. Mendel's laws of inheritance-discovery
- Code : [MP PMT 1993]
- (a) 1, 3 and 2 (b) 1, 2 and 3  
(c) 3, 1 and 2 (d) 2, 1 and 3
32. The term "genotype and gene" were coined by [DPMT 1993; MH CET 2004; MP PMT 2009]
- (a) H.J. Muller (b) T. Boveri  
(c) W.S. Sutton (d) W.L. Johanssen
33. Select the correct statement from the ones given below with respect to dihybrid cross [CBSE PMT (Pre.) 2010]
- (a) Tightly linked genes on the same chromosome show very few recombinations  
(b) Tightly linked genes on the same chromosome show higher recombinations  
(c) Genes far apart on the same chromosome show very few recombinations  
(d) Genes loosely linked on the same chromosome show similar recombinations as the tightly linked ones
34. When both alleles express their effect on being present together, the phenomenon is called [CPMT 2009; AIPMT 2015]
- Or
- Which Mendelism idea is depicted by a cross in which the  $F_1$  generation resembles both the parents [NEET 2013]
- (a) Dominance (b) Codominance  
(c) Pseudodominance (d) Amphidominance
35. What type of gametes will form by genotype  $RrYy$  [MP PMT 1993; RPMT 2002]
- (a)  $RY, Ry, rY, ry$  (b)  $RY, Ry, ry, ry$   
(c)  $Ry, Ry, Yy, ry$  (d)  $Rr, RR, Yy, YY$
36. Heterozygote tall plant ( $Tt$ ) is crossed with homozygous dwarf ( $tt$ ) plant. Then what will be percentage of dwarf plants in the next generation [Odisha JEE 2010]
- (a) 0% (b) 50%  
(c) 25% (d) 100%
37. In dihybrid cross, the pattern of inheritance represented by the punnett square given below, where yellow ( $Y$ ) is dominant over white ( $y$ ) and round ( $R$ ) is dominant over wrinkled ( $r$ ) seeds [NCERT]
- |    |    |    |    |    |
|----|----|----|----|----|
|    | YR | Yr | yR | yr |
| YR | F  | J  | N  | R  |
| Yr | G  | K  | O  | S  |
| yR | H  | L  | P  | T  |
| yr | I  | M  | Q  | U  |
- A plant of type 'H' will produce seeds with the genotype identical to seeds produced by the plants of
- (a) Type M (b) Type J  
(c) Type P (d) Type N

38. The term 'allelomorphic' implies [MP PMT 1997]  
 (a) Any two characters  
 (b) A pair of contrasting characters  
 (c) Sex linked characters  
 (d) A pair of non-contrasting characters
39. The alleles are [KCET 1994; MHCET 2004; RPMT 2005; MP PMT 2005; Haryana PMT 2005]  
 (a) A pair of genes governing a specific character such as tallness or dwarfness or alternate form of gene  
 (b) Multiple forms of genes  
 (c) Genes governing eye characters  
 (d) Genes present in allosomes
40. Alleles which show independent effect are called [CBSE PMT 1996; AMU (Med.) 2012]  
 (a) Supplementary alleles (b) Codominant alleles  
 (c) Epistatic alleles (d) Complementary alleles
41. When a gene exists in more than one form, the different forms are called [MP PMT 1994; CPMT 2002; AIPMT (Cancelled) 2015]  
 (a) Heterozygous (b) Complementary genes  
 (c) Genotypes (d) Alleles
42. An allele is said to be dominant if [NCERT; CBSE PMT 1999]  
 (a) It is expressed only in heterozygous combination  
 (b) It is expressed only in homozygous combination  
 (c) It is expressed in both homozygous and heterozygous condition  
 (d) It is expressed only in second generation
43. When a true breeding pea plant that has yellow seeds is pollinated by a plant that has green seeds, all the  $F_1$  plants have yellow seeds. This means that the allele for yellow is [MP PMT 1993]  
 Or  
 A character which is expressed in hybrid is called [WB JEE 2009]  
 (a) Heterozygous (b) Dominant  
 (c) Recessive (d) Lethal
44. In Mendel's experiments with garden pea, round seed shape (RR) was dominant over wrinkled seeds (rr), yellow cotyledon (YY) was dominant over green cotyledon (yy). What are the expected phenotypes in the  $F_2$  generation of the cross  $RRYY \times rryy$  [CBSE PMT 2006]  
 (a) Only wrinkled seeds with green cotyledons  
 (b) Round seeds with yellow cotyledons, and wrinkled seeds with yellow cotyledons  
 (c) Only round seeds with green cotyledons  
 (d) Only wrinkled seeds with yellow cotyledons
45. An organism with two identical alleles for a given trait is [NCERT; MP PMT 1993; CPMT 1994]  
 (a) Homozygous (b) Segregating  
 (c) Dominant (d) A hermaphrodite
46. How many different types of genetically different gametes will be produced by a heterozygous plant having the genotype  $AABbCc$  [CBSE PMT 1998]  
 (a) Two (b) Four  
 (c) Six (d) Nine
47. When two odd characters are present in a gene, this is known as [RPMT 1999]  
 (a) Bigamous (b) Heterogamous  
 (c) Polymorphic (d) Heteromorphic
48. When a tall plant with round seeds (TTRR) crossed with a dwarf plant with wrinkle seeds (ttrr), the  $F_1$  generation consists of tall plants with round seeds. What would be the proportion of dwarf plant with wrinkle seeds in  $F_1$  generation [KCET 2007]  
 (a)  $1/4$  (b)  $1/16$   
 (c) 0 (d)  $1/2$
49. In pea plants, yellow seeds are dominant to green. If a heterozygous yellow seeded plant is crossed with a green seeded plant, what ratio of yellow and green seeded plants would you expect in  $F_1$  generation [CBSE PMT 2007]  
 (a) 50 : 50 (b) 9 : 1  
 (c) 1 : 3 (d) 3 : 1
50. The dwarfness in plants of  $F_2$  generation is  
 (a) Recessive (b) Dominant  
 (c) Both the above (d) None of the above
51. A common test to find the genotype of a hybrid is by [CBSE PMT 2007]  
 (a) Crossing of one  $F_2$  progeny with male parent  
 (b) Crossing of one  $F_2$  progeny with female  
 (c) Studying the sexual behaviour of  $F_1$  progenies  
 (d) Crossing of one  $F_1$  progeny with male parent
52. When yellow round heterozygous pea plants are self fertilized, the frequency of occurrence of  $RrYY$  genotype among the offspring is [Kerala PMT 2012]  
 (a)  $9/16$  (b)  $3/16$   
 (c)  $2/16$  (d)  $1/16$   
 (e)  $6/16$
53. Mendel law is still true because [CPMT 1995]  
 (a) It takes place in sexually reproducing plants  
 (b) It takes place in asexually reproducing plants  
 (c) It takes place in both the above plants  
 (d) It takes place in apomictic reproducing plants
54. Character chosen by Mendel are located on how many chromosome [Haryana PMT 2005]  
 (a) 4 (b) 14  
 (c) 7 (d) 49
55. Which of the following is dominant character according to Mendel [AFMC 2000]  
 (a) Dwarf plant and yellow fruit  
 (b) Terminal fruit and wrinkled seed  
 (c) White testa and yellow pericarp  
 (d) Green coloured pod and rounded seed
56. Mendel found that the reciprocal crosses yielded identical results. From that he concluded that ..... [KCET 2010]  
 (a) There is independent assortment of trait  
 (b) Sex plays a role in deciding the dominance of a trait  
 (c) There is no dominance of any trait  
 (d) Sex has no influence on the dominance of traits
57. Test cross is used to [Odisha JEE 2010; CPMT 2010]  
 (a) Check heterozygosity in  $F_1$  generation  
 (b) Check heterozygosity in  $F_2$  generation  
 (c) Check independent assortment  
 (d) Check segregation
58. In Mendel's experiment nature of seed coat, flower colour, position of flower, pod colour, stem height etc. are referred to as [RPMT 1997]  
 (a) Alleles (b) Genotypes  
 (c) Phenotypes (d) All of above



59. A collection of plants and seeds having diverse alleles of all the genes of a crop is called [CBSE PMT (Pre.) 2011]  
(a) Genome (b) Herbarium  
(c) Germplasm (d) Gene library
60. Mendel enunciated [MP PMT 1995, 98]  
(a) Two principles of inheritance  
(b) Three principles of inheritance  
(c) Four principles of inheritance  
(d) Five principles of inheritance
61. Which of the following match is correct [CPMT 2010]  
(a) Independent assortment-segregation of factor  
(b) Lamarck-natural selection  
(c) Hatch and Slack-chemiosmotic theory  
(d) Peter Mitchell-proposed Z scheme
62. The first law of Mendel [CPMT 2003]  
(a) Law of inheritance  
(b) Law of variation  
(c) Law of independent assortment  
(d) Law of segregation
63. An exception to Mendel's law is [NCERT; Pb. PMT 2000; RPMT 2002, 06]  
(a) Law of independent assortment  
(b) Law of segregation  
(c) Law of dominance  
(d) Law of linkage
64. If Mendel had studied the seven traits using a plant with 12 chromosomes instead of 14, in what way would his interpretation have been different [NCERT; CBSE PMT 1998]  
(a) He could have mapped the chromosome  
(b) He would have discovered blending or incomplete dominance  
(c) He would not have discovered the law of independent assortment  
(d) He would have discovered sex linkage
65. Mendel's principle of segregation was based on the separation of alleles in the garden pea during [MP PMT 1993]  
(a) Pollination (b) Embryonic development  
(c) Seed formation (d) Gamete formation
66. A pure tall and a pure dwarf plant were crossed to produce offsprings. Offsprings were self crossed, then find out the ratio between true breeding tall to true breeding dwarf [MP PMT 2007]  
**Or**  
In hybridisation,  $Tt \times tt$  ( $F_1$  hybrid and a recessive parent) give rise to the progeny of ratio [CBSE PMT 1999; RPMT 1999; BVP 2000; Pb. PMT 2000; BHU 2003]  
(a) 1 : 1 (b) 3 : 1  
(c) 2 : 1 (d) 1 : 2 : 1
67. Mendel's law of heredity can be explained with the help of [CBSE PMT 1999]  
(a) Mitosis (b) Meiosis  
(c) Both mitosis and meiosis (d) None of the above
68. A cross between plants having  $RRYY$  and  $rryy$  composition will yield plants with [MP PMT 1993; BHU 2003]  
(a) Round and yellow seeds  
(b) Round and green seeds  
(c) Wrinkled and yellow seeds  
(d) Wrinkled and green seeds
69. Laws of Mendel are valid for [MP PMT 2005]  
(a) Asexual reproduction (b) Sexual reproduction  
(c) Vegetative reproduction (d) All above
70. Among the seven pairs of contrasting traits in pea plants as studied by Mendel, the number of traits related to flower, pod and seed respectively were [NCERT; MP PMT 1997; BVP 2003; AMU (Med.) 2009, 2012; WB-JEE 2011]  
(a) 2, 2, 2 (b) 2, 2, 1  
(c) 1, 2, 2 (d) 1, 1, 2
71. A cross in which an organism showing a dominant phenotype is crossed with the recessive parent in order to know its genotype is called [RPMT 1995; CPMT 1995; CBSE PMT (Pre.) 2010; CBSE PMT (Mains) 2010, 12; WB-JEE 2016]  
**Or**  
A cross between hybrid and recessive parent is [NCERT]  
(a) Monohybrid cross (b) Back cross  
(c) Test cross (d) Dihybrid cross
72. Some of the dominant traits studied by Mendel were [NCERT; AMU (Med.) 2012]  
(a) Round seed shape, constricted pod shape and axial flower position  
(b) Green pod colour, inflated pod shape and axial flower position  
(c) Yellow seed colour, violet flower colour and yellow pod colour  
(d) Axial flower position, green pod colour and green seed colour
73. The cross used to ascertain whether the plant is homozygous or heterozygous is [CBSE PMT 1994; BHU 1994, 2002; CPMT 2001; MP PMT 2006; AIIMS 2008]  
**Or**  
A cross between a homozygous recessive and a heterozygous plant is called [BHU 1995; MHCET 2003]  
(a) Linkage cross (b) Reciprocal cross  
(c) Test cross (d) Monohybrid cross
74. In a dihybrid cross where two parents differ in two pairs of contrasting traits like seed colour yellow ( $YY$ ) and seed colour green ( $yy$ ) with seed shape round ( $RR$ ) and seed shape wrinkled ( $rr$ ), the number of green coloured seeds ( $yy$ ) among sixteen products of  $F_2$  generation will be [NCERT; VITEEE 2008; AMU (Med.) 2012]  
(a) 2 (b) 4  
(c) 6 (d) 8
75. The genotypes of offspring in a genetic cross is called graphical representation to calculate the probability of all possible [Kerala PMT 2010]  
(a) Pedigree analysis (b) Karyotype  
(c) Punnett square (d) Chromosome map  
(e) Genotype ratio

76. A pea plant parent having violet coloured flowers with unknown genotype was crossed with a plant having white coloured flowers, in the progeny 50% of the flowers were violet and 50% were white. The genotypic constitution of the parent having violet coloured flowers was [DUMET 2010]  
(a) Homozygous (b) Merozygous  
(c) Heterozygous (d) Hemizygous
77. In man, the blue eye colour is recessive to the brown eye colour. If the boy has brown eye and his mother is blue eyed, what would be the phenotype of his father [KCET 2007]  
(a) Black eye (b) Brown eye  
(c) Green eye (d) Blue eye
78. When a cross is made between offspring and its parents, it is known as [MP PMT 1993]  
**Or**  
When a plant of  $F_1$  generation is crossed with homozygous dominant parents, it is known as [MP PMT 1998]  
(a) Monohybrid cross (b) Dihybrid cross  
(c) Back cross (d) Reciprocal cross
79. The colour based contrasting traits in seven contrasting pairs, studied by Mendel in pea plant were [NCERT; AMU (Med.) 2012]  
(a) 1 (b) 2  
(c) 3 (d) 4
80. Pure tall plants are crossed with pure dwarf plants. In the  $F_1$  generation all plants were tall. These tall plants of  $F_1$  generation were selfed and the ratio of tall to dwarf plants obtained was 3 : 1. This is called [BHU 2005]  
(a) Dominance (b) Inheritance  
(c) Co-dominance (d) Heredity
81. Which of the following is genotypic ratio of Mendel's monohybrid cross [NCERT; EAMCET 1993; KCET 1994; MP PMT 1996, 2005; J & K CET 2010; Odisha JEE 2010]  
(a) 1 : 3 (b) 3 : 1  
(c) 1 : 2 : 1 (d) 1 : 1 : 1 : 1
82. In a monohybrid cross when  $F_1$  is crossed with homozygous dominant parent then which type of offsprings will obtain [RPMT 2002]  
(a) Dominant : recessive 3:1 (b) Only recessive  
(c) Dominant : recessive 1:1 (d) No recessive
83. A dihybrid for qualitative trait is crossed with homozygous recessive individual of its type, the phenotypic ratio is [Odisha JEE 2005]  
(a) 1 : 2 : 1 (b) 3 : 1  
(c) 1 : 1 : 1 : 1 (d) 9 : 3 : 3 : 1
84. Which of the following depicts the Mendel's dihybrid ratio [NCERT; RPMT 1995; MP PMT 1995, 98; AFMC 2000; BVP 2001]  
(a) 3 : 1 (b) 9 : 3 : 3 : 1  
(c) 9 : 7 (d) 15 : 1
85.  $F_2$  generation in a Mendelian cross showed that both genotypic and phenotypic ratios are same as 1 : 2 : 1. It represents a case of [NCERT; CBSE PMT (Pre.) 2012]  
(a) Co-dominance  
(b) Dihybrid cross  
(c) Monohybrid cross with complete dominance  
(d) Monohybrid cross with incomplete dominance
86. Blue flowered and white flowered plant on crossing gave progeny of blue and white flowered in the ratio of 60 : 40. What ratio of blue and white is expected if the blue flowered are self pollinated [RPMT 1997]  
(a) 76 : 24 (b) 40 : 60  
(c) 52 : 48 (d) 84 : 16
87. Pure homozygous offsprings in a dihybrid cross in the  $F_2$  generation will be [AIIMS 1993]  
(a)  $1/2$  (b)  $1/4$   
(c)  $1/8$  (d)  $1/16$
88. In Mendelian monohybrid cross, phenotypic ratio in  $F_2$  is 3:1. How many types of gametes are formed in  $F_1$  generation [Bihar MDAT 1995]  
(a) Only one type (b) Two types  
(c) Four types (d) Eight types
89. When two genetic loci produce identical phenotypes in *cis* as well as in *trans* position, they are considered to be [CBSE PMT 1995; BHU 1999]  
(a) Pseudo alleles (b) The parts of the same gene  
(c) Multiple alleles (d) Different genes
90. If in a garden pea plant, a cross is made between red flowered and white flowered plants. What will be the phenotypic ratio in  $F_2$  generation [AFMC 2000; CBSE PMT 2002; Kerala CET 2003]  
(a) 1 : 2 : 1 (b) 9 : 3 : 3 : 1  
(c) 3 : 1 (d) 1 : 3
91. Test cross of dihybrid ratio is 1 : 1 : 1 : 1. It proves that [Odisha JEE 2011]  
(a)  $F_1$  hybrid produces four different progeny  
(b)  $F_1$  hybrid is homozygous  
(c) Two different progeny are produced by  $P_1$  parents  
(d) None of these
92. Mendel's principle of segregation means that the germ cells always receive [DUMET 2010]  
(a) One pair of alleles (b) One quarter of the genes  
(c) One of the paired alleles (d) Any pair of alleles
93. Mendel crossed a pure white-flowered recessive pea plant with a dominant pure red-flowered plant. The first generation of hybrids from the cross should show [MP PMT 1994, 97; RPMT 1995; AIIMS 1999, 2002; MHCET 2002; DPMT 2003]  
(a) 50% white-flowered and 50% red-flowered plants  
(b) All red-flowered plants  
(c) 75% red-flowered and 25% white-flowered plants  
(d) All white-flowered plants
94. If in a dihybrid cross Mendel had used two such characters which have linked, he would have faced difficulty in explaining the results on the basis of his [CBSE PMT 1990; RPMT 2005]  
**Or**  
In Mendelism, linkage was not observed due to [CPMT 1999; Odisha JEE 2011]  
(a) Law of segregation  
(b) Law of multiple factor hypothesis  
(c) Law of independent assortment  
(d) Law of dominance

95. If dwarf pea plant was treated with Gibberellic acid, it grew as tall as the pure tall pea plant. If this treated plant is crossed with a pure tall plant then the phenotypic ratio of is likely to be [BCECE 2005]  
 (a) All dwarf  
 (b) 50% dwarf 50% tall  
 (c) 75% tall 25% dwarf  
 (d) All tall
96. In a testcross involving  $F_1$  dihybrid flies, more parental-type offspring were produced than the recombinant type offspring. This indicates [NEET (Phase-I) 2016]  
 (a) The two genes are located on two different chromosomes  
 (b) Chromosomes failed to separate during meiosis  
 (c) The two genes are linked and present on the same chromosome  
 (d) Both of the characters are controlled by more than one gene
97. A farmer crossed a walnut combed chicken with a single combed one and obtained all walnut combed chickens in  $F_1$ . The genotype of the parents was [AIIMS 1993]  
 (a)  $Rr Pp \times rr pp$  (b)  $RR PP \times rr pp$   
 (c)  $RR pp \times rr pp$  (d)  $RR Pp \times rr pp$
98. When heterozygous red (dominant) flower is crossed with white flower the progeny would be [BVP 2004; DPMT 2007]  
 (a) 350 red : 350 white  
 (b) 450 red : 250 white  
 (c) 380 red : 320 white  
 (d) None of these
99. A double heterozygous tall plant with yellow colour (colour of cotyledon) is selfed the ratio of dwarf plants with green cotyledon is [MHCET 2002]  
 Or  
 Probability of genotype  $TTr$  in  $F_2$  generation of a dihybrid cross is [MH CET 2004]  
 (a)  $\frac{1}{16}$  (b)  $\frac{1}{4}$   
 (c)  $\frac{1}{6}$  (d)  $\frac{2}{16}$
100. In sweet pea plants the presence of dominant C and P genes is essential for development of purple colour. The ratio of plants producing flowers of different colours in the progeny of the cross  $Cc Pp \times Cc pp$  will be [AFMC 1993]  
 (a) 2 white and 6 purple coloured flowers  
 (b) 2 purple and 6 white coloured flowers  
 (c) 3 white and 5 purple coloured flowers  
 (d) 3 purple and 5 white coloured flowers
101. When a tall and red flowered individual is crossed with a dwarf and white flowered individual, phenotype in the progeny is dwarf and white. What will be the genotype of tall and red flowered individual [AFMC 2001]  
 Or  
 Which genotype represents a true dihybrid condition [CBSE PMT 1991]  
 (a)  $TTRR$  (b)  $TtRR$   
 (c)  $TtRr$  (d)  $TTRr$
102. Normal maize has starchy seeds which remain smooth when dry. A mutant form has sugary seeds which go crinkled when dry. When a mutant was crossed with a normal plant, an  $F_1$  was produced which had smooth seeds. What would be the relative ratios of the different seed types, if the  $F_1$  was allowed to self [AIIMS 1993]  
 (a) 1 smooth : 3 sugary (b) 3 smooth : 1 sugary  
 (c) 1 smooth : 1 sugary (d) All sugary
103. If a plant heterozygous for tallness is selfed, the  $F_2$  generation has both tall and dwarf plants. This proves the principle of [Odisha JEE 2011]  
 Or  
 When heterozygous tall plants are self-pollinated than tall and dwarf plants are obtained this is explain to [RPMT 1999]  
 (a) Dominance (b) Segregation  
 (c) Independent assortment (d) Incomplete dominance
104. From a single ear of corn, a farmer planted 200 kernels which produced 140 tall and 40 dwarf plants. The genotype of these offsprings are most likely [DPMT 1993]  
 (a)  $TT$ ,  $Tt$  and  $tt$  (b)  $TT$  and  $tt$  only  
 (c)  $TT$  and  $Tt$  only (d)  $Tt$  and  $tt$  only
105. From a cross  $Aa BB \times aa BB$ , following genotypic ratio will be obtained in  $F_1$  generation [NCERT; CBSE PMT 1990]  
 (a) 1  $Aa BB$  : 1  $aa BB$   
 (b) 1  $Aa BB$  : 3  $aa BB$   
 (c) 3  $Aa BB$  : 1  $aa BB$   
 (d) All  $Aa BB$  : No  $aa BB$
106. Hybrid breakdown refers to the condition when offspring are physiologically inferior to the following generation [DPMT 2004]  
 (a)  $F_1$  (b)  $F_2$   
 (c)  $P_1$  (d) All of these
107. If the cells of an organism heterozygous for two pairs of characters viz.  $Aa$  and  $Bb$  undergo meiosis, what will be the genotypes of the gametes produced [JIPMER 1994]  
 (a)  $Aa$  and  $Bb$  (b)  $AB$ ,  $aB$ ,  $Ab$  and  $ab$   
 (c)  $aB$  and  $Ab$  (d)  $Ab$  and  $ab$
108. When  $AABB$  and  $aabb$  are crossed, in  $F_2$  generation the ratio of  $AaBb$  will be [RPMT 1997; J & K CET 2008]  
 (a) 1/16 (b) 2/16  
 (c) 8/16 (d) 4/16
109. In a typical mendelian cross which is a dihybrid cross, one parent is homozygous for both dominant traits and another parent is homozygous for both recessive traits. In the  $F_2$  generation, both parental combinations and recombinations appear. The phenotypic ratio of parental combinations to recombinations is [KCET 2011]  
 (a) 10 : 6 (b) 12 : 4  
 (c) 9 : 7 (d) 15 : 1
110. In Mendelian dihybrid cross when heterozygous Round Yellow are self crossed, Round Green offsprings are represented by the genotype [Kerala PMT 2011]  
 (a)  $RrYy$ ,  $RrYY$ ,  $RRYy$  (b)  $Rryy$ ,  $RRyy$ ,  $rryy$   
 (c)  $rYy$ ,  $rYY$  (d)  $Rryy$ ,  $RRyy$   
 (e)  $RrYy$ ,  $rryy$ ,  $Rryy$



111. If a cross is made between AA and aa, the nature of  $F_1$  progeny will be [CPMT 2004]  
 (a) Genotypically AA, phenotypically a  
 (b) Genotypically Aa, phenotypically a  
 (c) Genotypically Aa, phenotypically A  
 (d) Genotypically aa, phenotypically A
112. When a tall plant with rounded seeds (TTRR) is crossed with a dwarf plant with wrinkled seeds (ttrr), the  $F_1$  generation consists of tall plants with rounded seeds. How many types of gametes an  $F_1$  plant would produce [CPMT 2004]  
 (a) One (b) Three  
 (c) Four (d) Eight
113. In a plant, red fruit (R) is dominant over yellow fruit (r) and tallness (T) is dominant over shortness (t). If a plant with RRTt genotype is crossed with a plant that is rrtt. [CBSE PMT 2004; AIIMS 2007]  
 (a) 75% will be tall with red fruit  
 (b) All the offspring will be tall with red fruit  
 (c) 25% will be tall with red fruit  
 (d) 50% will be tall with red fruit
114. A self-fertilizing trihybrid plant forms [CBSE PMT 2004]  
 (a) 8 different gametes and 16 different zygotes  
 (b) 8 different gametes and 32 different zygotes  
 (c) 8 different gametes and 64 different zygotes  
 (d) 4 different gametes and 16 different zygotes
115. Match the genetic phenomena with their respective ratios
- | Column - I                   | Column - II      |
|------------------------------|------------------|
| A. Inhibitory gene ratio     | 1. 9 : 3 : 4     |
| B. Complementary gene ratio  | 2. 1 : 1 : 1 : 1 |
| C. Recessive epistasis ratio | 3. 12 : 3 : 1    |
| D. Dihybrid test cross ratio | 4. 13 : 3        |
| E. Dominant epistasis ratio  | 5. 9 : 7         |
- [Kerala PMT 2007, 09]
- (a) A-5, B-4, C-3, D-2, E-1  
 (b) A-4, B-5, C-1, D-2, E-3  
 (c) A-1, B-2, C-4, D-3, E-5  
 (d) A-2, B-1, C-4, D-5, E-3  
 (e) A-5, B-4, C-1, D-2, E-3
116. If a tall plant is crossed with a dwarf plant and obtained progeny is half tall and half dwarf plants. Then the genotype of progeny will be [BHU 2003; RPMT 2006; WB JEE 2011, 12]  
 (a) TT × tt (b) Tt × tt  
 (c) TT × Tt (d) Tt × Tt
117. Mendel's law of independent assortment is applicable for [Odisha JEE 2002]  
 (a) All genes in all organism  
 (b) All genes of pea plant only  
 (c) All linked genes only  
 (d) All non-linked genes only
118. Hybrid vigour is induced by [CPMT 2001]  
 (a) Clonal selection (b) Crossing of plant  
 (c) Crossing two plants (d) Species differentiation
119. A tall true breeding garden pea plant is crossed with a dwarf true breeding garden pea plant. When the  $F_1$  plant were selfed the resulting genotypes were in the ratio of [NEET (Phase-I) 2016]  
 (a) 1 : 2 : 1 :: Tall homozygous : Tall heterozygous : Dwarf  
 (b) 1 : 2 : 1 :: Tall heterozygous : Tall homozygous : Dwarf  
 (c) 3 : 1 :: Tall : Dwarf  
 (d) 3 : 1 :: Dwarf : Tall
120. A true breeding plant is [NEET (Phase-II) 2016]  
 (a) Always homozygous recessive in its genetic constitution  
 (b) One that is able to breed on its own  
 (c) Produced due to cross pollination among unrelated plants  
 (d) Near homozygous and produces offspring of its own kind
121. Which one from those given below is the periods for Mendel's hybridization experiments [NEET 2017]  
 (a) 1856-1863 (b) 1840-1850  
 (c) 1857-1869 (d) 1870-1877
122. Among the following characters, which one was not considered by Mendel in his experiments on pea [NEET 2017]  
 (a) Stem - Tall or Dwarf  
 (b) Trichomes - Glandular or non-glandular  
 (c) Seed - Green or Yellow  
 (d) Pod - Inflated or Constricted

### Interaction of gene and cytoplasmic inheritance

1. Some genomic representation of skin colour are given below  
 (i) AA bb CC (ii) AA bb cc  
 (iii) AA BB CC (iv) aa bb cc  
 Which of the option is correct for showing the darkness of colour of the skin in decreasing order [GUJCET 2014]  
 (a) i → iv → ii → iii (b) iii → ii → i → iv  
 (c) iii → i → ii → iv (d) i → iii → ii → iv
2. Fruit colour in squash is an example of [CBSE PMT 2014]  
 (a) Complementary genes (b) Inhibitory genes  
 (c) Recessive epistasis (d) Dominant epistasis
3. Leaf colour in *Mirabilis jalapa* is an example of [DPMT 2006]  
 (a) Non-Mendelian inheritance  
 (b) Mendelian inheritance  
 (c) Chemical inheritance  
 (d) Both (b) and (c)
4. Genes present in the cytoplasm of eukaryotic cells, are found in [AIIMS 2008]  
 (a) Mitochondria and inherited via egg cytoplasm  
 (b) Lysosomes and peroxisomes  
 (c) Golgi bodies and smooth endoplasmic reticulum  
 (d) Plastids and inherited via male gamete

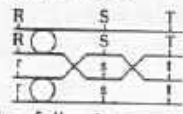
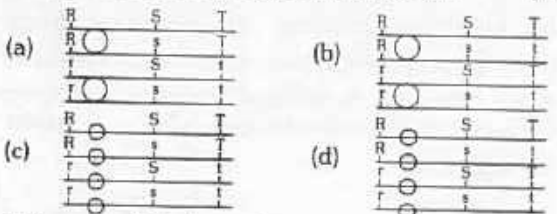
5. *Lathyrus odoratus* is an example of which of the following genes [CPMT 2000]  
(a) Supplementary genes (b) Complementary genes  
(c) Lethal genes (d) Codominant genes
6. Besides activating the egg another role of a sperm is to carry to egg [Odisha JEE 2009]  
(a) RNA (b) Mitochondria  
(c) DNA (d) Ribosomes
7. In which one of the following, complementary gene interaction ratio of 9 : 7 is observed [Kerala PMT 2009]  
(a) Fruit shape in Shepherd's purse  
(b) Coat colour in mouse  
(c) Feather colour in fowl  
(d) Flower colour in pea  
(e) Four 'O' clock plant
8. Two or more independent genes present on different chromosomes which determine nearly same phenotype are called [Odisha JEE 2012]  
(a) Supplementary genes (b) Complementary genes  
(c) Duplicate genes (d) None of these
9. A human male produces sperms with the genotypes AB, Ab, aB and ab pertaining to two diallelic characters in equal proportions. What is the corresponding genotype of the person [CBSE PMT 2007]  
(a) AaBb (b) AaBB  
(c) AABb (d) AABB
10. In which mode of inheritance do you expect more maternal influence among the offspring [CBSE PMT 2006]  
(a) Y-linked (b) X-linked  
(c) Autosomal (d) Cytoplasmic
11. In *Mirabilis* a hybrid for red (RR) and white (rr) flower produces pink (Rr) flower. A plant with pink flower is crossed with white flower the expected phenotypic ratio is [RPMT 2006]  
(a) Red : Pink : White (1 : 2 : 1)  
(b) Pink : White (1 : 1)  
(c) Red : Pink (1 : 1)  
(d) Red : White (3 : 1)
12. Grain colour in wheat is determined by three pairs of polygenes. Following the cross AABBCC (dark colour) × aabbcc (light colour), in  $F_2$  generation what proportion of the progeny likely to resemble either parent [AIIMS 2005, 07, 08]  
(a) None (b) Less than 5 per cent  
(c) One third (d) Half
13. The most likely reason for the development of resistance against pesticides in insects damaging a crop is [CBSE PMT 2004]  
(a) Directed mutation  
(b) Acquired heritable changes  
(c) Random mutations  
(d) Genetic recombination
14. In shorthorn cattle genes for red ( $r_1$ ) and white ( $r_2$ ) coat colour occur. Crosses between red ( $r_1r_2$ ) and white ( $r_2r_2$ ) produced ( $r_1r_2$ ) roan. This is an example of [BHU 2003]  
(a) Complementary genes (b) Epistasis  
(c) Codominance (d) Incomplete dominance
15. In *Antirrhinum* two plants with pink flowers were hybridized. The  $F_1$  plants produced red, pink and white flowers in the proportion of 1 red, 2 pink and 1 white. What could be the genotype of the two plants used for hybridization. Red flower colour is determined by RR, and white by rr genes [CBSE PMT (Mains) 2010]  
(a) rrrr (b) RR  
(c) Rr (d) rr
16. The gene interaction when one gene masks the effect [DPMT 2006; CPMT 2011]  
Or  
When a gene pair hides the effect of another, the phenomenon is called [NCERT]  
(a) Complementary gene action  
(b) Supplementary gene action  
(c) Duplicate gene action  
(d) Epistasis
17. What will be the ratio in  $F_2$  generation if red-flowered variety of *Mirabilis jalapa* is crossed with white-flowered variety [NCERT; MP PMT 1995, 98, 99; BHU 2003]  
Or  
Phenotypic ratio in plant *Snapdragon* in  $F_2$  is [AMU (Med.) 2010]  
(a) 1 : 1 : 1 : 1 (b) 1 : 2 : 1  
(c) 2 : 1 (d) 3 : 1
18. What would be the colour of flowers in  $F_1$  progeny as a result of a cross between homozygous red and homozygous white-flowered *Snapdragon* [MP PMT 1997]  
(a) Red (b) White  
(c) Red and white (d) Pink
19. 9 : 3 : 3 : 1 ratio is modified to 9 : 7 ratio due to [NCERT; CPMT 1998; CBSE PMT 2001]  
(a) Complementary gene (b) Epistatic gene  
(c) Hypostatic gene (d) Supplementary gene
20. Incomplete dominance is found in [MP PMT 2001; J & K CET 2010; Kerala PMT 2010]  
(a) *Pisum sativum* (b) *Antirrhinum majus*  
(c) Both (a) and (b) (d) None of these
21. Complete dominance is absent in [JIPMER 2002]  
Or  
Incomplete dominance is shown by [MH CET 2004]  
(a) *Pisum sativum* (b) *Mirabilis jalapa*  
(c) *Lathyrus odoratus* (d) *Oenothera lamarckiana*
22. When an albino female plant of maize is crossed with normal green male plant, all plants in the progeny are albino because [CMC Vellore 1994]  
(a) Plastids are inherited through maternal plants  
(b) Albinism is dominant over green character  
(c) The crossing results in structural changes in green plastids  
(d) Green plastids of male parents become mutated

23. Kappa particles indicate [MP PMT 2003]  
 (a) Nuclear inheritance  
 (b) Cytoplasmic inheritance  
 (c) Mutation  
 (d) Nucleo-cytoplasmic inheritance
24. *Mirabilis jalapa* is a good example of [AIIMS 2001]  
 (a) Complete dominance (b) Plastid inheritance  
 (c) Both (a) and (b) (d) None of the above
25. Which of the following is associated with multiple phenotypes [CPMT 1999; AIIMS 2000; JIPMER 2001; AFMC 2001; AMU (Med.) 2006; DPMT 2006; Kerala PMT 2008; Odisha JEE 2009, 11; J & K CET 2010]  
 (a) Epistasis (b) Pleiotropy  
 (c) Polygenic inheritance (d) Mutation
26. Human skin colour is controlled by several gene pairs. Let us assume here that there are just three gene pairs on different chromosomes and that for each pair there are two alleles – an incompletely dominant one that codes for melanin deposition. If a very dark skinned person marries a very light skinned woman, what will be the chance that their offspring will have very dark skin [Kerala PMT 2006]  
 (a) 0 (b) 1/4  
 (c) 5/8 (d) 9/64  
 (e) 3/64
27. After crossing two plants, the progenies are found to be male sterile. The phenomenon is found to be maternally inherited and is due to some genes which reside in [CBSE PMT 1997; Pb. PMT 2000]  
 (a) Nucleus (b) Chloroplast  
 (c) Mitochondria (d) Cytoplasm
28. Plasmids so found in bacteria are [MP PMT 2003]  
 (a) Extra nuclear DNA (b) Food particles  
 (c) Dead protoplasmic parts (d) None of the above
29.  $F_1$  hybrid is intermediate between the two parents. The phenomenon is [MHCET 2004]  
 (a) Codominance (b) Dominance  
 (c) Blending inheritance (d) Incomplete dominance
30. Extranuclear inheritance (cytoplasmic inheritance) is a consequence of presence of genes in [MP PMT 1993; CBSE PMT 2004]  
 (a) Ribosomes and chloroplasts  
 (b) Lysosomes and ribosomes  
 (c) Mitochondria and chloroplasts  
 (d) Endoplasmic reticulum and mitochondria
31. Genes for cytoplasmic male sterility in plants are generally located in [CBSE PMT 2005]  
 (a) Mitochondrial genome (b) Cytosol  
 (c) Chloroplast genome (d) Nuclear genome
32. The phenotypic ratio obtained in quantitative inheritance of a dihybrid cross is [DPMT 2004]  
 Or  
 In a cross between red kernelled and white kernelled varieties of wheat showing polygenic inheritance the phenotypic ratio in  $F_2$  generation will be. [MH CET 2015]  
 (a) 1 : 2 : 1 (b) 1 : 4 : 6 : 4 : 1  
 (c) 1 : 6 : 15 : 20 : 15 : 6 : 1 (d) 9 : 3 : 3 : 1
33. A plasmid [MP PMT 1999]  
 (a) Lives together with chromosome  
 (b) Shows dependent assortment  
 (c) Can replicate independently  
 (d) Cannot replicate
34. The  $F_2$  generation offspring in a plant showing incomplete dominance, exhibit [KCET 2006; MP PMT 2009; J & K CET 2012]  
 (a) Variable genotypic and phenotypic ratios  
 (b) A genotypic ratio of 1 : 1  
 (c) A phenotypic ratio of 3 : 1  
 (d) Similar phenotypic and genotypic ratios of 1 : 2 : 1

### Linkage and Crossing over

1. The evidence that crossing over occurs at four stranded stage and not at two stranded stage of the chromosomes, comes from [DPMT 1993]  
 (a) 2 : 2 : 2 : 2 arrangement of ascospores in *Neurospora*  
 (b) 4 : 4 arrangement of ascospores in *Neurospora*  
 (c) Studies of meiosis in maize  
 (d) Studies on linkage maps of chromosomes in *Drosophila*
2. The four daughter cells derived from a single meiosis differ from each other due to [BHU 1994]  
 (a) Difference in chromosome number  
 (b) Crossing over only  
 (c) Independent assortment of chromosomes only  
 (d) Crossing over as well as independent assortment of chromosomes
3. Coupling and repulsion are the two faces of [NCERT; JIPMER 1994]  
 (a) Crossing over (b) Linkage  
 (c) Chiasmata (d) Mutation
4. The map distance between genes A and B is 3 units, between B and C 10 units and between C and A 7 units. The order of the genes in a linkage map constructed on the above data would perhaps be [CMC Vellore 1994]  
 (a) A, B, C (b) A, C, B  
 (c) B, C, A (d) B, A, C
5. Alleles of different genes that are on the same chromosome may occasionally be separated by a phenomenon known as [MP PMT 1993]  
 Or  
 Linked gene are separated by [CPMT 1994]  
 (a) Pleiotropy (b) Epistasis  
 (c) Continuous variation (d) Crossing over
6. Which one of the following pairs is correctly matched [MP PMT 1993; AIPMT 2015]  
 (a) Morgan Discovered the process of linkage  
 (b) Linus Pauling Isolated DNA for the first time  
 (c) Francis Crick Discovered the phenomenon of transformation  
 (d) H. Khorana Discovered that a sequence of 3 nucleotides codes for a single amino acid
7. Which of the following animal was selected by Morgan for studying linkage [MHCET 2015]  
 (a) *Apis indica*  
 (b) *Agrobacterium tumefaciens*  
 (c) *Drosophila melanogaster*  
 (d) *E. coli*



8. In Morgan's experiments on linkage, the percentage of white eyed, miniature winged recombinants in  $F_2$  generation is [Kerala PMT 2009]  
 (a) 1.3 (b) 37.2  
 (c) 62.8 (d) 73.2  
 (e) 98.7
9. Two genes  $R$  and  $Y$  are located very close on the chromosomal linkage map of maize plant. When  $RRYY$  and  $rryy$  genotypes are hybridized, the  $F_2$  segregation will show [CBSE PMT 2007]  
 (a) Higher number of the recombinant types  
 (b) Segregation in the expected 9 : 3 : 3 : 1 ratio  
 (c) Segregation in 3 : 1 ratio  
 (d) Higher number of the parental types
10. The number of linkage group in  $E. coli$  is/are [DPMT 2007, 10; MP PMT 2012]  
 (a) 4 (b) 2  
 (c) 1 (d) 5
11. Crossing-over occurs in the [NCERT; Bihar MDAT 1995; MP PMT 1995, 2007, 09, 12; JIPMER 2002; CPMT 2009]  
 (a) Leptotene stage (b) Pachytene stage  
 (c) Anaphase stage (d) Diakinesis stage
12. Mendel observed that some characters did not assort independently. Later researchers found it to be due to [MP PMT 1995, 98]  
 (a) Crossing-over  
 (b) Linkage  
 (c) Dominance of one trait over the other  
 (d) Amitosis
13. Exchange of genetic material between chromatids of homologous chromosomes during meiosis is called [CBSE PMT 1996; DPMT 2007; MP PMT 2012; AIIMS 2013]  
**Or**  
 Recombination is involved in the process of [DUMET 2009]  
 (a) Synapsis (b) Chiasmata  
 (c) Transformation (d) Crossing over
14. The scientists who have given the theory of linkage are [NCERT; MP PMT 2001]  
 (a) Morgan and Castle (b) Beadle and Tatum  
 (c) Watson and Crick (d) Bateson and Punnett
15. Which one of the following is the most suitable medium for culture of *Drosophila melanogaster* [CBSE PMT 2006]  
 (a) Ripe banana (b) Cow dung  
 (c) Moist bread (d) Agar agar
16. Depending upon the distance between any two genes which is inversely proportional to the strength of linkage, cross overs will vary from [NCERT; AMU (Med.) 2012]  
 (a) 50–100% (b) 0–50%  
 (c) 75–100% (d) 100–150%
17. Linkage decreases the frequency of [CPMT 1998]  
 (a) Hybrid (b) Dominant allele  
 (c) Recessive allele (d) Both (a) and (b)
18. Crossing over in diploid organism is responsible for [NCERT; CBSE PMT 1991, 98; MP PMT 2010]  
 (a) Dominance of genes  
 (b) Linkage between genes  
 (c) Segregation of alleles (genes)  
 (d) Recombination of linked allele (genes)
19. The figure shows a homologous (bivalent) pair of chromosomes during meiosis
- 
- Which one of the following option correctly illustrates the final products of the second meiotic division [NCERT]
- 
20. Linkage was first observed in [AFMC 2000]  
 (a) Field pea (b) Sweet pea  
 (c) Pea (d) Grass pea
21. What is the unit of crossing over  
 (a) Cistron (b) Mutton  
 (c) Recon (d) None of the above
22. Crossing over that results in genetic recombination in higher organisms occurs between [CBSE PMT 2004; DPMT 2004; BVP 2004; VITEEE 2006; DUMET 2010]  
 (a) Two daughter nuclei  
 (b) Two different bivalents  
 (c) Sister chromatids of a bivalent  
 (d) Non-sister chromatids of a bivalent
23. When closely placed genes on the same chromosome are inherited together the phenomenon is known as [Kerala PMT 2004]  
 (a) Qualitative inheritance (b) Crossing over  
 (c) Gene interaction (d) Multiple allelism  
 (e) Linkage
24. Genetic maps of chromosomes are based on the frequency of [Kerala PMT 2006]  
 (a) Non-disjunction (b) Translocation  
 (c) Dominance (d) Genetic recombination  
 (e) Chromosomal aberration
25. Number of linkage group in *Pisum sativum* is [BVP 2004]  
**Or**  
 How many pairs of contrasting characters in pea plants were studied by Mendel in his experiments [AIPMT (Cancelled) 2015]  
 (a) 2 (b) 5  
 (c) 7 (d) 9
26. Sexual reproduction leads to [CPMT 2002; RPMT 2005]  
 (a) Genetic recombination (b) Polyploidy  
 (c) Aneuploidy (d) Euploidy
27. In the diplotene stage [DPMT 1993]  
 (a) Chromosomes are thin and long  
 (b) Homologous chromosomes undergo crossing over and chiasmata are seen  
 (c) Bivalents become very short and chiasmata move towards ends of chromosomes  
 (d) Chromosomes have reached poles

28. For the preparation of genetic maps, the recombination frequencies between genes are additive over short distances but not over long distances due to [BHU 1994]  
 (a) Multiple cross overs (b) Lethal mutation  
 (c) Epistasis (d) Synaptonemal complex
29. Genetic recombination occur through [CPMT 2002; RPMT 2005, 06]  
 (a) Mitosis and fertilization (b) Mitosis and meiosis  
 (c) Meiosis and fertilization (d) None of the above
30. When synapsis is complete all along the chromosome, the cell is said to have entered a stage called [AIIMS 2005]  
 (a) Zygotene (b) Pachytene  
 (c) Diplotene (d) Diakinesis
31. What will be the number of linkage groups in maize if it has 10 pairs of chromosomes [MP PMT 1999]  
**Or**  
 What will be the number of linkage groups in a cell having  $2n = 20$  [NCERT]  
 (a) 5 (b) 10  
 (c) Zero (d) 20
32. In case of incomplete linkage, the parental combinations obtained in  $F_1$  generation are [Odisha JEE 2012]  
 (a) 100% (b) More than 50%  
 (c) 25% (d) Less than 50%
33. Chiasma shows the sites of [NCERT; MHCET 2003]  
 (a) Spindle formation (b) Synapsis  
 (c) Crossing over (d) None of these
34. Which of the following statements is not true of two genes that show 50% recombination frequency [NEET 2013]  
 (a) If the genes are present on the same chromosome, they undergo more than one crossovers in every meiosis  
 (b) The genes may be on different chromosomes  
 (c) The genes are tightly linked  
 (d) The genes show independent assortment

### Chromosomes and Genes

1. How many pairs of homologous chromosomes are present in human [Odisha JEE 2008]  
 (a) 46 (b) 44  
 (c) 22 (d) 23
2. The name chromatin was coined by [Kerala PMT 2010]  
 (a) Flemming (b) Robert Brown  
 (c) George Palade (d) Camillo Golgi  
 (e) Rudolf Virchow
3. Polytenic chromosomes were first observed by  
 (a) Batanetzky-1980  
 (b) Heitz and Bauer-1935  
 (c) Balbiani-1881  
 (d) Stevens and Wilson-1905
4. The terminal end of a chromosome is called [MP PMT 1999]  
 (a) Centromere (b) Chromomere  
 (c) Telomere (d) Metamere
5. Which of the following is the correct sequence of units of genetics arranged in descending order of size [BHU 2012]  
 (a) Gene  $\rightarrow$  Cistron  $\rightarrow$  Muton  $\rightarrow$  Recon  
 (b) Gene  $\rightarrow$  Muton  $\rightarrow$  Cistron  $\rightarrow$  Recon  
 (c) Gene  $\rightarrow$  Recon  $\rightarrow$  Cistron  $\rightarrow$  Muton  
 (d) Gene  $\rightarrow$  Cistron  $\rightarrow$  Recon  $\rightarrow$  Muton
6. Centromere is a part of chromosome which helps in the [CBSE PMT 1995; MP PMT 1998; BHU 2001]  
 (a) Division of centrosomes  
 (b) Formation of spindle fibres  
 (c) Movement of chromosomes  
 (d) Formation of nuclear spindle
7. The chromosome number in meiocyte is 34. The organism could be [KCET 2015]  
 (a) Ophioglossum (b) Dog  
 (c) Onion (d) Apple
8. The distance between the genes a, b, c and d in mapping units are  $a-d = 3.5$ ;  $b-c = 1$ ;  $a-b = 6$ ;  $c-d = 1.5$ ;  $a-c = 5$ . Find out the sequence of arrangement of these genes [Kerala PMT 2008]  
 (a) acdb (b) abcd  
 (c) acbd (d) adbc  
 (e) adcb
9. Number of autosomes in human sperm is [MP PMT 1995, 2003, 10]  
 (a) 11 (b) 22  
 (c) 44 (d) 45
10. In a certain species of animal, genes T, U, V and W occur on the same chromosome. The following table gives their cross-over values (COVs)
- | linked gene pair | COV |
|------------------|-----|
| T and U          | 25  |
| T and V          | 5   |
| V and U          | 30  |
| U and W          | 10  |
| V and W          | 20  |
- Which of the following option shows the appropriate order of the genes on the chromosome [NCERT]  
 (a) V, W, T, U (b) T, V, W, U  
 (c) T, W, U, V (d) V, T, W, U
11. The long and short arms of chromosome are designated respectively as [DUMET 2010]  
 (a) p and q arms (b) q and p arms  
 (c) m and p arms (d) l and s arms
12. An unfertilized human egg contains [CBSE PMT 1991, 92; CPMT 1993; MP PMT 1993, 99, 2000; Manipal 1995]  
 (a) Two X chromosomes  
 (b) One X and Y chromosome  
 (c) One Y chromosome only  
 (d) One X chromosome only

13. The structure present over chromosome is  
[MP PMT 1995, 2003; CBSE PMT 1997; BHU 2002]

Or

The structure of the chromosome to which spindle fibre is attached is  
[MP PMT 1993, 95; Pb. PMT 2000]

- (a) Nucleolus (b) Centromere  
(c) Centrochome (d) Golgi bodies
14. Match column I with column II and select the correct option

Column I (Name of the organism)		Column II (Haploid chromosome number in gamete)	
A.	<i>Ophioglossum</i>	1.	23
B.	Rice	2.	24
C.	Potato	3.	12
D.	Man	4.	630

[Kerala PMT 2011]

- (a) A-1, B-2, C-3, D-4 (b) A-2, B-3, C-4, D-1  
(c) A-3, B-4, C-2, D-1 (d) A-4, B-3, C-2, D-1  
(e) A-4, B-3, C-1, D-2
15. Who used the word "chromosome" [MP PMT 1997]  
(a) Huxley (b) Flemming 1888  
(c) Kolliker 1888 (d) Waldeyer 1888
16. The theory of recombination of linked gene due to crossing over of chromosome during zygotene of meiosis was put forward by  
(a) T.H. Morgan (b) Punnet  
(c) Mendel (d) Connes
17. Solenoid is a structure of [Kerala PMT 2006]  
(a) Nucleosomal organization with 10 nm thickness  
(b) Condensed chromatin fibre with 30 nm diameter  
(c) Highly condensed form of chromatid with 300 nm thickness  
(d) Well organised chromatid with 700 nm thickness  
(e) Well organised chromosome with 1400 nm thickness
18. Total collection of genes at any time in a unit of evolution is [Odisha JEE 2009]

Or

The sum of genes in a population is called [CPMT 1993]

- (a) Gene bank (b) Gene library  
(c) Genome (d) Gene pool
19. The distance between two genes in a chromosome is measured in cross-over units which represent [AIIMS 1998; BHU 2008]  
(a) Ratio of crossing over between them  
(b) Percentage of crossing over between them  
(c) Number of crossing over between them  
(d) None of these
20. The chromosomal number in the meiocytes of housefly is [Kerala PMT 2011]  
(a) 8 (b) 12  
(c) 21 (d) 23  
(e) 34

21. A chromosome, in which the centromere is situated close to its end so that one arm is very short and other very long is

[MP PMT 1997, 98, 2002; AIIMS 2002;

DUMET 2009; AMU (Med.) 2009; AIPMT (Cancelled) 2015]

- (a) Acrocentric (b) Metacentric  
(c) Sub-metacentric (d) Telocentric
22. In polytene chromosomes dark bands are visible. These bands are formed by the apposition of [AFMC 2006]  
(a) Protein particles  
(b) Chromomeres on chromonemata  
(c) Nucleosomes  
(d) None

23. In eukaryotes basic structural unit made of histone and DNA [DPMT 2006]

Or

What are those structures that appear as beads-on-string in the chromosomes when viewed under electron microscope

[CBSE PMT (Pre.) 2011]

- (a) Nucleosome (b) Nucleolus  
(c) Chromosome (d) Lysosome
24. Heterochromatin remains condensed in which part of chromosome [RPMT 2006]  
(a) Secondary constriction-I (b) Secondary constriction-II  
(c) Telomeres (d) Both (a) and (b)
25. Chromosomal theory of inheritance was based on [MP PMT 2006]

- (a) Segregation of genes  
(b) Diploidy and haploidy  
(c) Sex linkage  
(d) Presence of sex chromosomes
26. Number of (approximately) genes in *E. coli* are [CPMT 2005]  
(a) 4000 (b) 6000  
(c) 10000 (d) 18000
27. The largest gene in man is [Kerala PMT 2009]  
(a) Dystrophin  
(b) Insulin gene  
(c) Beta globin gene of haemoglobin  
(d) Tumor suppressor gene  
(e) Oncogene
28. Tizio and Levan's contribution is very significant because they [CBSE PMT 1993]  
(a) Gave the number of human chromosomes  
(b) Pointed out mutational changes  
(c) Identified Barr bodies  
(d) Detected sex linkage
29. Depending upon size and centromere position, the 46 chromosomes have been divided into a number of groups [CBSE PMT 1993; MP PMT 2002]  
(a) 6 (b) 5  
(c) 7 (d) 10
30. The grouping of human chromosomes is based on [CBSE PMT 1993]  
(a) Secondary constrictions alone  
(b) Dot-like satellites alone  
(c) Banding patterns alone  
(d) All the above



31. Geneticist plot the relative locations of genes on chromosomes by which of these methods [JIPMER 1993]  
 (a) Using powerful microscopes  
 (b) Calculating the number of genes  
 (c) Determining the frequency of crossing over  
 (d) Exposing animals to radiations
32. Genes are made up of [MP PMT 2009]  
 Or  
 Genes are chemically [BHU 2002]  
 (a) Histones (b) Hydrocarbons  
 (c) Polynucleotides (d) Lipoproteins
33. Genes are located in [MP PMT 2012]  
 (a) Ribosomes (b) Lysosomes  
 (c) Chromosomes (d) Spherosomes
34. The chemical nature of chromatin is [WB JEE 2009; MP PMT 2013]  
 (a) Nucleic acids  
 (b) Nucleic acids & histone proteins  
 (c) Nucleic acids, histone & non histone proteins  
 (d) Nucleic acids & non- histone proteins
35. Experimental verification of the chromosomal theory of inheritance was given by [Kerala PMT 2011]  
 (a) Gregor Johann Mendel (b) Hugo de Vries  
 (c) Langdon Down (d) Henking  
 (e) Thomas Hunt Morgan
36. Number of histone proteins in each nucleosome core is [Odisha JEE 2012]  
 (a) 8 (b) 10  
 (c) 12 (d) 14
37. Karyotype is [CPMT 2009]  
 (a) Chromosome complement which is specific for each species of living organism  
 (b) All organism possessing same type of chromosomes  
 (c) Division of nucleus  
 (d) None of the above
38. What would be the number of chromosomes of the aleurone cells of a plant with 42 chromosomes in its roots tip cells [CBSE PMT (Pre.) 2011]  
 (a) 21 (b) 42  
 (c) 63 (d) 84
39. The genes, which are confined to differential region of Y chromosome only, are called [CBSE PMT 1994; AIIMS 1998; MP PMT 2000; CPMT 2003]  
 (a) Mutant (b) Autosomal  
 (c) Holandric (d) Completely sex-linked
40. Crossing over takes place at a stage between [MP PMT 2007]  
 (a) Leptotene and diplotene (b) Pachytene and diplotene  
 (c) Zygotene and pachytene (d) Zygotene and diplotene
41. What is the chromosome number of plasmodium [RPMT 2000]  
 (a) 18 (b) 14  
 (c) 10 (d) 9
42. The polytene chromosomes were discovered for the first time in [CBSE PMT 1995; BHU 2012; MP PMT 2012]  
 (a) *Chironomus* (b) Fruitfly  
 (c) *Drosophila* (d) House fly
43. A child receives [CBSE PMT 1995]  
 (a) 25% genes from his father  
 (b) 50% genes from his father  
 (c) 75% genes from his father  
 (d) 100% genes from his father
44. Telomere repetitive DNA sequences control the function of eukaryote chromosomes because they [CBSE PMT 2007]  
 (a) Act as replicons  
 (b) Are RNA transcription initiator  
 (c) Help chromosome pairing  
 (d) Prevent chromosome loss
45. The genes are in the form of [CPMT 1998]  
 (a) Sequence of nucleotide (b) Base pair  
 (c) Proportion of base pair (d) None of these
46. The genome of *Caenorhabditis elegans* consists of [Kerala PMT 2007]  
 (a) 3 billion base pairs and 30,000 genes  
 (b) 180 million base pairs and 13,000 genes  
 (c) 4.7 million base pairs and 4,000 genes  
 (d) 97 million base pairs and 18,000 genes  
 (e) 12 million base pairs and 6,000 genes
47. Match the numbers of genes given in Column – I with names of organisms in Column – II and choose the correct alternatives
- | Column – I |                        | Column – II |                                |
|------------|------------------------|-------------|--------------------------------|
| A.         | 450 to 700 genes       | 1.          | <i>Escherichia coli</i>        |
| B.         | 4000 genes             | 2.          | <i>Drosophila melanogaster</i> |
| C.         | 13,000 genes           | 3.          | <i>Mycoplasma</i>              |
| D.         | 32,000 to 50,000 genes | 4.          | <i>Homo sapiens</i>            |
| E.         | 35,000 to 45,000 genes | 5.          | <i>Oryza sativa</i>            |
- [Kerala PMT 2007]
- (a) A-2, B-1, C-5, D-3, E-4  
 (b) A-3, B-1, C-2, D-5, E-4  
 (c) A-3, B-2, C-1, D-5, E-4  
 (d) A-2, B-3, C-1, D-5, E-4  
 (e) A-1, B-3, C-2, D-5, E-4
48. Arrangement of chromosomes in the order of decreasing length is termed [Manipal 2005]  
 (a) Pedigree (b) Eugenetics  
 (c) Idiogram (d) Dysengenics
49. The condensation of the chromosomes are maximal with visible centromeres at which phase of cell cycle [MP PMT 1994]  
 (a)  $G_1$  phase (b) S phase  
 (c)  $G_2$  phase (d) M phase

50. A normal metaphase chromosome with a middle centromere is [MP PMT 1994, 2012; Kerala CET 2005; CPMT 2005]  
Or  
Chromosomes whose arms are equal are called [KCET 1999]  
(a) Metacentric (b) Sub-metacentric  
(c) Acrocentric (d) Telocentric
51. The males of grasshoppers and moths possess two sets of autosomes and [MP PMT 1994]  
(a) X and Y chromosomes  
(b) Only X chromosome  
(c) Only Y chromosome  
(d) Neither X nor Y chromosome
52. Relative morphologies of chromosomes of an individual indicate his/her [MP PMT 1994]  
(a) Genotype (b) Phenotype  
(c) Pedigree chart (d) Karyotype
53. For making important contributions in respect of the nature of gene, the Noble Prize was rewarded to [MP PMT 2010]  
(a) T. H. Morgan (b) De Vries  
(c) H.J. Muller (d) Darwin
54. In humans, most number of genes are located on chromosome [Kerala PMT 2012]  
(a) 1 (b) 6  
(c) X (d) 21  
(e) Y
55. Number of autosomes in a normal female is [CPMT 1995; J & K CET 2012]  
(a) 21 (b) 22  
(c) 23 (d) 44
56. The point at which the polytene chromosomes appear to be attached together is known as [CBSE PMT 1995; KCET 2006]  
(a) Centriole (b) Chromocentre  
(c) Centromere (d) Chromomere
57. Balbiani discovered special type of chromosome from the salivary gland of *Chironomus* larva which are recognized by the presence of [MP PMT 1995]  
(a) Bands (b) Loops  
(c) Both bands and loops (d) All of the above
58. Who used the frequency of recombination between gene pairs on the same chromosome as a measure of the distance between genes and mapped their position on the chromosome [Kerala PMT 2012]  
(a) Gregor Mendel (b) Correns  
(c) Tschermak (d) Watson and Crick  
(e) Alfred Sturtevant
59. Polytene or giant chromosomes are found in [KCET 1994; AFMC 1999; DPMT 2006; WB JEE 2012; WB-JEE 2016]  
(a) Salivary glands of man  
(b) Salivary glands of woman  
(c) Salivary glands of all animals  
(d) Salivary glands of *Drosophila*
60. Lampbrush chromosomes are visible [CBSE PMT 1996; DPMT 2006]  
(a) In diplotene of meiosis (b) In prophase of meiosis  
(c) In interphase (d) In metaphase of meiosis
61. In sex linkage, the speciality is [BHU 2006]  
(a) Atavism (b) Criss-cross inheritance  
(c) Reversion (d) Gene flow
62. Doubling of the chromosomes is termed as [Odisha JEE 2012]  
(a) Duplication (b) Transcription  
(c) Translation (d) None of these
63. Lampbrush chromosomes are found inside [CPMT 1999; MP PMT 2002]  
(a) Salivary glands of *Drosophila*  
(b) Salivary glands of silk moth  
(c) Oocytes of frog  
(d) Nucleus of man
64. Genetically active area of chromosome is called [BVP 2000]  
(a) Euchromatin (b) Heterochromatin  
(c) Heptan (d) Cistron
65. *Drosophila melanogaster* has 8 chromosomes in somatic cell. How many linkage groups will be there [BVP 2000]  
(a) 4 (b) 8  
(c) 2 (d) 5
66. Two sister chromatids are attached with [BVP 2000]  
(a) Spindle fibre (b) Centromere  
(c) Chromocentre (d) Chromatid
67. Balbiani rings are present in [BVP 2000; MH CET 2001; KCET 2004]  
(a) Polysomes (b) Autosomes  
(c) Polytene chromosomes (d) None of the above
68. Chromosomes can be stained with [MH CET 2001; WB JEE 2009; MP PMT 2009]  
(a) Iodine (b) Aniline blue  
(c) Safranin (d) Aceto carmine
69. In plant A,  $2n = 12$  and in plant B,  $2n = 16$ , then the ploidy number of cross breeding plant is [Odisha JEE 2010]  
(a) 14 (b) 28  
(c) 12 (d) 16
70. Crossing over takes place between [RPMT 2001]  
(a) Two chromosomes  
(b) Two non-homologous chromosomes  
(c) Two homologous chromosomes  
(d) None
71. Whereas the number of chromosomes is reduced to half in first reduction division of meiosis, then what is the need for second mitotic division [MP PMT 2001]  
(a) For the segregation of replicated chromosomes  
(b) For equal distribution of haploid chromosomes  
(c) For the formation of four gametes  
(d) For equal distribution of genes on chromosomes
72. In humans chromosomal condition of male is [JIPMER 2002]  
(a) 44 AA + XO (b) 44 AA + XX  
(c) 44 AA + XY (d) 44 AA + XXY
73. The carriers of hereditary material are [MP PMT 2002]  
(a) Chromosomes (b) Gene  
(c) Gametes (d) Gametocytes

74. The twenty third pair of chromosomes in man is known as [MP PMT 2002]  
(a) Chromatid (b) Heterosome  
(c) Autosome (d) Gene
75. In recent past human chromosomes have been studied by a technique using specific, often fluorescent dyes, known as [MP PMT 2002]  
(a) Dyeing technique (b) Banding technique  
(c) Ultra dyeing technique (d) Karyotyping technique
76. The chromosomes as thread like structures in nucleus was first described by [MP PMT 2002]  
(a) Mendel (b) Strasburger  
(c) Darwin (d) Levitzky
77. The function of chromosomes of carrying the genetic information from one cell generation to another is performed by [MP PMT 2002]  
(a) RNA (b) DNA  
(c) Histones (d) Calcium
78. The chromosomes which determine the somatic characters are called [MH CET 2002]  
(a) Sex chromosomes (b) Heterosomes  
(c) Autosomes (d) None of the above
79. The linkage map of X-chromosome of fruit fly has 66 units with yellow body gene (y) at one end and bobbed hair (b) gene at the other end. The recombination frequency between these two genes (y and b) should be [CBSE PMT 2003]  
(a) 100 % (b) 66 %  
(c) 50 % (d) 5.50 %
80. Chromosome number is [MP PMT 2003]  
(a) Fixed for a species (b) Fixed for an ecosystem  
(c) Fixed for a community (d) Fixed for a biosphere
81. Science which links heredity with environments is [MP PMT 2004]  
(a) Genetics (b) Gene ecology  
(c) Ecology (d) Ecophysiology
82. In man sexlinked characters are mainly transmitted through [MP PMT 2004]  
(a) Y-chromosome  
(b) Autosomes  
(c) X-chromosome  
(d) X-chromosome, Y-chromosome and Autosomes
83. A male human is heterozygous for autosomal genes A and B and is also hemizygous for hemophilic gene h. What proportion of his sperms will be abh [CBSE PMT 2004]  
(a)  $\frac{1}{16}$  (b)  $\frac{1}{4}$   
(c)  $\frac{1}{8}$  (d)  $\frac{1}{32}$
84. The recessive genes located on X-chromosome in humans are always [CBSE PMT 2004]  
(a) Expressed in males (b) Expressed in females  
(c) Lethal (d) Sub-lethal
85. The total number of nitrogenous bases in human genome is estimated to be about [AIIMS 2004, 08]  
(a) 3.5 million (b) 35 thousand  
(c) 35 million (d) 3.1 billion
86. In order to calculate map distance of genes on a chromosome, one must know the [AIEEE Pharmacy 2004]  
(a) Number of mutant genes  
(b) Cross over percentage  
(c) Recombination frequency of each gene locus  
(d) Non-cross over percentage
87. At a particular locus, frequency of 'A' allele is 0.6 and that of 'a' is 0.4. What would be the frequency of heterozygotes in a random mating population of equilibrium [CBSE PMT 2005]  
(a) 0.16 (b) 0.48  
(c) 0.36 (d) 0.24
88. Polytene chromosomes are formed by [Kerala PMT 2004; CPMT 2005]  
(a) Endoreduplication of chromosomes  
(b) Somatic pairing of homologous chromosomes  
(c) Somatic pairing of non-homologous chromosomes  
(d) Germinal pairing of non-homologous chromosomes
89. Telomerase is an enzyme which is a [CBSE PMT 2005]  
(a) Repetitive DNA (b) RNA  
(c) Simple protein (d) Ribonucleoprotein
90. Percentage of recombination between A and B is 9% and C is 17%, B and C is 26%, then the arrangement of genes is [Odisha JEE 2004]  
(a) ABC (b) ACB  
(c) BCA (d) BAC.
91. Chromosome complement with  $2n-1$  is called as [BHU 2005, 08; WB JEE 2008]  
(a) Monosomy (b) Nullsomy  
(c) Trisomy (d) Tetrasomy
92. A gene is said to be dominant, if [CBSE PMT 1992, 2002]  
(a) It is never expressed in any condition  
(b) It is expressed only in heterozygous condition  
(c) It expresses its effect only in homozygous stage  
(d) It is expressed both in homozygous and heterozygous conditions
93. Chromosomes were seen first time by [RPMT 2003]  
(a) Waldeyer (b) Flemming  
(c) Hofmeister (d) Strasburger
94. Genes located at the same locus of chromosomes are called [CBSE PMT 1997; AIPMT (Cancelled) 2015]  
(a) Polygenes (b) Oncogenes  
(c) Multiple alleles (d) None of these
95. Jumping genes are found in [MP PMT 2003]  
(a) Eukaryotes  
(b) Bacteriophage  
(c) Bacteria  
(d) Eukaryotes and prokaryotes
96. Some genes in bacteria and virus may code for more than one polypeptide, they are called as [Odisha JEE 2011]  
(a) Overlapping genes (b) Jumping gene  
(c) Split gene (d) None of these
97. The person who discovered 'Y' chromosomes was  
(a) Mc Carthy (b) Mc Clung  
(c) Gregor Mendel (d) Netti Stevens
98. "Nu body" was shown by [BVP 2003]  
(a) Darlington (b) Johanssen  
(c) Woodcock (d) Temin and Baltimore



99. "Cytochimera" means  
(a) Cells having haploid number of chromosomes  
(b) Cells having two nuclei  
(c) Cells having different chromosomes other than vegetative cells  
(d) None of the above
100. Genes carried on chromosomes was first proved by [VITEEE 2006]  
(a) Mendel (b) Watson  
(c) Crick (d) Bridges
101. In *Pisum sativum* there are 14 chromosomes. How many pairs with different chromosomal composition can be prepared [BHU 2005]  
(a) 14 (b) 7  
(c)  $2^{14}$  (d)  $2^7$
102. Which of the following is incorrectly paired [Kerala CET 2005]  
(a) Sry-gene-X-chromosome  
(b)  $2n-2$ -nullisomic  
(c) Nucleoid prokaryote  
(d) Polytene chromosome-*Drosophila*  
(e) Trisomy-Down's syndrome
103. Gene controls [MP PMT 2010]  
(a) Heredity but not protein synthesis  
(b) Protein synthesis but not heredity  
(c) Both heredity and protein synthesis  
(d) Biochemical action of some enzymes
104. Smallest structure having the power of replicating itself is  
Or  
The factor responsible for expression of character transmitted from parents to offsprings [MH CET 2006]  
(a) Chloroplast (b) Gene  
(c) Mitochondria (d) Ribosome
105. The core of nucleosome is made up of [AFMC 2000; MH CET 2007]  
(a)  $H_1, H_2A, H_2B, H_3$  (b)  $H_1, H_2A, H_2B, H_4$   
(c)  $H_1, H_2A, H_2B, H_3, H_4$  (d)  $H_2A, H_2B, H_3, H_4$
106. Nucleosome consists of [MP PMT 1999, 2012]  
(a) Nucleolus (b) Genes  
(c) Microfilaments (d) Histones
107. Structural element of chromatin is [WB JEE 2011]  
(a) Histone (b) Acid protein and DNA  
(c) Nuclear matrix (d) Nucleosome
108. The salivary gland chromosomes in the dipteran larvae, are useful in gene mapping because [CBSE PMT 2005]  
(a) These are fused  
(b) These are much longer in size  
(c) These are easy to stain  
(d) They have endoreduplicated chromosomes
109. Nucleosomes are [MP PMT 2003]  
(a) Units of DNA (b) Units of RNA  
(c) Units of proteins (d) Units of chromosomes
110. Carrier of hereditary is [MP PMT 2005]  
(a) Gene (b) DNA  
(c) Chromosome (d) All of above
111. Plant A is having chromosome no.  $2n = 12$  and B having  $2n = 16$ . Both are crossed to form allotetraploid C. What is the chromosome number of C [Odisha JEE 2010]  
(a) 32 (b) 14  
(c) 28 (d) 7
112. Holandric genes are [DPMT 1993; BHU 2000; AIIMS 2010; MP PMT 2013]  
(a) Carried by 'X' chromosomes  
(b) Carried by different parts of 'Y' chromosomes  
(c) Carried by 'X' and 'Y' chromosomes  
(d) Carried by autosomes
113. Which organism was used by Beadle and Tatum to proposed one gene-one enzyme hypothesis [CPMT 2004; BVP 2004; CBSE PMT 2007; MP PMT 2007]  
(a) *E. coli* (b) *Nostoc*  
(c) *Drosophila* (d) *Neurospora*
114. A gene is made up of [MP PMT 1996, 2011; BVP 2002]  
(a) DNA (b) RNA  
(c) Either DNA or RNA (d) Amino acids
115. Which one of the following conditions of the zygotic cell would lead to the birth of a normal human female child [CMC Vellore 1993; CBSE PMT (Mains) 2011]  
(a) Only one X chromosome  
(b) One X and one Y chromosome  
(c) Two X chromosomes  
(d) Only one Y chromosome
116. Separation of the two chromatids of a chromosome takes place in mitosis during [DPMT 1993; AMU (Med.) 2006]  
(a) Prophase (b) Anaphase  
(c) Metaphase (d) Telophase
117. The modern concept of gene is [CPMT 1994]  
(a) A segment of DNA capable of crossing over  
(b) A functional unit of DNA  
(c) A segment of DNA  
(d) A segment of chromosome
118. "One gene one enzyme" theory was proposed by [NCERT; CPMT 1994, 2006; BHU 1995, 2008; AMU (Med.) 2000; DPMT 2001, 04; MP PMT 2003, 06; BVP 2004; RPMT 2006; CBSE PMT 2006]  
(a) G.W. Beadle and E.L. Tatum  
(b) O.T. Avery and M. McCarthy  
(c) J.H. Tijo and A. Levan  
(d) C.E. Ford and J.H. Tijo
119. One functional unit of gene which specifies synthesis of one polypeptide is known as [NCERT; MP PMT 2001; J & K CET 2008]  
Or  
The equivalent of a structural gene [NEET (Phase-II) 2016]  
(a) Recon (b) Clone  
(c) Codon (d) Cistron
120. The theory of jumping genes was propounded by or Noble prize for the concept of jumping gene was given to [MHCET 2001; BVP 2001, 03, 04; MP PMT 2002; CPMT 2003; BHU 2006, 12]  
(a) Mendel (b) Morgan  
(c) Barbara Mc Clintock (d) Sanger

121. The terms *cistron*, *recon* and *muton* were proposed by

[NCERT; MP PMT 2009]

- (a) W. Ingram (b) Bateson  
(c) J. Lederberg (d) S. Benzer

122. A normal spontaneous rate for a single gene is one mutation in every.....replication [Odisha JEE 2004]

- (a)  $10^3$  to  $10^5$  (b)  $10^5$  to  $10^7$   
(c)  $10^6$  to  $10^9$  (d)  $10^7$  to  $10^{10}$

123. Genes are [MP PMT 2005, 12]

- (a) Morphological units (b) Hereditary units  
(c) Basic units (d) All of these

124. The eukaryotic chromosomes are made up of [MP PMT 1994, 2011; WB JEE 2010]

- (a) DNA (b) RNA  
(c) DNA and proteins (d) DNA and lipids

125. Chromosome Y is [VITEEE 2006]

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

126. Nucleosomes are bounded by

- (a) RNA (b) Histone  $H_4$   
(c) Histone  $H_3$  (d) DNA

127. Who postulated the 'Chromosome Theory of Inheritance' [MP PMT 1997; Kerala PMT 2007]  
Or

The behaviour of the chromosomes was parallel to the behaviour of genes during meiosis was noted by [Keral PMT 2012]

- (a) De Vries (b) Mendel  
(c) Sutton and Boveri (d) Morgan

128. In split genes, the coding sequences are called [NCERT; CBSE PMT 1995]

- (a) Cistrons (b) Operons  
(c) Exons (d) Introns

129. Which one of the following true [MP PMT 2005]

- (a) One gene one protein  
(b) One gene one polypeptide  
(c) One gene many polypeptide  
(d) All of the above

130. The bacterial genome refers to the total number of genes located upon a **or** The term 'genome' refers to the total number of genes combined in a [CPMT 1994, 2010; AIIMS 1994; MP PMT 1994, 95, 98; Manipal 1995; Odisha JEE 2009]

- (a) Haploid set of chromosomes  
(b) Diploid set of chromosomes  
(c) Tetraploid set of chromosomes  
(d) Hexaploid set of chromosomes

131. Different types of chromosomes can be recognised by the position of the following separating the two arms [KCET 1994]

- (a) Centromere (b) Genes  
(c) Spindle (d) Nucleus

132. Nucleosome core is intimately associated with [WB-JEE 2016]

- (a) 160 bp of DNA (b) 210 bp of DNA  
(c) 250 bp of DNA (d) 100 bp of DNA

## Multiple allelism

1. 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 [NCERT; AIIMS 1992]

- (a) Any autosome  
(b) X chromosome of female  
(c) X chromosome of male  
(d) Either on autosome or X chromosome

2. In humans, height and skin colour shows a lot of variation. They are example of [CBSE PMT 2006, 07; AIIMS 2008, 13; VITEEE 2008; MH CET 2015]

- (a) Multiple alleles  
(b) Pleiotropic inheritance  
(c) Polygenic / Quantitative inheritance  
(d) Pseudoalleles

3. In human beings, the colour of skin is controlled by [CPMT 1995; Kerala CET 1999, 2002; CBSE PMT 2007]

- (a) Multiple alleles (b) Lethal genes  
(c) Polygenic effect (d) None of these

4. Which of the following is genetically dominant in man [MP PMT 2007]

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

5. Which of the following genotypes does not produce any sugar polymer on the surface of the RBC [Kerala PMT 2010]

- (a)  $I^A I^A$  (b)  $I^B i$   
(c)  $I^A I^B$  (d)  $i i$   
(e)  $I^B I^B$

6. ABO blood grouping is controlled by gene *I* which has three alleles and show co-dominance. There are six genotypes. How many phenotypes in all are possible [KCET 2007; CBSE PMT (Pre./Mains) 2010; Kerala PMT 2012]

- (a) Six (b) Three  
(c) Four (d) Five

7. Inheritance of ABO blood grouping is an example of [J & K CET 2008; Kerala PMT 2010]

- (a) Dominance (b) Co-dominance  
(c) Incomplete dominance (d) Both (a) & (b)

8. The most popularly known blood grouping is the ABO grouping. It is named ABO and not ABC, because "O" in it refers to having [CBSE PMT 2009]

- (a) Other antigens besides A and B on RBCs  
(b) Overdominance of this type of the genes for A and B types  
(c) One antibody only-either anti-A or anti-B on the RBCs  
(d) No antigens A and B on RBCs

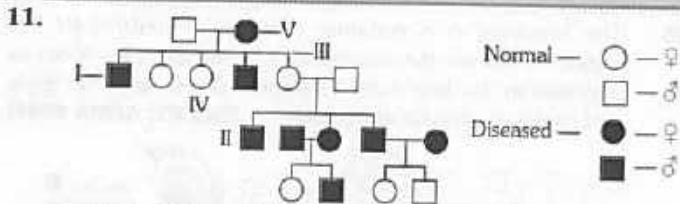
9. Inheritance of blood group is a condition of [CBSE PMT 1990; AFMC 2006; Kerala PMT 2008, 09]

- (A) Co-dominance (B) Incomplete dominance  
(C) Multiple allelism (D) Multiple gene  
(a) A, B (b) B, D  
(c) B, C (d) A, D  
(e) A, C

10. A woman with blood group 'O' has a child with blood group 'O'. She claims that a man with blood group 'A' as the father of her child. What would be the genotype of the father, if her claim is right [BHU 2008; EAMCET 2009]  
 (a)  $I^O I^O$  (b)  $I^A I^B$   
 (c)  $I^A I^O$  (d)  $I^B I^O$
11. The condition of erythroblastosis foetalis occurs only when [KCET 2012]  
 (a) The husband is  $Rh^+$  and wife is  $Rh^-$   
 (b) The husband is  $Rh^-$  and wife is  $Rh^+$   
 (c) The mother is  $Rh^+$  and foetus is  $Rh^-$   
 (d) The mother is  $Rh^-$  and foetus is  $Rh^+$
12. The offspring produced from a marriage have only O or A blood groups. Of the genotypes given below, the possible genotypes of the parents would be [KCET 2009]  
 (a)  $I^A I^A$  and  $I^A I^O$  (b)  $I^O I^O$  and  $I^O I^O$   
 (c)  $I^A I^A$  and  $I^O I^O$  (d)  $I^A I^O$  and  $I^O I^O$
13. Blood stains are found at the site of a murder. If DNA profiling technique is to be used for identifying the criminal, which of the following is ideal for use [KCET 2009]  
 (a) Serum (b) Erythrocytes  
 (c) Leucocytes (d) Platelets
14. A person with blood group 'A' can be given blood of which group [MP PMT 1993, 2005; RPMT 2006]  
 (a) A and B (b) B and O  
 (c) A and O (d) A, B, AB and O
15. Antisera used to detect Rh blood group [MP PMT 1993]  
 (a) Anti A (b) Anti B  
 (c) Anti C (d) Anti D
16. Human blood groups are example of a [MP PMT 2010]  
 (a) Gradualism (b) Cline  
 (c) Gradient of diploidy (d) Polymorphism
17. Who was the scientist to introduce ABO blood groups [CBSE PMT 1993; CPMT 1994; BCECE 2005]  
 (a) Wiener (b) Levine  
 (c) Fisher (d) Landsteiner
18. One of the following is not the types of blood groups or blood factors [KCET 2004]  
 (a) Lewis and Duffy (b) Bufts and Kips  
 (c) ABO and Rh (d) Rh and MN
19. Mating among close relations is referred [CBSE PMT 1994]  
 (a) Permanent marriage (b) Line breeding  
 (c) Inbreeding (d) Cross breeding
20. Genes exhibiting multiple effects phenotype are known as [NCERT; MP PMT 2009; Odisha JEE 2012]  
 (a) Complementary genes (b) Pleiotropic genes  
 (c) Cistrons (d) Pseudogenes
21. For a child having blood group B, if father has blood group A, what may be the blood group of the mother  
 (a) O or A (b) O  
 (c) B or AB (d) A
22. If a child has O type of blood group and the father B type, the genotype of the father will be [CBSE PMT 1992; MP PMT 2009]  
 (a)  $I^O I^O$  (b)  $I^A I^B$   
 (c)  $I^O I^B$  (d)  $I^B I^B$
23. Person with blood group AB is considered as universal recipient because he has [MP PMT 1992, 96, 99, 2003, 06; CPMT 1995; CBSE PMT 2014]  
 (a) No antigen on RBC and no antibody in the plasma  
 (b) Both A and B antigens in the plasma but no antibodies  
 (c) Both A and B antigens on RBC but no antibodies in the plasma  
 (d) Both A and B antibodies in the plasma
24. Rh factor may be responsible for [MP PMT 1992; WB JEE 2010]  
 (a) Turner's syndrome (b) AIDS  
 (c) Sickle-cell anaemia (d) Erythroblastosis foetalis
25. Parents of blood groups O and AB cannot have a child of group AB because [MP PMT 1992]  
 (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
26. Identify the wrong statement [KCET 2015]  
 (a) Alleles b and c also produce sugar  
 (b) Alleles  $I^A$  and  $I^B$  produce sugar  
 (c) When  $I^B$  and b or i are present only  $I^B$  is expressed  
 (d) Both  $I^A$  and  $I^B$  are present together and they express because of co-dominance
27. Which of the following is the number of alleles for blood group in an individual [J & K CET 2012]  
 (a) 1 (b) 2  
 (c) 3 (d) 4
28. Rh factor is named after  
 (a) Man (b) Rat  
 (c) Monkey (d) Chimpanzee
29. A person with antigens A and B and no antibodies belongs to blood group or In which blood group antibodies are absent [CBSE PMT 1991; CPMT 1993, 94; MP PMT 1996, 98, 99, 2011; Odisha JEE 2010]  
 (a) A (b) B  
 (c) AB (d) O
30. 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 these
31. Universal donors have no antigens in RBC and have both a and b antibodies. They belong to blood group [CPMT 1994; JIPMER 1994; MP PMT 1994, 96, 99, 09, 12; Pb PMT 2004]  
 Or  
 Which blood group can be given to patients of any blood group [J & K CET 2005; Odisha JEE 2008; MP PMT 2010]  
 (a) A (b) B  
 (c) AB (d) O
32. Four children belonging to the same parents have the following blood groups A, B, AB and O. Hence, the genotypes of the two parents are [KCET 2011]  
 (a) Both parents are homozygous for 'A' group  
 (b) One parent is homozygous for 'A' and another parent is homozygous for 'B'  
 (c) One parent is heterozygous for 'A' and another parent is heterozygous for 'B'  
 (d) Both parents are homozygous for 'B' group



33. When red blood corpuscles containing both A and B antigens are mixed with your blood serum, they agglutinate. Hence your blood group is ..... type [KCET 2010]  
(a) AB (b) O  
(c) A (d) B
34. Persons of blood group A contain [MP PMT 1994]  
(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
35. Blood group agglutinin is [CPMT 2009]  
(a) Glycoprotein (b) Phosphoprotein  
(c) Haemoprotein (d) Phospholipid
36. The animal which has oval RBCs [Manipal 2005]  
(a) Humans (b) Camel  
(c) Dog (d) Fish
37. Blood groups are named because of the agglutinin A and B present in [CPMT 1992, 93]  
(a) Plasma (b) RBC  
(c) WBC (d) Platelet
38. A person with unknown blood group under ABO system, has suffered much blood loss in an accident and needs immediate blood transfusion. His one friend who has a valid certificate of his own blood type, offers for blood donation without delay. What would have been the type of blood group of the donor friend [NCERT; Odisha JEE 2005; CBSE PMT (Pre.) 2011, 12]  
(a) Type A (b) Type B  
(c) Type AB (d) Type O
39. 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 [MP PMT 1994]  
(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
40. With regard to the ABO blood typing system, if a man who has type B blood and a woman who has type O blood were to have children, what blood types could the children have [KCET 2012]  
(a) A or O (b) B or O  
(c) AB or O (d) A, B, AB or O.
41. If two persons with 'AB' blood group marry and have sufficiently large number of children, these children could be classified as 'A' blood group : 'AB' blood group 'B' blood group in 1 : 2 : 1 ratio. Modern technique of protein electrophoresis reveals presence of both 'A' and 'B' type proteins in 'AB' blood group individuals. This is an example of [NEET 2013]  
(a) Complete dominance (b) Co-dominance  
(c) Incomplete dominance (d) Partial dominance
42. The probability of having a child with blood group O to parents with blood groups A and B is [CPMT 1995]  
(a) 4 out of 4 (b) 3 out of 4  
(c) 2 out of 4 (d) 1 out of 4
43. Example of qualitative inheritance is [CPMT 1995]  
(a) Colour of skin  
(b) Colourblindness  
(c) Klinefelter's syndrome  
(d) Alkaptonuria
44. The father has blood group AB and mother 'O'. The child is supposed to have which of the following bloodgroups [AFMC 1995]  
(a) 'A' or 'B' (b) 'A' only  
(c) 'B' or 'O' (d) B only
45. A child of a mother with blood group A and a father with blood group AB may have any one of the following blood groups except [NCERT; Manipal 1995; CPMT 2005; MHCET 2005]  
(a) A (b) B  
(c) AB (d) O
46. Donors and recipients in a blood transfusion process can be [MP PMT 1995]  
(a) Only father and son  
(b) Only brother and sister  
(c) Only maternal uncle and niece  
(d) All the above
47. Which of the following substances, if introduced into the blood stream, would cause coagulation of blood at the site of its introduction [CBSE PMT 2005]  
(a) Fibrinogen (b) Prothrombin  
(c) Heparin (d) Thromboplastin
48. Detection of blood group is done by agglutination test using antiserum. According to this [KCET 1994]  
(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 these
49. Mother homozygous B, and father is A. What will be the possible blood group in their progeny [DPMT 2007]  
(a) AB & B possible (b) AB & A possible  
(c) A and B possible (d) O possible
50. Rh-ve person donated blood to Rh+ve person for the second time. Then [KCET 2007]  
(a) Rh-ve person will die  
(b) Nothing happens to Rh+ve person  
(c) Rh+ve blood starts reacting to Rh-ve blood  
(d) Rh+ve person will die
51. Rh factor is present in [BHU 2006]  
(a) All vertebrates  
(b) All mammals  
(c) All reptiles  
(d) Man and rhesus monkey only
52. If the foetus is Rh+ and mother is Rh- then [BVP 2004]  
(a) Foetus will transmit antigen to mother blood  
(b) Foetus will transmit antibody to mother blood  
(c) Foetus is attacked by antibodies to mother blood  
(d) Foetus is attacked by antigen to mother blood



In the above given pedigree, assume that no outsider marrying in, carry a disease. Write the genotypes of II and III

[EAMCET 2009]

- (a) All  $X^dY$  (b)  $X^{DY}$  and  $X^{DX^d}$   
 (c)  $X^dXX^dY$  and  $X^dY^D$  (d)  $X^dX^d$  and  $X^dY$
12. Which one of the following is a genetically transmitted character [Kerala PMT 2004]  
 (a) Colour blindness (b) Hydrocephalus  
 (c) Hemophilia (d) Muscular dystrophy  
 (e) All of these
13. A normal-visioned man whose father was colour-blind, marries a woman whose father was also colour-blind. They have their first child as a daughter. What are the chances that this child would be colour-blind [NCERT; CBSE PMT 1990; KCET 2009; CBSE PMT (Pre.) 2012]  
 (a) 50% (b) 100%  
 (c) 0% (d) 25%
14. A man who is suffering from a recessive X-linked disease marries a normal woman. Then what is true about its progeny [CPMT 2009]  
 (a) All sons are diseased  
 (b) All daughter's are diseased  
 (c) All sons are normal  
 (d) None of the above
15. Person whose father is colourblind marries a lady whose mother is daughter of a colourblind man. Their children will be [DPMT 1993; AIIMS 2013]  
 (a) All normal  
 (b) All colour blind  
 (c) All sons colour blind  
 (d) Some sons normal and some colour blind
16. Which one of the following symbols and its representation, used in human pedigree analysis is correct [CBSE PMT (Pre.) 2010]  
 (a)  $\blacklozenge$  = male affected  
 (b)  $\square \text{---} \square$  = mating between relatives  
 (c)  $\circ$  = unaffected male  
 (d)  $\square$  = unaffected female
17. Pick out the correct statements [NEET (Phase-I) 2016]  
 (A) Haemophilia is a sex-linked recessive disease  
 (B) Down's syndrome is due to aneuploidy  
 (C) Phenylketonuria is an autosomal recessive gene disorder  
 (D) Sickle cell anaemia is a X-linked recessive gene disorder  
 (a) (A) and (D) are correct  
 (b) (B) and (D) are correct  
 (c) (A), (C) and (D) are correct  
 (d) (A), (B) and (C) are correct

18. The most common type of haemophilia results from the congenital absence of

[CPMT 2004; MP PMT 2007; WB JEE 2008]

- (a) Factor II (b) Factor V  
 (c) Factor VIII (d) Factor XI

19. Which of the following diseases belongs to the same category as colourblindness in man

[AIEEE Pharmacy 2003; BHU 2005; Odisha JEE 2011]

- (a) Nightblindness (b) Presbyopia  
 (c) Diabetes incipidus (d) Haemophilia

20. X-linked recessive gene is [MP PMT 2007]

- (a) Always expressed in male  
 (b) Always expressed in female  
 (c) Lethal  
 (d) Sub lethal

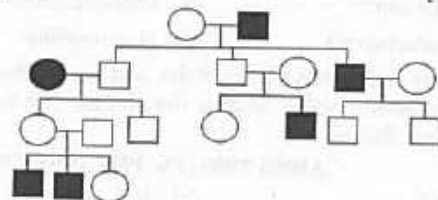
21. If a colourblind woman marries and a normal visioned man, their sons will be [CBSE PMT 1994, 99, 2006; BHU 1996; MP PMT 2000, 05; CPMT 2005]

- (a) Three-fourths colourblind and one-fourth normal  
 (b) All colourblind  
 (c) All normal visioned  
 (d) One-half colourblind and one-half normal

22. A man known to be victim of haemophilia marries a normal woman whose father was known to be a bleeder. Then it is expected that [Pb. PMT 1999; CBSE PMT 2000]

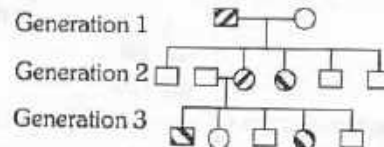
- (a) All their children will be bleeders  
 (b) Half of their children will be bleeders  
 (c) One fourth of their children will be bleeders  
 (d) None of their children will be bleeder

23. In the following human pedigree, the filled symbols represent the affected individuals. Identify the type of given pedigree [AIPMT 2015]



- (a) X-linked recessive (b) Autosomal recessive  
 (c) X-linked dominant (d) Autosomal dominant

24. Given below is a pedigree chart showing the inheritance of a certain sex-linked trait in humans



Key :

- $\square$  Unaffected male  
 $\blacksquare$  Affected male  
 $\circ$  Unaffected female  
 $\bullet$  Affected female

[NCERT; AIIMS 2005]

The trait traced in the above pedigree chart is

- (a) Dominant X-linked (b) Recessive X-linked  
 (c) Dominant Y-linked (d) Recessive Y-linked

25. Which of the following statement about colour blindness is correct [AMU (Med.) 2010]  
 (a) 2% men are red colour blind, 6% are green colour blind  
 (b) 6% men are red colour blind, 2% are green colour blind  
 (c) 10% men are red colour blind, 5% are green colour blind  
 (d) 5% men are red colour blind, 10% are green colour blind
26. Sex linked disease is [NCERT; CPMT 1993, 99, 2003; MP PMT 1994, 98; AFMC 1996, 2001; Pb. PMT 2000; MHCET 2000; J & K CET 2002; BHU 2004; Kerala PMT 2010; WB-JEE 2016]  
 (a) Haemophilia (b) Colourblindness  
 (c) Sickle-cell anaemia (d) Both (a) and (b)
27. A woman with normal vision, but whose father was colour blind, marries a colour blind man. Suppose that the fourth child of this couple was a boy. Thus boy [CBSE PMT 2005]  
 (a) Will be partially colour blind since he is heterozygous for the colour blind mutant allele  
 (b) Must have normal colour vision  
 (c) Must be colour blind  
 (d) May be colour blind or may be of normal vision,
28. A man and a woman, who do not show any apparent signs of a certain inherited disease, have seven children (2 daughters and 5 sons). Three of the sons suffer from the given disease but none of the daughters are affected which of the following mode of inheritance do you suggest for this disease [CBSE PMT 2005]  
 (a) Autosomal dominant (b) Sex-linked dominant  
 (c) Sex-limited recessive (d) Sex-linked recessive
29. Which of the following is not a hereditary disease [CBSE PMT 2005]  
 (a) Cretinism (b) Cystic fibrosis  
 (c) Thalassaemia (d) Haemophilia
30. 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 [AIIMS 1999; Pb. PMT 1999; CPMT 2000]  
 (a) 25% (b) 50%  
 (c) 75% (d) 100%
31. A colour blind man marries a woman with normal sight who has no history of colour blindness in her family. What is the probability of their grandson being colour blind [AIPMT 2015]  
 (a) 1 (b) Nil  
 (c) 0.25 (d) 0.5
32. Sickle cell anaemia is due to [CBSE PMT 1990]  
 (a) Hormones (b) Viruses  
 (c) Genes (d) Bacteria
33. Sex linked inheritance was discovered by  
 (a) McClung (b) Mendel  
 (c) Landsteiner (d) Morgan
34. Colour blindness is caused by a single [RPMT 2006]  
 (a) Dominant gene in woman (b) Dominant gene in man  
 (c) Recessive gene in man (d) Recessive gene in woman
35. The following is a pedigree chart of a family with five children. It shows the inheritance of attached, ear - lobes as opposed to the free ones. The squares represent the male and circles the female individuals [NCERT; AIIMS 2004]
- 
- Which one of the following conclusions drawn is correct  
 (a) The parents are homozygous dominant  
 (b) The parents are homozygous recessive  
 (c) The parents are heterozygous  
 (d) The trait is Y - linked
36. Female rarely experience the physiologic defect of haemophilia because they do so only when they are  
 (a) Heterozygous for the defect  
 (b) Homozygous for the defect  
 (c) Carrier for the defect  
 (d) Wives of haemophilic husbands
37. A colour blind son will born when [CPMT 1993]  
 (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
38. Sex influenced characters are due to [MH CET 2004]  
 (a) Y-linked genes  
 (b) X-linked genes  
 (c) Autosomal genes  
 (d) Y-linked gene modification
39. A colourblind man has a colourblind sister but a normal brother than phenotype of its parents is [CPMT 1993]  
 (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
40. The frequency of a character is found to be increasing when [CPMT 1993; MP PMT 2001]  
 (a) It is dominant  
 (b) It is recessive  
 (c) It is adaptable  
 (d) It is inheritable
41. 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
42. The daughter born to haemophilic father and normal mother could be [AIIMS 1992]  
 (a) Normal (b) Carrier  
 (c) Haemophilic (d) None
43. Haemophilia is caused due to lack of [AIIMS 1992]  
 (a) ADH (b) AHF  
 (c) STH (d) ACTH



44. A marriage between normal visioned man and colourblind woman will produce which of the following types of offsprings [BHU 2004]  
 (a) Normal sons and carrier daughters  
 (b) Colourblind sons and carrier daughters  
 (c) Colourblind sons and 50% carrier daughters  
 (d) 50% colourblind sons and 50% carrier daughters
45. Sex-linked genes of man are [Wardha 2005]  
 (a) Present on X-chromosome  
 (b) Present on autosomes  
 (c) Present on short arm (p) of Y-chromosome  
 (d) Present on long arm (q) of Y-chromosome
46. If a normal woman marries a colourblind man, their [MP PMT 2002; Kerala PMT 2004; RPMT 2006]  
 (a) All sons 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
47. A girl of normal vision whose father was colourblind marries a man of normal vision whose father was also colourblind. Their sons would be (of total number of sons) [MP PMT 1995]  
 (a) All colourblind  
 (b) 50% colourblind  
 (c) All normal  
 (d) 25% colourblind
48. All the sons are haemophilic and daughter are normal of a haemophilic father and normal mother. This character is [CBSE PMT 1996]  
 (a) X-linked recessive  
 (b) Y-linked recessive  
 (c) X-linked dominant  
 (d) Y-linked dominant
49. 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)
50. Haemophilia is more commonly seen in human males than in human females because [NCERT; CBSE PMT 2005; WB JEE 2008; NEET (Phase-I) 2016]  
 (a) This disease is due to an X-linked dominant mutation  
 (b) A greater proportion of girls die in infancy  
 (c) This disease is due to an X-linked recessive mutation  
 (d) This disease is due to a Y-linked recessive mutation
51. 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 [Kerala CET 2003]  
 (a) All sons are normal and all daughters are colourblind  
 (b) Both the sons and daughters are colourblind  
 (c) All the sons are colourblind and all daughters are normal  
 (d) 50% sons are colourblind and all daughters are phenotypically normal

52. Match the symbol with associated statement

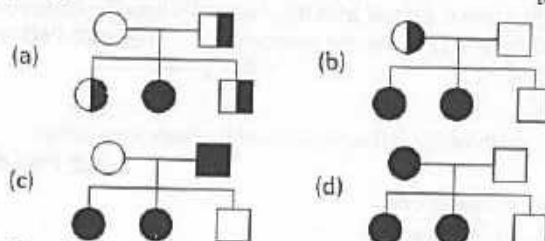
1.	2.
3.	4.
5.	6.
7.	8.
9.	10.
11.	12.
13.	14.
15.	16.

- A. Heterozygous individuals with autosomal recessive  
 B. Diseased (or death)  
 C. Female carrier of an X-linked recessive gene  
 D. Individuals with normal trait  
 E. Consanguineous mating (marriage of blood relatives)  
 F. Unknown sex  
 G. Mating  
 H. Male  
 I. Female  
 J. Affected individual  
 K. Abortion or still birth

[NCERT]

	B	C	E	F
(a)	6	4	14	12
(b)	16	13	2	11
(c)	3	1	2	7
(d)	16	1	2	7

53. If husband is PTC taster and wife is PTC non taster. Their daughters are non tasters but their son is taster. This is not related with a sex-linked trait. Out of four a, b, c, d which pedigree is correct [NCERT]



54. 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

55. 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; CBSE PMT 2004; MH CET 2004; AIIMS 2008]

(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

56. A colourblind daughter is born when

[MP PMT 1998; Kerala CET 2002; Odisha JEE 2005; WB-JEE 2016]

(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

57. Brachydactyly is due to

[RPMT 2006]

(a) Dominant gene on the autosome  
(b) Recessive gene on the autosome  
(c) Dominant gene on the sex chromosome  
(d) None of the above

58. Which disease is genetically linked [MP PMT 1996, 99]

(a) Haemophilia (b) Dysentery  
(c) Plague (d) Tuberculosis

59. Haemophilic man marries a normal woman. Their offsprings will be [MP PMT 1993, 97; CBSE PMT 1999; CPMT 1999; Haryana PMT 2005]

(a) All girls (b) All normal  
(c) All haemophilic (d) All boys haemophilic

60. When an allele fails to explain itself in presence of the other allele, the former is said to be [CBSE PMT 1991]

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

61. A woman with two genes for haemophilia and one gene for colourblindness on one of the X chromosomes marries a normal man. How will the progeny be [NCERT; CBSE PMT 1998; KCET 2012]

(a) All sons and daughters haemophilic and colourblind  
(b) Haemophilic and colourblind daughters  
(c) 50% haemophilic colourblind sons and 50% normal sons  
(d) 50% haemophilic daughters and 50% colourblind daughters

62. A fruit fly is heterozygous for sex-linked genes when mated 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

63. In which of the following colourblindness is inherited

[MP PMT 2000]

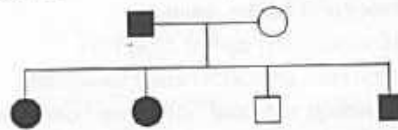
(a) In males only  
(b) In females only  
(c) In both males and females  
(d) In none of the above

64. 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

65. Study the pedigree chart of a certain family given below and select the correct conclusion which can be drawn for the character [NCERT; CBSE PMT (Mains) 2010]



(a) The female parent is heterozygous  
(b) The parents could not have a normal daughter for this character  
(c) The trait under study could not be colour-blindness  
(d) The male parent is homozygous dominant

66. 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

67. Gene for colourblindness is located on

[MH CET 2002; MP PMT 2003, 07; Odisha JEE 2011]

(a) Homologous part of X-chromosome  
(b) Non-homologous part of X-chromosome  
(c) Homologous part of Y-chromosome  
(d) Non-homologous part of Y-chromosome

68. Which of the following conditions is not X-linked

[MP PMT 2010]

(a) Colour blindness (b) Haemophilia  
(c) Down's syndrome (d) Myopia

69. 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; BVP 2002]

(a) Autosomal dominant (b) Sex linked recessive  
(c) Sex limited character (d) Sex linked dominant

70. Pattern baldness, moustaches and beard in human males are examples of [CBSE PMT 2003]

Or

The traits which are expressed in only a particular sex though their genes occurs in the opposite sex too are known as [Odisha JEE 2012]

(a) Sex-determining traits (b) Sex linked traits  
(c) Sex limited traits (d) Sex differentiating traits

71. One of the genes present exclusively on the X-chromosome in humans is concerned with [AIIMS 2003]

(a) Baldness  
(b) Red-green colour blindness  
(c) Facial hair/moustaches in males  
(d) Night blindness

72. One of the following is not true to haemophilia

[Kerala CET 2003]

(a) Royal disease (b) Bleeder's disease  
(c) X-linked disease (d) Y-linked disease

73. Which one is the **incorrect** statement with regards to the importance of pedigree analysis [NEET (Karnataka) 2013]  
 (a) It confirms that DNA is the carrier of genetic information  
 (b) It helps to understand whether the trait in question is dominant or recessive  
 (c) It confirms that the trait is linked to one of the autosome  
 (d) It helps to trace the inheritance of a specific trait
74. If a colour-blind man marries a woman who is homozygous for normal colour vision, the probability of their son being colour-blind is [NEET (Phase-II) 2016]  
 (a) 1 (b) 0  
 (c) 0.5 (d) 0.75
9. 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
10. Twin is [Haryana PMT 2005]  
 (a) Developed from same zygote  
 (b) Developed from different zygote  
 (c) Two different sperm  
 (d) Two different ovum
11. 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)

### Twins & I.Q., Eugenics, Euthenics and Euphenics

1. Fraternal twins are produced when [CMC Vellore 1993]  
 (a) A fertilized egg divided into two  
 (b) An egg is fertilized by two sperms  
 (c) A divided egg has two set of chromosomes  
 (d) Two eggs are fertilized simultaneously
2. Identical twins are [Odisha JEE 2010]  
 (a) Heterozygous (b) Homozygous  
 (c) Monozygotic (d) Dizygotic
3. An organism which receives identical alleles of a particular gene from both parents is [CBSE PMT 1993]  
 (a) Heterozygote (b) Holometabolous  
 (c) Homosapiens (d) Homozygote
4. Study of improvement of human race by providing ideal nature is [CBSE PMT 1990; MP PMT 1998]  
**Or**  
 Improvement of genetic characters and present day generation on the basis of best nutrition and training is called [MP PMT 1995]  
 (a) Eugenics (b) Euphenics  
 (c) Euthenics (d) None of these
5. The best method to improve the genetic quality of mankind is  
 (a) Marriage restrictions  
 (b) Sterilizations  
 (c) Control of immigrations  
 (d) Sexual separation of defectives
6. Study of human race is called [AFMC 1997; Haryana PMT 2005; MP PMT 2005]  
 (a) Eugenics (b) Entomology  
 (c) Ecology (d) Pathology
7. 'Eugenics' pertains to [CBSE PMT 1990]  
 (a) Improvement of mankind by improving his heredity  
 (b) Preserving human sperms for artificial insemination  
 (c) Study of human genetics  
 (d) Controlling size of a human family
8. Sometimes the separation of twins is incomplete and these are born attached or remain so even after. Such twins are known as [MH CET 2002]  
**Or**  
 Conjoint twins are also known as  
 (a) Fraternal (b) Dizygotic  
 (c) Identical (d) Siamese

## NCERT

### Exemplar Questions

1. All genes located on the same chromosome [NCERT]  
 (a) Form different groups depending upon their relative distance  
 (b) Form one linkage group  
 (c) Will not form any linkage groups  
 (d) Form interactive groups that affect the phenotype
2. Conditions of a karyotype  $2n + 1$ ,  $2n - 1$  and  $2n + 2$ ,  $2n - 2$  are called [NCERT]  
 (a) Aneuploidy (b) Polyploidy  
 (c) Allopolyploidy (d) Monosomy
3. Distance between the genes and percentage of recombination shows [NCERT]  
 (a) A direct relationship (b) An inverse relationship  
 (c) A parallel relationship (d) No relationship
4. If a genetic disease is transferred from a phenotypically normal but carrier female to only some of the male progeny, the disease is [NCERT]  
 (a) Autosomal dominant (b) Autosomal recessive  
 (c) Sex-linked dominant (d) Sex-linked recessive
5. In sickle cell anaemia glutamic acid is replaced by valine. Which one of the following triplets codes for valine [NCERT]  
 (a) G G G (b) A A G  
 (c) G A A (d) G U G
6. Person having genotype  $I^A I^B$  would show the blood group as AB. This is because of [NCERT]  
 (a) Pleiotropy (b) Co-dominance  
 (c) Segregation (d) Incomplete dominance
7. Z Z / Z W type of sex determination is seen in [NCERT]  
 (a) Platypus (b) Snails  
 (c) Cockroach (d) Peacock
8. A cross between two tall plants resulted in offspring having few dwarf plants. What would be the genotypes of both the parents [NCERT]  
 (a) TT and Tt (b) Tt and Tt  
 (c) TT and TT (d) Tt and tt



9. In a dihybrid cross, if you get 9:3:3:1 ratio it denotes that [NCERT]  
 (a) The alleles of two genes are interacting with each other  
 (b) It is a multigenic inheritance  
 (c) It is a case of multiple allelism  
 (d) The alleles of two genes are segregating independently
10. Which of the following will not result in variations among siblings [NCERT]  
 (a) Independent assortment of genes  
 (b) Crossing over  
 (c) Linkage  
 (d) Mutation
11. Mendel's Law of independent assortment holds good for genes situated on the [NCERT]  
 (a) Non-homologous chromosomes  
 (b) Homologous chromosomes  
 (c) Extra nuclear genetic element  
 (d) Same chromosome
12. Occasionally, a single gene may express more than one effect. The Phenomenon is called [NCERT]  
 (a) Multiple allelism (b) Mosaicism  
 (c) Pleiotropy (d) Polygeny
13. In a certain taxon of insects some have 17 chromosomes and the others have 18 chromosomes. The 17 and 18 chromosome-bearing organisms are [NCERT]  
 (a) Males and females, respectively  
 (b) Females and males, respectively  
 (c) All males  
 (d) All females
14. The inheritance pattern of a gene over generations among humans is studied by the pedigree analysis. Character studied in the pedigree analysis is equivalent to [NCERT]  
 (a) Quantitative trait (b) Mendelian trait  
 (c) Polygenic trait (d) Maternal trait
15. It is said that Mendel proposed that the factor controlling any character is discrete and independent. His proposition was based on the [NCERT]  
 (a) Results of  $F_3$  generation of a cross  
 (b) Observations that the offspring of a cross made between the plants having two contrasting characters shows only one character without any blending  
 (c) Self pollination of  $F_1$  offsprings  
 (d) Cross pollination of  $F_1$  generation with recessive parent
16. Two genes 'A' and 'B' are linked. In a dihybrid cross involving these two genes, the  $F_1$  heterozygote is crossed with homozygous recessive parental type (aa bb). What would be the ratio of offspring in the next generation [NCERT]  
 (a) 1:1:1:1 (b) 9:3:3:1  
 (c) 3:1 (d) 1:1
17. In the  $F_2$  generation of a Mendelian dihybrid cross the number of phenotypes and genotypes are [NCERT]  
 (a) Phenotypes – 4; genotypes – 16  
 (b) Phenotypes – 9; genotypes – 4  
 (c) Phenotypes – 4; genotypes – 8  
 (d) Phenotypes – 4; genotypes – 9
18. Mother and father of a person with 'O' blood group have 'A' and 'B' blood group, respectively. What would be the genotype of both mother and father [NCERT]  
 (a) Mother is homozygous for 'A' blood group and father is heterozygous for 'B'  
 (b) Mother is heterozygous for 'A' blood group and father is homozygous for 'B'  
 (c) Both mother and father are heterozygous for 'A' and 'B' blood group, respectively  
 (d) Both mother and father are homozygous for 'A' and 'B' blood group, respectively



## Critical Thinking

### Objective Questions

1. Which is Gynandromorph type of animal [GUJCET 2015]  
 (a) Drosophila (b) Beetles  
 (c) Silk worms (d) All of the above
2. The segregation of paired hereditary factors that Mendel postulated occurs during [CBSE PMT 1993]  
 (a) Anaphase of first meiotic division  
 (b) Metaphase of second meiotic division  
 (c) During interphase between two meiotic divisions  
 (d) Prophase of first meiotic division
3. A cell at telophase stage is observed by a student in a plant brought from the field. He tells his teacher that this cell is not like other cells at telophase stage. There is no formation of cell plate and thus the cell is containing more number of chromosomes as compared to other dividing cells. This would result in [NEET (Phase-I) 2016]  
 (a) Aneuploidy (b) Polyploidy  
 (c) Somaclonal variation (d) Polyteny
4. Match the terms in column-I with their description in column-II and choose the correct option [NEET (Phase-I) 2016]

	Column I		Column II
(a)	Dominance	(i)	Many genes govern a single character
(b)	Codominance	(ii)	In a heterozygous organism only one allele expresses itself
(c)	Pleiotropy	(iii)	In a heterozygous organism both alleles express themselves fully
(d)	Polygenic inheritance	(iv)	A single gene influences many characters

- |     |      |       |      |       |
|-----|------|-------|------|-------|
|     | (a)  | (b)   | (c)  | (d)   |
| (a) | (ii) | (i)   | (iv) | (iii) |
| (b) | (ii) | (iii) | (iv) | (i)   |
| (c) | (iv) | (i)   | (ii) | (iii) |
| (d) | (iv) | (iii) | (i)  | (ii)  |

1. A selection that acts to eliminate one extreme from an array of phenotypes is [EAMCET 2009]

(a) Disruptive (b) Directional  
(c) Stabilizing (d) Coevolution

2. A tobacco plant which is heterozygous for albinism (a recessive character) is self pollinated if 1200 seeds are subsequently germinated, how many of the seedlings would have the parental genotype [Manipal 1995; BHU 2008]

(a) 300 (b) 600  
(c) 900 (d) 1200

3. Match the column I, II, and III

Column I		Column II		Column III	
(A)	Sickle Cell Anaemia	(i)	Due to recessive PP genes	(P)	Arrangement of valine in place of Glutamic acid
(B)	Phenyl Ketonuria	(ii)	Due to absence of homogentisic oxidase enzyme	(Q)	Inborn error of metabolism
(C)	Alkaptonuria	(iii)	Follows Mendelian Principles	(R)	Urine turns black when exposed to air
(D)	Thalassaemia	(iv)	Characters caused by homozygous recessive genes	(S)	The required haemoglobin is not generated in the blood

[GUJCET 2015]

- (a) (A - ii - S) (B - iii - R) (C - i - Q) (D - iv - P)  
(b) (A - iv - P) (B - i - Q) (C - ii - R) (D - iii - S)  
(c) (A - iv - P) (B - iii - R) (C - i - S) (D - ii - R)  
(d) (A - iii - R) (B - i - Q) (C - iv - P) (D - ii - S)

4. Find the odd one out, with respect to X-linkage

[MHCET 2015]

(a) Haemophilia (b) Myopia  
(c) Nephritis (d) Night blindness

5. In case of incomplete dominance in  $F_2$  generation

[BHU 1995, 2008]

- (a) Genotypic ratio is 3 : 1  
(b) Phenotypic ratio is 3 : 1  
(c) Genotypic ratio = phenotypic ratio  
(d) Nothing can be concluded

6. One of the parents of a cross has a mutation in its mitochondria. In that cross, that parent is taken as a male. During segregation of  $F_2$  progenies that mutation is found in

[CBSE PMT 2004]

- (a) All the progenies  
(b) Fifty percent of the progenies  
(c) One-third of the progenies  
(d) None of the progenies

7. When a cell with 40 chromosomes undergoes meiosis, each of the four resulting cells has [BHU 2001; CPMT 2003]

(a) 20 chromosomes (b) 40 chromosomes  
(c) 80 chromosomes (d) 10 chromosomes

8. Chromosomal number in a cell of a flowering plant is

(a) Only haploid (b) Only diploid  
(c) Many types (d) None of these

9. How many genome types are present in a typical green plants cell [CBSE PMT 1998]

(a) Two (b) Three  
(c) More than five (d) More than ten

10. Find out the correct statement [Kerala PMT 2007]

- (a) Monosomy and nullisomy are the two types of euploidy  
(b) Polyploidy is more common in animals than in plants  
(c) Polyploids occur due to the failure in complete separation of sets of chromosomes  
(d)  $2n-1$  condition results in trisomy  
(e) Non-homologous chromosomal duplication results in autopolyploidy

11. Match the items in Column - I with Column - II and choose the correct alternative

Column - I		Column - II	
A.	Sickle-cell anaemia	1.	7 <sup>th</sup> chromosome
B.	Phenylketonuria	2.	4 <sup>th</sup> chromosome
C.	Cystic fibrosis	3.	11 <sup>th</sup> chromosome
D.	Huntington's disease	4.	X-chromosome
E.	Color blindness	5.	12 <sup>th</sup> chromosome

[NCERT; Kerala PMT 2006, 07, 08; VITEEE 2006; AMU (Med.) 2009]

- (a) A-1, B-3, C-4, D-2, E-5  
(b) A-2, B-3, C-4, D-5, E-1  
(c) A-2, B-1, C-3, D-5, E-4  
(d) A-4, B-5, C-3, D-2, E-1  
(e) A-3, B-5, C-1, D-2, E-4

12. Which of the following statement is correct [GUJCET 2014]

- (a) In honey - bee, functional male does not undergo meiosis during gamatic formation  
(b) In flagellaria, male is heterogametic  
(c) In Bonellia, a hormone - like substance secreted by the proboscis is responsible for femaleness  
(d) Due to the addition of one extra 'X' chromosome in Drosophila in uninucleated state gynandromorphy is observed

13. If an inheritable mutation is observed in a population at high frequency, it is referred to as [KCET 2015]

(a) Sequence annotation (b) DNA polymorphism  
(c) Linkage (d) Expressed sequence Tag

14. The cause of Cat-cry syndrome is due to [RPMT 2006]

(a) Loss of a segment of X-chromosome  
(b) Loss of a segment of 5<sup>th</sup> chromosome  
(c) Loss of segment of Y-chromosome  
(d) None of the above

15. When two genes are situated very close to each other in a chromosome

(a) The percentage of crossing over between them is very high  
(b) Hardly any cross over are detected  
(c) No crossing over can take place between them  
(d) Only double cross overs can take place between them

20. Who is known as father of physiological genetics or father of biochemical genetics [Haryana PMT 2001; MHCET 2001; AIIMS 2009]
- (a) Slatyer (b) Charles Elton  
(c) Taylors (d) Archibald Garrod
21. When a cluster of genes shows linkage behaviour they [CBSE PMT 2003]
- (a) Induce cell division  
(b) Do not show a chromosome map  
(c) Show recombination during meiosis  
(d) Do not show independent assortment
22. Which of the following best illustrates FEEDBACK in development [NEET (Karnataka) 2013]
- (a) Tissue (X) secretes RNA which changes the development of tissue (Y)  
(b) As tissue (X) develops, it secretes enzymes that inhibit the development of tissue (Y)  
(c) As tissue (X) develops, it secretes something that induces tissue (Y) to develop  
(d) As tissues (X) develops, it secretes something that shows down the growth of tissue (Y)
23. Match the column I with column II and choose the correct option
- | Column I                                   | Column II                 |
|--------------------------------------------|---------------------------|
| A. Incomplete dominance                    | i. Hershey and Chase      |
| B. Linkage                                 | ii. <i>Antirrhinum sp</i> |
| C. Transforming principle                  | iii. Griffith             |
| D. Proved that DNA is the Genetic material | iv. Morgan                |
- [Kerala PMT 2012]
- (a) A-i; B-iv; C-iii; D-ii (b) A-iv; B-ii; C-iii; D-i  
(c) A-ii; B-iii; C-iv; D-i (d) A-ii; B-iv; C-i; D-iii  
(e) A-ii; B-iv; C-iii; D-i
24. The fruit fly *Drosophila melanogaster* was found to be very suitable for experimental verification of chromosomal theory of inheritance by Morgan and his colleagues because [CBSE PMT (Mains) 2010]
- (a) It reproduces parthenogenetically  
(b) A single mating produces two young flies  
(c) Smaller female is easily recognisable from larger male  
(d) It completes life cycle in about two weeks
25. The exchange of one part of a chromosome to the other part of same or another chromosome is called [AFMC 2002]
- Or
- The movement of gene from one linkage group to another is called [AIPMT 2015; AIPMT (Cancelled) 2015; NEET (Phase-II) 2016]
- (a) Inversion (b) Mutation  
(c) Translocation (d) Linkage
26. 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
27. How many nucleosomes are found in helical coil of 30 nm chromatin fibre [RPMT 2000]
- (a) 10 (b) 12  
(c) 06 (d) 09
28. Biological marriage of one of the following should be avoided [AFMC 1995, 2001]
- 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 [CBSE PMT 1990, 2002; MP PMT 1995, 2000, 03, 06; AIEEE Pharmacy 2003; RPMT 2005; AFMC 2006; BHU 2008]
- Or
- In which of the following situations, is there a risk factor for children of incurring erythroblastosis foetalis [KCET 2010]
- (a)  $Rh^+$  male and  $Rh^-$  female  
(b)  $Rh^+$  male and  $Rh^+$  female  
(c)  $Rh^-$  male and  $Rh^+$  female  
(d)  $Rh^-$  male and  $Rh^-$  female
29. Primary source of allelic variation is [AIIMS 2005]
- (a) Independent assortment (b) Recombination  
(c) Mutation (d) Polyploidy
30. Persons with the following syndrome have a tendency of tall structure, mental defects and a strong antisocial behaviour [KCET 1994]
- (a) XYY syndrome  
(b) Down's syndrome  
(c) Klinefelter's syndrome  
(d) Turner's syndrome
31. Recessive characters are expressed [AFMC 1995]
- (a) Only when they are present on X chromosomes of male  
(b) Only when they are present on X chromosomes of female  
(c) On any autosome  
(d) On both the chromosomes of female
32. Marriages between close relatives and cousins is not advisable because
- (a) More mutations 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
33. 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 of 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
34. In order to find out the different types of gametes produced by a pea plant having the genotype AaBb, it should be crossed to a plant with the genotype [CBSE PMT 2005]
- (a) aaBB (b) AaBb  
(c) AABB (d) aabb



35. Each chromosome at the anaphase stage of a bone marrow cell in our body has [CBSE PMT 1995]  
(a) Two chromatids (b) No chromatids  
(c) Only one chromatid (d) Several chromatids
36. A child's blood group is 'O'. The parents blood groups cannot be [CBSE PMT 1994; Kerala PMT 2005]  
(a) AB and O (b) B and O  
(c) A and B (d) A and A
37. Identical twins are produced when [MP PMT 2001]  
(a) One fertilized egg divided into 2 blastomeres and both separate  
(b) One sperm fertilizes two eggs  
(c) One egg fertilized with two sperms  
(d) Two eggs are fertilized
38. The process of genetic mutation is [MP PMT 2002]  
(a) Reversible (b) Irreversible  
(c) Partially reversible (d) Continuous
39. Allelic sequence variations where more than one variant (allele) at a locus in a human population with a frequency greater than 0.01 is referred to as [Kerala PMT 2011]  
(a) Incomplete dominance (b) Multiple allelism  
(c) SNP (d) EST  
(e) DNA polymorphism
40. A man with blood group 'AB' marries a woman with 'O' blood group. In this situation [MP PMT 1994]  
(a) The blood groups 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
41. Knowing that albinism is determined by a recessive gene in man, presence of albinism in children born to a couple proves that [AIIMS 1992]  
(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
42. 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 the true person  
(c) Both the parentage claims are false  
(d) Mother is the true person and father claimed is not true
43. 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
44. 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 [MP PMT 2001]  
(a) 6 : 2 (b) 2 : 6  
(c) 4 : 4 (d) 3 : 5
45. Transition type of gene mutation is caused when [MP PMT 1997]  
(a) GC is replaced by TA (b) CG is replaced by GC  
(c) AT is replaced by CG (d) AT is replaced by GC
46. Euploidy is best explained by [Pb. PMT 1999]  
(a) Exact multiples of a haploid set of chromosomes  
(b) One chromosome less than the haploid set of chromosomes  
(c) One chromosome more than the haploid set of chromosomes  
(d) One chromosome more than the diploid set of chromosomes
47. Match list I with List II and select the correct answer using code given below  
List I (syndrome)  
(1) Patau's syndrome (2) Kline-Felter'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<sup>th</sup>  
(D)  $46 + 1 = 47$ , Chromosome 21<sup>st</sup>  
Code [MP PMT 2001]  
(a) 1 2 3 4 (b) 1 2 3 4  
(c) A B C D (d) D C B A  
(e) 1 2 3 4 (f) 1 2 3 4  
(g) C B D A (h) C A D B

## Assertion & Reason

Read the assertion and reason carefully to mark the correct option out of the options given below :

- (a) If both the assertion and the reason are true and the reason is a correct explanation of the assertion  
(b) If both the assertion and reason are true but the reason is not a correct explanation of the assertion  
(c) If the assertion is true but the reason is false  
(d) If both the assertion and reason are false  
(e) If the assertion is false but reason is true

1. Assertion : Somaclonal variations may be present in plants produced from callus.

Reason : Somaclonal variations are caused due to recombination during meiosis.

[EAMCET 2009]

2. Assertion : In humans, the gamete contributed by the male determines whether the child produced will be male or female.

Reason : Sex in humans is a polygenic trait depending upon a cumulative effect of some genes on X-chromosome and some on Y-chromosome. [AIIMS 2005, 08]

3. Assertion : Persons suffering from haemophilia fail to produce blood clotting factor VIII.  
Reason : Prothrombin producing platelets in such persons are found in very low concentration. [AIIMS 2005]
4. Assertion : Mustard gas acts as a mutagen.  
Reason : It transfers alkyl groups to the bases in DNA.
5. Assertion : The DNA fingerprint is the same for every cell, tissue and organ of a person.  
Reason : DNA fingerprint is used for treatment of inherited disorders like Huntington's disease, Alzheimer's and Sickle cell anaemia. [GUJCET 2015]
6. Assertion : Among the primates, chimpanzee is the closest relative of the present day humans.  
Reason : The banding pattern in the autosome numbers 3 and 6 of man and chimpanzee is remarkably similar. [AIIMS 2004]
7. Assertion : If pollen mother cells has 42 chromosomes. The pollen has only 21 chromosomes.  
Reason : Pollens are formed after meiosis in pollen mother cell. [AIIMS 1997]
8. Assertion : Clones are produced by sexual reproduction and same sexual process.  
Reason : These are prepared by group of cells descended from many cells or by inbreeding of a heterozygous line. [AIIMS 2002]
9. Assertion : Hybrids are generally back crossed.  
Reason : Back cross is done to increase the traits of the parent.
10. Assertion : A gene may have several allelomorphs.  
Reason : Wild form can mutate in more than one ways.
11. Assertion : Phenylketonuria is a recessive hereditary disease caused by body's failure to oxidise an amino acid phenylalanine to tyrosine, because of a defective enzyme.  
Reason : It results the presence of phenylalanine acid in urine. [AIIMS 2000]
12. Assertion : The genetic complement of an organism is called genotype.  
Reason : Genotype is the type of hereditary properties of an organism. [AIIMS 1999, 2007]
13. Assertion : Holandric genes are found on Y chromosome.  
Reason : Inheritance of Holandric genes are always from father to son. [AIIMS 1996]
14. Assertion : Haemophilia never occurs in women.  
Reason : Gene for haemophilia is located on X chromosome. [AIIMS 1994, 96]
15. Assertion : Haploids are used to study mutation.  
Reason : Most of the mutations are recessive.
16. Assertion : The shape of chromosomes is based on the position of centromere.  
Reason : During anaphase, the chromosome bends in the region of centromere.
17. Assertion : Heterochromatin is genetically inactive.  
Reason : It lacks genes.
18. Assertion : Kinetochore helps in the movement of chromosomes.  
Reason : It has points for attachment of microtubules.
19. Assertion : Restriction endonuclease recognize short palindromic sequence and cut at specific sites.  
Reason : When a restriction endonuclease acts on Palindrome, it cleaves both the strands of DNA molecules. [GUJCET 2015]
20. Assertion : The lampbrush chromosomes are called diplotene chromosomes bivalents.  
Reason : The number of loops is maximum during diplotene.
21. Assertion : In humans, most sex-linked genes are present on the X chromosome.  
Reason : X-chromosome contains a large number of genes with major effects on phenotype.
22. Assertion : Human chromosomes have been studied through banding technique.  
Reason : Banding technique is useful in studying chromosomal aberrations.

## Answers

### Mendelism

1	a	2	c	3	c	4	b	5	c
6	d	7	b	8	b	9	d	10	b
11	a	12	a	13	b	14	a	15	d
16	d	17	c	18	c	19	b	20	b
21	c	22	c	23	b	24	a	25	a
26	a	27	b	28	b	29	a	30	a
31	c	32	d	33	a	34	b	35	a
36	b	37	d	38	b	39	a	40	b
41	d	42	c	43	b	44	b	45	a
46	b	47	b	48	c	49	a	50	a
51	d	52	c	53	a	54	a	55	d
56	d	57	a	58	c	59	c	60	b
61	a	62	d	63	d	64	c	65	d
66	a	67	b	68	a	69	b	70	a
71	c	72	b	73	c	74	b	75	c
76	c	77	b	78	c	79	c	80	a
81	c	82	d	83	c	84	b	85	d
86	a	87	c	88	b	89	b	90	c
91	a	92	c	93	b	94	c	95	d
96	c	97	b	98	a	99	a	100	d

53. A woman is married for the second time. Her first husband was ABO blood type A, and her child by that marriage was type O. Her new husband is type B and their child is type AB. What is the woman's ABO genotype and blood type  
[AIIMS 2009]  
(a)  $I^A I^O$ ; Blood type A (b)  $I^A I^B$ ; Blood type AB  
(c)  $I^B I^O$ ; Blood type B (d)  $I^O I^O$ ; Blood type O
54. Who discovered Rh factor [MP PMT 1998]  
(a) Huxley (b) Landsteiner  
(c) Landsteiner and Weiner (d) Weiner
55. If one parent has blood group A and the other parent has blood group B, the offsprings have which blood group  
[MP PMT 1998; AIPMT (Cancelled) 2015]  
(a) AB (b) O  
(c) BO (d) A, B, AB, O
56. The problem due to Rh<sup>+</sup> factor arises when the blood two ( $Rh^+$  and  $Rh^-$ ) mix up [CBSE PMT 1999]  
(a) In a test tube (b) Through transfusion  
(c) During pregnancy (d) Both (a) and (c)
57. Which of the following are most abundant types of antibodies [CBSE PMT 1999; VITEEE 2008; Odisha JEE 2011; BHU 2012]  
(a) IgA (b) IgE  
(c) IgG (d) IgM
58. When dominant and recessive alleles express themselves together, it is called [CBSE PMT 2001]  
(a) Dominance (b) Co-dominance  
(c) Amphidominance (d) Pseudodominance
59. In erythroblastosis foetalis, which factors of the mother pass through placenta into the foetus [JIPMER 2002]  
(a) Rh antigens (b) Rh antibodies  
(c) ABO antibodies (d) Agglutinins
60. Which one of the following is hereditary character of blood [AFMC 2003]  
(a) Blood group (b) Haem  
(c) Nucleus (d) None of the above
61. You are required to draw blood from a patient and to keep it in a test tube for analysis of blood corpuscles and plasma. You are also provided with the following four types of test tubes. Which of them will you **not** use for the purpose.  
[CBSE PMT 2004]  
(a) Test tube containing heparin  
(b) Test tube containing sodium oxalate  
(c) Test tube containing calcium bicarbonate  
(d) Chilled test tube
62. A man with blood group B marries a woman with blood group A and their first child is having blood group B. What is the genotype of child [CPMT 2004]  
(a)  $I^a I^b$  (b)  $I^a I^a$   
(c)  $I^b I^a$  (d)  $I^b I^b$
63. Marriage between persons having AB blood groups would produce [WB-JEE 2016]  
(a) Offsprings with AB blood group only  
(b) Offsprings with A, B and AB blood groups  
(c) Offsprings with A and B blood groups only  
(d) Offsprings with A, B, AB and O blood groups
64. Among the following characters, which one was not considered by Mendel in his experiments on pea [NEET 2017]  
(a) Stem – Tall of Dwarf  
(b) Trichomes – Glandular or non-glandular  
(c) Seed – Green or Yellow  
(d) Pod – Inflated or Constricted

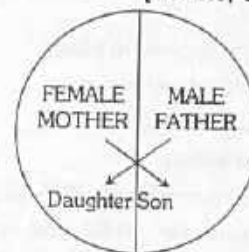
## Genetic variation

1. A person affected by disease having chromosome complement XXX is called/having [Odisha JEE 2008]  
(a) Klinefelter's syndrome  
(b) Down's syndrome  
(c) Super female  
(d) Turner's syndrome
2. With respect to phenylketonuria identify which statement is not correct [KCET 2015]  
(a) It is a case of aneuploidy  
(b) It is an example of pleiotropy  
(c) Caused due to autosomal recessive trait  
(d) It is an error in metabolism
3. Mating between two individuals differing in genotype to produce genetic variation is called [J & K CET 2005]  
(a) Domestication (b) Introduction  
(c) Hybridisation (d) Mutation
4. Sickle cell anaemia is most resistant to which disease [Odisha JEE 2008]  
(a) Malaria (b) Filariasis  
(c) Dengue (d) Chicken pox
5. If an albino man marries with a normal woman and 50 offsprings are albino and 50 are normal, the woman is [MP PMT 1998]  
(a) Heterozygous normal (b) Homozygous normal  
(c) Heterozygous carrier (d) None of these
6. Occurrence of cell containing multiples of  $2n$  genomes in diploid organisms is known as [VITEEE 2008]  
(a) Aneuploidy (b) Allopolyploidy  
(c) Amphiploidy (d) Endopolyploidy
7. The genetic defect-adenosine deaminase (ADA) deficiency may be cured permanently by [CBSE PMT 2009; Kerala PMT 2012]  
(a) Periodic infusion of genetically engineered lymphocytes having functional ADA cDNA  
(b) Administering adenosine deaminase activators  
(c) Introducing bone marrow cells producing ADA into cells at early embryonic stages  
(d) Enzyme replacement therapy



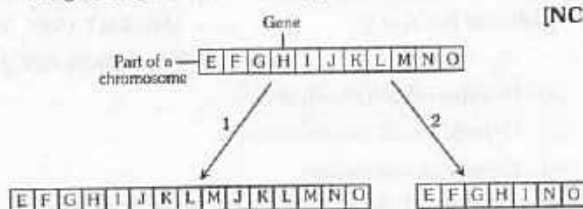
8. The hereditary disease in which the urine of a person turns black on exposure to air due to the presence of homogentisic acid is known as [BPV 2001; BHU 2012]  
 (a) Ketonuria (b) Phenylketonuria  
 (c) Haematuria (d) Alkaptonuria
9. Which is the most common mechanism of genetic variation in the population of a sexually-reproducing organism [AIPMT (Cancelled) 2015]  
 (a) Chromosomal aberrations  
 (b) Genetic drift  
 (c) Recombination  
 (d) Transduction
10. To be evolutionary successful, a mutation must be [MP PMT 1996]  
 (a) Germplasm DNA (b) Somatoplasm DNA  
 (c) Cytoplasm (d) RNA
11. Which of the chromosomal formulation is responsible for the expression of meta-male character in *Drosophila* [Kerala PMT 2007; WB-JEE 2016]  
 (a)  $2A+3X$  (b)  $3A+3X$   
 (c)  $4A+3X$  (d)  $3A+XY$   
 (e)  $2A+XY$
12. Sickle cell anaemia is [CBSE PMT 2009]  
 (a) An autosomal linked dominant trait  
 (b) Caused by substitution of valine by glutamic acid in the beta globin chain of haemoglobin  
 (c) Caused by a change in a single base pair of DNA  
 (d) Characterized by elongated sickle like RBCs with a nucleus
13. Alzheimer disease in humans is associated with the deficiency of [CBSE PMT 2009]  
 (a) Dopamine  
 (b) Glutamic acid  
 (c) Acetylcholine  
 (d) Gamma aminobutyric acid (GABA)
14. Industrial melanism as observed in peppered moth proves that [CBSE PMT 2007]  
 (a) The true black melanic forms arise by a recurring random mutation  
 (b) The melanic form of the moth has no selective advantage over lighter form in industrial area  
 (c) The lighter-form moth has no selective advantage either in polluted industrial area or non-polluted area  
 (d) Melanism is a pollution-generated feature
15. Mongoloid condition is related to or In mongolism a patient shows [MP PMT 1995; CBSE PMT 2001]  
 (a) Monosomy (b) Trisomy  
 (c) Nullisomy (d) None of the above
16. Which of the following is a genetic disease [CBSE PMT 1990; JIPMER 1993]  
 (a) Phenylketonuria (b) Blindness  
 (c) Cataract (d) Leprosy
17. Which one of the following conditions in humans is correctly matched with its chromosomal abnormality/linkage [CPMT 1994; MP PMT 1994, 98, 2000, 02; Kerala CET 2003; BVP 2003; CBSE PMT 2008; J & K CET 2012]  
**Or**  
 An abnormal human male phenotype involving an extra X-chromosome is a case of [CBSE PMT 1995, 96; CPMT 1996; MP PMT 1997, 2003, 04]  
 (a) Erythroblastosis foetalis - X-linked  
 (b) Down syndrome - 44 autosomes + XO  
 (c) Klinefelter's syndrome - 44 autosomes + XXY  
 (d) Colour blindness - Y-linked
18. Mutation is [NCERT; MP PMT 1993]  
 (a) Sudden change in morphology  
 (b) Change in characters  
 (c) Change in heritable characters  
 (d) None of these
19. A person who is trisomic for twenty first pair of chromosomes is [CPMT 1993, 2000; DPMT 1993; MP PMT 1993; WB JEE 2008; J & K CET 2012; NEET (Karnataka) 2013]  
**Or**  
 Number of sex chromosomes is normal in [MP PMT 1992]  
 (a) Klinefelter's syndrome (b) Down's syndrome  
 (c) Turner's syndrome (d) None of these
20. The monosomic condition in human beings depicted as XO is referred to as [CPMT 1994; MP PMT 1994, 98, 2002, 03; AIIMS 1999; Pb. PMT 2000; JIPMER 2002; DPMT 2007; BHU 2008]  
 (a) Criminal syndrome (b) Down's syndrome  
 (c) Klinefelter's syndrome (d) Turner's syndrome
21. Point (Gene mutation) mutation involves [MP PMT 1995; JIPMER 2001; AMU (Med.) 2009; CBSE PMT 2009]  
 (a) Insertion (b) Change in single base pair  
 (c) Duplication (d) Deletion
22. The number of chromosomes in Turner's syndrome is [CBSE PMT 1993]  
 (a) 45 (b) 43  
 (c) 44 (d) 42
23. Which of the following disorders is not hereditary [J & K CET 2005]  
 (a) Haemophilia (b) Cataract  
 (c) Sickle-cell anaemia (d) Colour blindness
24. Disorders of amino acid metabolism results in [CBSE PMT 1993; Kerala PMT 2004]  
 (a) Alkaptonuria (b) Phenylketonuria  
 (c) Albinism (d) All the above
25. The **incorrect** statement with regard to haemophilia is [NEET 2013]  
 (a) A single protein involved in the clotting of blood is affected  
 (b) It is a sex-linked disease  
 (c) It is a recessive disease  
 (d) It is a dominant disease

26. The point mutations A to G, C to T, C to G and T to A in DNA are [JIPMER 1993]  
 (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
27. Genomic mutation is [AFMC 2008]  
 (a) Change in number genes  
 (b) Change in number of chromosomes  
 (c) Change in shape of chromosomes  
 (d) All of these
28. Haploids are more suitable for mutation studies than the diploids. This is because [CBSE PMT 2008]  
 (a) Haploids are more abundant in nature than diploids  
 (b) All mutations, whether dominant or recessive are expressed in haploids  
 (c) Haploids are reproductively more stable than diploids  
 (d) Mutagens penetrate in haploids more effectively than in diploids
29. The number of chromosomes in Down's syndrome is [MP PMT 1992, 98, 2005; CPMT 2002; BVP 2002; MHCET 2002; CBSE PMT 2002, 05; DPMT 2003, 06; AFMC 2005; RPMT 2006; BHU 2006; WB JEE 2009]  
 (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
30. When a mutation is limited to the substitution of one nucleotide for another, it is called [Kerala PMT 2008; MP PMT 2010]  
 (a) Translocation  
 (b) Point mutation  
 (c) Base inversion  
 (d) Sugar phosphate deletion  
 (e) Frame shift
31. A man having Klinefelter's syndrome is [J & K CET 2005; MP PMT 2005]  
 (a) Intersex with secondary sexual characters on the side of female  
 (b) Male with secondary sexual characters of female  
 (c) Female with secondary sexual characters of male  
 (d) Normal fertile male
32. An abnormal human baby with 'XXX' sex chromosomes was born due to [AIPMT (Cancelled) 2015]  
 (a) Formation of abnormal ova in the mother  
 (b) Fusion of two ova and one sperm  
 (c) Fusion of two sperms and one ovum  
 (d) Formation of abnormal sperms in the father
33. Edward's syndrome, Patau's syndrome and Down's syndrome are due to [MP PMT 1997, 2003; CPMT 1999; Odisha JEE 2005]  
 (a) Mutation due to malnutrition  
 (b) Change in sex chromosomes  
 (c) Change in autosomes  
 (d) Change in both sex chromosomes and autosomes
34. Which of these is not a Mendelian disorder [Kerala PMT 2008, 10]  
 (a) Cystic fibrosis (b) Sickle cell anaemia  
 (c) Colour blindness (d) Haemophilia  
 (e) Turner's syndrome
35. Moody describes the mutation as [MP PMT 2010]  
 (a) Sports (b) Saltation  
 (c) Factors (d) Shotgun
36. Which of the following mutations is not hereditary [MP PMT 2012]  
 (a) Genetic (b) Gametic  
 (c) Somatic (d) Germinal
37. Represented below is the inheritance pattern of a certain type of traits in humans. Which one of the following conditions could be an example of this pattern [NCERT; CBSE PMT (Mains) 2012]



- (a) Phenylketonuria (b) Sickle cell anaemia  
 (c) Haemophilia (d) Thalassemia
38. What would be the number of chromosomes 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
39. A man whose father was colour blind marries a woman who had a colour blind mother and normal father. What percentage of male children of this couple will be colour blind [CBSE PMT 2014]  
 (a) 50% (b) 75%  
 (c) 25% (d) 0%
40. Trisomic condition of Down's syndrome arises due to [CBSE PMT 1991; MP PMT 2003; DUMET 2009]  
 (a) Triploidy  
 (b) Translocation  
 (c) Non-disjunction  
 (d) Dicentric bridge formation

41. The given figure shows two types of chromosome mutation [NCERT]



These are called

- (a) 1 - Inversion, 2 - Substitution  
(b) 1 - Inversion, 2 - Deletion  
(c) 1 - Duplication, 2 - Substitution  
(d) 1 - Duplication, 2 - Deletion
42. Hugo de Vries formulated the "Mutation Theory" based on the experiments he conducted on [NCERT; CPMT 1993; MP PMT 1994; CBSE PMT 2005; Odisha JEE 2005, 08; RPMT 2006; VITEEE 2006]  
(a) *Althea rosea*  
(b) *Pisum sativum*  
(c) *Drosophila melanogaster*  
(d) *Oenothera lamarckiana*
43. A hereditary disease which is never passed on from father to son is [J & K CET 2005]  
(a) Autosomal linked disease  
(b) X-chromosomal linked disease  
(c) Y-chromosomal linked disease  
(d) None of these
44. Somaclonal variation appears in plants [DUMET 2009]  
(a) Growing in polluted soil or water  
(b) Exposed to gamma rays  
(c) Raised in tissue culture  
(d) Transformed by recombinant DNA technology
45. In *Drosophila*, gene for white eye mutation is also responsible for depigmentation of body parts. Thus a gene that controls several phenotypes is called [Kerala CET 2005]  
(a) Oncogene  
(b) Epistatic gene  
(c) Hypostatic gene  
(d) Pleiotropic gene  
(e) Sex-linked gene
46. The functional unit of mutation is [JIPMER 1994; MP PMT 1994; AFMC 1995]  
(a) Gene  
(b) Muton  
(c) Recon  
(d) Cistron
47. The most striking example of point mutation is found in a disease called [CBSE PMT 1995]  
**Or**  
In which of the following disorders, blood has a defective haemoglobin [KCET 2006]  
(a) Night blindness  
(b) Thalassemia  
(c) Down's syndrome  
(d) Sickle-cell anaemia
48. Which following pair of diseases is caused by two genes located on human X-Chromosome [WB JEE 2012]  
(a) Colour blindness and phenylketonuria  
(b) Colour blindness and haemophilia  
(c) Colour blindness and albinism  
(d) Colour blindness and hypertrichosis

49. Which of the following is not related to chromosomal aberration [MP PMT 1995]  
(a) Euploidy  
(b) AIDS  
(c) Aneuploidy  
(d) Klinefelter's syndrome

50. Sickle cell anaemia is [DUMET 2009; WB JEE 2011]  
(a) Autosomal dominant inheritance  
(b) X-linked recessive inheritance  
(c) Autosomal recessive inheritance  
(d) X-linked dominant inheritance

51. The frequency of a mutant gene in a population is expected to increase, if the gene is [CBSE PMT 1994]  
(a) Recessive  
(b) Dominant  
(c) Sex linked  
(d) Favourably selected

52. Albinism is a congenital disorder (non synthesis of melanin) resulting from the lack of the enzyme [CBSE PMT 1994; BHU 2003, 12]  
(a) Catalase  
(b) Fructokinase  
(c) Tyrosinase  
(d) Xanthine oxidase

53. Sometimes chromosome number increase or decrease due to [AFMC 1996]  
(a) Non-disjunction of chromosome  
(b) Genetic repete  
(c) Mutation  
(d) All of these

54. Match the following

List-I		List-II	
(A)	XX-XO, method of sex determination	(I)	♀ Heterogametic
(B)	1.5 X/A ratio	(II)	Turner's syndrome
(C)	Karyotype 45	(III)	Hemiptera
(D)	ZW-ZZ method of sex determination	(IV)	Metafemale

The correct match is

[EAMCET 2009]

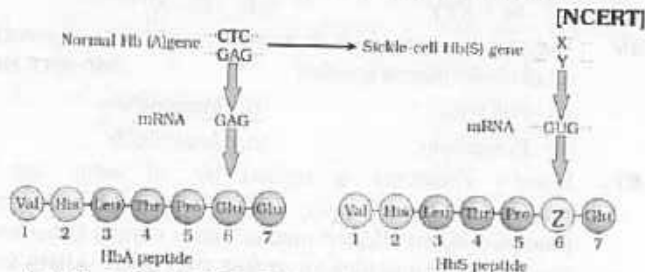
	A	B	C	D
(a)	I	IV	III	II
(b)	III	IV	II	I
(c)	IV	I	II	III
(d)	I	IV	II	III

55. The idea of mutations was brought forth by [NCERT; DPMT 2006; Kerala PMT 2009; CBSE PMT (Mains) 2012]  
(a) Hugo de Vries, who worked on evening primrose  
(b) Gregor Mendel, who worked on *Pisum sativum*  
(c) Hardy Weinberg, who worked on allele frequencies in a population  
(d) Charles Darwin, who observed a wide variety of organisms during sea voyage

56. Edward syndrome is on account of ..... [MP PMT 1994, 99, 2000, 04]  
(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



57. Sickle-cell anaemia is an autosome linked recessive trait that can be transmitted from parents to the offspring when both the partners are carrier for all the gene (or heterozygous). The disease is controlled by a single pair of allele,  $Hb^A$  &  $Hb^S$ . Out of the three possible genotypes only homozygous individuals for  $Hb^S$  ( $Hb^S Hb^S$ ) are lethal. Select the right option in which X, Y and Z are correctly identified



- (a) X- CAC; -Y GTG; Z His (b) X- GTG; -Y CAC; Z Val  
(c) X- CAC; -Y GTG; Z Phe (d) X- CAC; -Y GTG; Z Val

58. Pick out the correct statements

- (i) Haemophilia is a sex-linked recessive disease  
(ii) Down's syndrome is due to aneuploidy  
(iii) Phenylketonuria is an autosomal dominant gene disorder  
(iv) Phenylketonuria is an autosomal recessive gene disorder  
(v) Sickle-cell anaemia is an X-linked recessive gene disorder [Kerala CET 2005; Kerala PMT 2008, 09]

- (a) (i), (iii) and (v) are correct  
(b) (i) and (iii) are correct  
(c) (ii) and (v) are correct  
(d) (i), (iv) and (v) are correct  
(e) (i), (ii) and (iv) are correct

59. Match column I with column II and find the correct answer

Column I	Column II
(A) Monoploidy	(1) $2n - 1$
(B) Monosomy	(2) $2n + 1$
(C) Nullisomy	(3) $2n + 2$
(D) Trisomy	(4) $2n - 2$
(E) Tetrasomy	(5) $n$
	(6) $3n$

[DPMT 2003, 06; Kerala PMT 2009; WB JEE 2010]

- (a) (A) — (5), (B) — (1), (C) — (4), (D) — (2), (E) — (3)  
(b) (A) — (5), (B) — (2), (C) — (4), (D) — (1), (E) — (3)  
(c) (A) — (6), (B) — (5), (C) — (3), (D) — (4), (E) — (2)  
(d) (A) — (2), (B) — (1), (C) — (3), (D) — (6), (E) — (5)  
(e) (A) — (1), (B) — (5), (C) — (3), (D) — (2), (E) — (4)

60. Height is

[CPMT 2005]

- (a) Somatogenic variation (b) Discontinuous variation  
(c) Continuous variation (d) Blastogenic variation

61. Which one of the following is not a mutagen

[MP PMT 1995, 97; 2000]

- (a) Ethyl methane sulphonate  
(b) Acetic acid  
(c) Nitrous acid  
(d) Ethylene oxide

62. Mutation rates are affected by

[MP PMT 1997;

Bihar CECE 2006; CBSE PMT (Pre.) 2011]

- (a) Temperature  
(b) X-rays  
(c) Gamma and beta radiation  
(d) All of the above

63. A mutation is most likely to have a selective advantage in evolution if

[CPMT 2005]

- (a) It affects dominant genes  
(b) It affects recessive genes  
(c) It affects whole chromosomes  
(d) The environment remains stable

64. Which one of the following is a wrong statement regarding mutations

[CBSE PMT (Mains) 2012]

- (a) Deletion and insertion of base pairs cause frame-shift mutations  
(b) Cancer cells commonly show chromosomal aberrations  
(c) UV and Gamma rays are mutagens  
(d) Change in a single base pair of DNA does not cause mutation

65. The gene for diabetes mellitus is

[BHU 2012]

- (a) Autosomal dominant (b) Autosomal recessive  
(c) Sex-linked dominant (d) Sex linked recessive

66. Gynaecomastia is the symptom of

[DPMT 2004; Kerala PMT 2007, 08, 10]

- (a) Down syndrome (b) SARS  
(c) Turner's syndrome (d) Klinefelter's syndrome

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

[CBSE PMT 1999]

- (a) Genetic mutation (b) Zygotic mutation  
(c) Somatic mutation (d) All of these

68. In human beings, multiple genes are involved in the inheritance of

[CBSE PMT 1999]

- (a) Colourblindness (b) Phenylketonuria  
(c) Sickle-cell anaemia (d) Skin colour

69. The formation of multivalents at meiosis in diploid organism is due to

[CBSE PMT 1998]

- (a) Monosomy (b) Inversion  
(c) Deletion (d) Reciprocal translocation

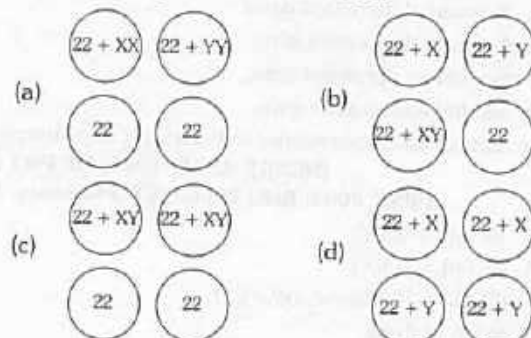
70. Mental retardation in man, associated with sex chromosomal abnormality is usually due to

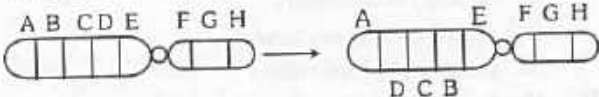
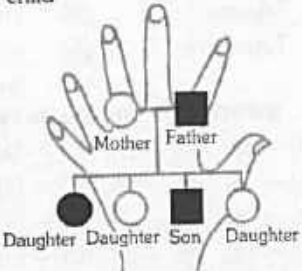
[CBSE PMT 1998]

- (a) Reduction in X complement  
(b) Increase in X complement  
(c) Moderate increase in Y complement  
(d) Large increase in Y complement

71. If chromosome complement  $44+XY$  of a gamete mother cell suffers a non-disjunction at the time of first meiotic division. Which sets of gametes will be correct

[NCERT]



72. If haploid chromosome number in a cell is 12. The monosomic number will be [MP PMT 1997, 2000]  
(a) 24 (b) 21  
(c) 25 (d) 23
73. Normally DNA molecule has A-T, G-C pairing. However, these bases can exist in alternative valency status, owing to rearrangements called [BHU 2000; WB JEE 2009]  
(a) Point mutation (b) Analogue substitution  
(c) Frame-shift mutation (d) Tautomerisational mutation
74. Who reported that Down's syndrome is due to extra 21st chromosome [BVP 2000]  
(a) J.L. Down (1866) (b) Lejeune (1959)  
(c) Klinefelter (1942) (d) Huntington (1872)
75. In agriculture mutation caused by a mutagen is [NCERT; BVP 2001]  
(a) Natural (b) Chemical  
(c) Spontaneous (d) Induced
76. Discontinuous variations are [AIIMS 2001]  
Or  
The reason of fault in gene duplication is [RPMT 1999]  
(a) Mutations (b) Acquired characters  
(c) Essential features (d) Nonessential features
77. Epicanthal skin fold and simian crease are characteristics of [KCET 2001; MH CET 2015]  
(a) Haploidy (b) Heteroploidy  
(c) Turner's syndrome (d) Down's syndrome
78. UV radiations cause [BHU 2012]  
(a) Formation of thymine dimers  
(b) Deletion of base pairs  
(c) Methylation of bases  
(d) Addition of base pairs
79. A person may have one gene for normal haemoglobin and one gene for sickle cell haemoglobin. This heterozygous condition is called [BHU 2002]  
(a) Genome (b) Anaemia  
(c) Gene trait (d) Sickle cell trait
80. If a diploid cell is treated with colchicine, then it becomes [CBSE PMT 2002]  
(a) Tetraploid (b) Diploid  
(c) Triploid (d) Monoploid
81. Which one of the following mutation partially or fully reverses the harmful effects of previous mutation [MP PMT 2002]  
(a) Indirect suppression (b) Intergenic mutation  
(c) Intragenic mutation (d) Suppressor mutation
82. Polydactyly in man is due to [J & K CET 2002]  
(a) Autosomal dominant gene  
(b) Autosomal recessive gene  
(c) Sex-linked dominant gene  
(d) Sex-linked recessive gene
83. The number of chromosomes in Klinefelter's syndrome is [NCERT; CPMT 1995; MP PMT 2003; DPMT 2004; BHU 2006; PET (Pharmacy) 2013]  
(a) 47 (44 + XXY)  
(b) 47 (44 + XXX)  
(c) 47 (46 + 1 chromosome 21)  
(d) None of these
84. Philadelphia chromosome is [MH CET 2002]  
(a) 13th chromosome (b) 22nd chromosome  
(c) 17th chromosome (d) 21st chromosome
85. Which of the following chromosomal constitution refers to Jacob's syndrome in human [BHU 2012]  
(a) 44 + XO (b) 44 + XXY  
(c) 44 + XYY (d) 45 + XYY
86. The condition in which there are more than two complete set of chromosome is called [MP PMT 2003]  
(a) Polytene (b) Monoploidy  
(c) Polyploidy (d) Aneuploidy
87. 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 [CBSE PMT 2003; AIIMS 2007]  
(a) 25% (b) 100%  
(c) 75% (d) 50%
88. Polyploidy can be induced by the application of [MP PMT 2009]  
(a) Auxin (b) Kinetin  
(c) Colchicine (d) Ethylene
89. Given below is the representation of a kind of chromosomal mutation  
What is the kind of mutation represented [AFMC 1997; AIIMS 2004]  
Or  
When a segment of a chromosome breaks and later rejoins after 180° rotation, it is known as [Keral PMT 2009]
- 
- (a) Deletion (b) Duplication  
(c) Inversion (d) Reciprocal translocation
90. In the given human hand pedigree which character is represented and what is the probability of disease occurrence in fifth child [NCERT]
- 
- (a) Polydactyly (X-linked recessive disorder), 50%  
(b) Polydactyly (X-linked dominant disorder), 50%  
(c) Polydactyly (autosomal recessive disorder), 50%  
(d) Polydactyly (autosomal dominant disorder), 50%
91. A recessive mutant is one which [Odisha JEE 2004]  
(a) Is not expressed  
(b) Is rarely expressed  
(c) Is expressed only in homozygous and hemixygous state  
(d) Is expressed only in heterozygous state.

92. Frequency of Down's syndrome increases when the maternal age is [Odisha JEE 2004]  
 (a) Above 35 years  
 (b) Below 35 years  
 (c) During 1st pregnancy  
 (d) In mothers of at least 3 children
93. Addition or deletion of a single nucleotide results in which type of mutation [BHU 2012; Odisha JEE 2012]  
 (a) Deficiency (b) Duplication  
 (c) Frameshift mutation (d) None of these
94. Change in the number of body parts is called [MP PMT 2009]  
 (a) Continuous variation (b) Discontinuous variation  
 (c) Meristic variation (d) Substantive variation
95. Turner's syndrome is an example of [Kerala PMT 2004]  
 (a) Monosomy (b) Bisomy  
 (c) Trisomy (d) Polyploidy  
 (e) Translocation
96. In man, which of the following genotypes and phenotypes may be the correct result of aneuploidy in sex chromosomes [CPMT 2004]  
 (a) 22 pairs + XXY males  
 (b) 22 pairs + XX females  
 (c) 22 pairs + XXXY females  
 (d) 22 pairs + Y females
97. The "cri-du-chat" syndrome is caused by change in chromosome structure involving [AIIMS 2005; Kerala PMT 2007, 08]  
**Or**  
 The loss of a chromosomal segment is due to [Kerala PMT 2011]  
 (a) Deletion (b) Duplication  
 (c) Inversion (d) Translocation
98. Somaclonal variation appears in [AIIMS 2005]  
 (a) Organism produced through somatic hybridization  
 (b) Plants growing in highly polluted conditions  
 (c) Apomictic plants  
 (d) Tissue culture raised plants
99. Mutation cannot change [KCET 2007]  
 (a) RNA (b) Environment  
 (c) Enzyme (d) DNA
100. Turner's syndrome in human is caused by  
 (a) Autosomal aneuploidy  
 (b) Sex chromosome aneuploidy  
 (c) Polyploidy  
 (d) Point mutation
101. Which of the following corresponds to mutagens [Odisha JEE 2012]  
 (a) Chemicals and radiations which cause changes in the genetic material of a cell  
 (b) Various archaebacteria that produce methane  
 (c) Chemicals which react with ozone molecules and destroy them  
 (d) RNA molecules that infect plant cells and cause diseases
102. Both sickle cell anaemia and Huntington's chorea are [CBSE PMT 2006]  
 (a) Pollutant-induced disorders  
 (b) Virus-related diseases  
 (c) Bacteria-related diseases  
 (d) Congenital disorders
103. 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 [CBSE PMT 1998]  
**Or**  
 If both parents are carriers for thalassemia, which is an autosomal recessive disorder, what are the chances of pregnancy resulting in an affected child [NEET 2013]  
 (a) 100% (b) 25%  
 (c) 50% (d) 75%
104. Loss or gain of one or more complete set of chromosomes along with the diploid complement is known as [Odisha JEE 2012]  
 (a) Aneuploidy  
 (b) Euploidy  
 (c) Reverse tandem duplication  
 (d) Substitution mutation
105. Addition of one or more haploid set of its own genome in an organism results in [VITEEE 2006]  
 (a) Autopolyploidy (b) Allopolyploidy  
 (c) Aneuploidy (d) Diploid
106. Autosomal mutant allele *HbS* causes [VITEEE 2006]  
 (a) Thalassemia (b) Albinism  
 (c) Sickle cell anaemia (d) Agammaglobulinemia
107. Due to nondisjunction of chromosomes during spermatogenesis, sperms carry both sex chromosomes (22A+XY) and some sperms do not carry any sex chromosome (22A+0). If these sperms fertilize normal eggs (22A+X), what types of genetic disorders appear among the offsprings [KCET 2010]  
 (a) Turner's syndrome and Klinefelter's syndrome  
 (b) Down's syndrome and Klinefelter's syndrome  
 (c) Down's syndrome and Turner's syndrome  
 (d) Down's syndrome and cri-du-chat syndrome
108. Genetic variation in a population arises due to [NEET (Karnataka) 2013]  
 (a) Recombination only  
 (b) Mutations as well as recombination  
 (c) Reproductive isolation and selection  
 (d) Mutations only
109. The chromosomal condition in Turner's syndrome is [Kerala PMT 2011; MP PMT 2011; KCET 2012]  
**Or**  
 A human female with Turner's syndrome [CBSE PMT 2014]  
 (a) 21 Trisomy with XY (b) 44 Autosomes + XXY  
 (c) 44 Autosomes + XYY (d) 44 Autosomes + XO  
 (e) 18 Trisomy with XY
110. The change in single base pair [Odisha JEE 2011]  
 (a) Results in new species  
 (b) Always changes the polypeptide chain  
 (c) May not change the phenotype  
 (d) Always changes the phenotype



111. Which is correct for Turner's syndrome

[NCERT; AMU (Med.) 2012]

- (a) It is a case of monosomy
- (b) It causes sterility in females
- (c) Absence of Barr body
- (d) All of the above

112. Sickle cell anaemia is caused by the substitution of

[AFMC 1997; Kerala CET 2004, 05; Kerala PMT 2012]

- (a) Valine by glutamic acid at sixth position of alpha chain of haemoglobin
- (b) Valine by glutamic acid at sixth position of beta chain of haemoglobin
- (c) Glutamic acid by valine at sixth position of alpha chain of haemoglobin
- (d) Glutamic acid by valine at sixth position of beta chain of haemoglobin
- (e) Glutamic acid by methionine at sixth position of alpha chain of haemoglobin

113. If a colour blind man marries a woman who is normal but carries this trait, the progeny will be [MP PMT 2013]

- (a) All normal females but carrier of the trait
- (b) All males and 50% females colour blind
- (c) All females and 50% males colour blind
- (d) 50% males and 50% females colour blind

114. Thalassaemia and sickle cell anemia are caused due to a problem in globin molecule synthesis. Select the correct statement [NEET 2017]

- (a) Both are due to a qualitative defect in globin chain synthesis
- (b) Both are due to a quantitative defect in globin chain synthesis
- (c) Thalassaemia is due to less synthesis of globin molecules
- (d) Sickle cell anemia is due to a quantitative problem of globin molecules

115. A disease caused by an autosomal primary non-disjunction is [NEET 2017]

- (a) Down's Syndrome
- (b) Klinefelter's Syndrome
- (c) Turner's Syndrome
- (d) Sickle Cell Anemia

### Sex determination

1. Sex chromosomes of a female bird are represented by

[Kerala PMT 2008]

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

2. When released from ovary human egg contain [AFMC 2006]

- (a) One Y chromosome
- (b) Two X chromosome
- (c) One X chromosome
- (d) XY chromosome

3. Barr bodies (seen in saliva test in Olympic games) are found in human and are associated with

[CBSE PMT 1992; MP PMT 1997, 98; KCET 1998;

BHU 1999, 2002; RPMT 2005]

- (a) Male autosome
- (b) Female autosome
- (c) Female sex chromosome
- (d) Male sex chromosome

4. The chromosomes responsible for the determination of sex are called

- (a) Autosomes
- (b) Allosomes
- (c) Multiple alleles
- (d) Heterosis

5. Sex chromosomes for the first time was discovered in which plant [DPMT 2006]

- (a) *Sphaerocarpus*
- (b) *Pisum sativum*
- (c) *Neurospora*
- (d) *Lathyrus odoratus*

6. XO type of sex determination is seen in

[CPMT 2010; Kerala PMT 2011]

- (a) Man
- (b) Grasshopper
- (c) *Drosophila*
- (d) Birds
- (e) Horses

7. The barr body is observed in [Kerala CET 2005]

- (a) Basophils of males
- (b) Neutrophils of females
- (c) Eosinophils
- (d) Neutrophils of males

8. In *Drosophila*, the sex is determined by [MP PMT 1994; 95; CBSE PMT 2003; AIEEE Pharmacy 2004; AIIMS 2011]

- (a) Whether the egg is fertilized or develops parthenogenetically
- (b) The ratio of number of X-chromosomes to the sets of autosomes
- (c) X and Y chromosomes
- (d) The ratio of pairs of X-chromosomes to the pairs of autosomes

9. In XO type of sex determination [Kerala PMT 2012]

- (a) Females produce two different type of gametes
- (b) Males produce two different types of gametes
- (c) Females produce gametes with Y chromosomes
- (d) Males produce single type of gametes
- (e) Males produce gametes with Y chromosomes

10. Which type of gene regulate sex-determination in Spinach plant [GUJCET 2015]

- (a) Homozygous genes
- (b) Heterozygous genes
- (c) Single gene
- (d) Multiple genes

11. The sex determination pattern in honeybee is called

[MP PMT 1993, 2005; AIIMS 1993]

- (a) Female haploidy
- (b) Haplodiploidy
- (c) Gametic diploidy
- (d) Gametogony

12. Sex of a human child is determined by

[MP PMT 1993, 2001, 03]

- (a) Size of the egg at the time of fertilization
- (b) Size of the sperm at the time of fertilization
- (c) Sex chromosome of father
- (d) Sex chromosome of mother

13. Lyon hypothesis deals with [DPMT 1993]

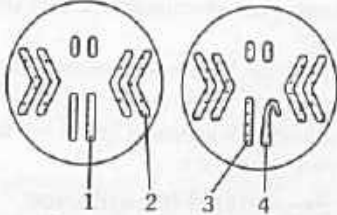
- (a) Centromere position
- (b) Genetic compatibility
- (c) Genetic incompatibility
- (d) Number of Barr bodies

14. Meta-females have [DPMT 1993]

- (a) XX
- (b) XO
- (c) XXXX
- (d) XXXXXX

15. Animal which remains male initially, then changes to female (*Tapeworm proglottides*) is called [CBSE PMT 1993]

- (a) Protandrous
- (b) Apomixis
- (c) Profixation
- (d) None of these

16. Barr bodies and drumsticks are of what significance to geneticists and biologists [AFMC 1993]  
 (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 calls  
 (d) They signify the presence of sex linked traits
17. A family has five girls and no son. Probability of son as the 6th child will be [NCERT; AFMC 2000; CPMT 2005]  
 (a) 50% (b) 75%  
 (c) Full (d) No chance
18. Barr bodies are [CBSE PMT 1993]  
 (a) Chromatin negative (b) Not influenced by stains  
 (c) Chromatin positive (d) Poorly staining
19. Chromosomal abnormality of an unborn baby (while in mother's womb) can be found out by a technique called [MP PMT 1995]  
 (a) Amniocentesis (b) CAT scanning  
 (c) Ultrasound (d) Tissue culture
20. In our society women are blamed for producing female children. Choose the correct answer for the sex-determination in humans [NEET (Karnataka) 2013]  
 (a) Due to some defect like aspermia in man  
 (b) Due to the genetic make up of the particular sperm which fertilizes the egg  
 (c) Due to the genetic make up of the egg  
 (d) Due to some defect in the women
21. Foetal sex can be determined by examining cells from amniotic fluid looking for [CBSE PMT 1991]  
 (a) Barr bodies (b) Chiasmata  
 (c) Sex chromosomes (d) Kinetochore
22. Chromosomes that determine male sex in *Melandrium* plant is [DPMT 2004]  
 (a) Y chromosome (b) X chromosome  
 (c) XX chromosome (d) None of these
23. Male child will be born if [CPMT 1993, 95; BHU 1995]  
 (a) Father is sexually more excited  
 (b) Sperm of male with Y chromosome fertilizes the egg  
 (c) Sperm of male with X chromosome fertilizes the egg  
 (d) None of the above
24. Barr body in mammals represents [CBSE PMT 1995]  
 (a) All the heterochromatin in female cells  
 (b) One of the two X chromosomes in somatic cells of females  
 (c) All the heterochromatin in male and female cells  
 (d) The Y chromosome in somatic cells of male
25. Based on Lyon's hypothesis, what will be the number of Barr bodies found in a human female suffering from Down's syndrome [AIEEE Pharmacy 2004]  
 (a) 0 (b) 1  
 (c) 2 (d) 3
26. *Drosophila* flies with one half of the body male and other half female is referred to as [CBSE PMT 1994]  
 Or  
 Loss of a X chromosome in a particular cell during its development, results into [CBSE PMT 1998]  
 (a) Gynandromorph (b) Hermaphrodite  
 (c) Super female (d) Intersex
27. In human female, barr bodies are formed by [CBSE PMT 1996]  
 (a) Inactivation of mother's X chromosome  
 (b) Inactivation of father's X chromosome  
 (c) Inactivation of both mother's and father's X chromosomes  
 (d) Inactivation of either mother's or father's X chromosome
28. Gynandromorphs develop in *Drosophila* when the two cells in the two-celled proembryo will have one of the following chromosomal sets [MP PMT 1997]  
 (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
29. Genic balance theory of sex determination was proposed by [MP PMT 1997, 99, 2000; KCET 2004]  
 (a) Morgan (b) Bridges  
 (c) Boveri (d) Wilkins
30. A medical technician while observing a human blood smear under the microscope notes the presence of barrbody close to the nuclear membrane in the WBC. This indicates that person under investigation is [Kerala CET 2003]  
 (a) Colour blind (b) Haemophilic  
 (c) Normal female (d) Normal male
31. The following figure refer to the chromosome complement of each sex of fruit fly
- 
- By which number is a Y chromosome labelled  
 (a) 4 (b) 3  
 (c) 2 (d) 1
32. Genetic identity of a human male is determined by [CBSE PMT 1997; Pb. PMT 1999; KCET 2000; CPMT 2000]  
 (a) Autosome (b) Nucleolus  
 (c) Sex chromosome (d) Cell organelles
33. The theory where ratio between the number of X-chromosomes and number of complete sets of autosomes will determine the sex is known as [MP PMT 2002]  
 (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
34. If somatic cells of a human male contain single barrbody, the genetic composition of the person would be [JIPMER 1993; MP PMT 2001]  
 Or  
 The genotype of a boy having sexual characters of a girl is  
 (a) XYY (b) XXY  
 (c) XO (d) XXXY
35. Chromosome theory of sex determination was propounded by [MP PMT 2002; BVP 2002]  
 (a) Bridges (b) Balbiani  
 (c) Goldschmidt (d) None of the above

36. Identify the wrong statement [Kerala PMT 2010]
- In male grasshoppers 50% of the sperms have no sex chromosome
  - Usually female birds produce two types of gametes based on sex chromosomes
  - The human males have one of their sex chromosomes much shorter than the other
  - The male fruit fly is heterogametic
  - In domesticated fowls the sex of the progeny depends on the type of sperm that fertilizes the egg

37. Random genetic drift in a population probably results from [CBSE PMT 2002, 03]

- Large population size
- Highly genetically variable individuals
- Interbreeding within small isolated population
- Constant low mutation rate

38. In melandrium the sex determination type is [Kerala CET 2003]

- XX-XY type
- XX-XO type
- ZZ-ZW type
- XY-XO type

39. Which one of the following conditions correctly describes the manner of determining the sex in the given example

[NCERT; CBSE PMT (Pre.) 2011; KCET 2015]

- Homozygous sex chromosomes (XX) produce male in *Drosophila*
- Homozygous sex chromosomes (ZZ) determine female sex in birds
- XO type of sex chromosomes determine male sex in grasshopper
- XO condition in humans as found in Turner Syndrome, determines female sex.

### Sex linked Inheritance

1. A colour blind man marries the daughter of a colour blind person. Then in their progeny [AIIMS 1992; MP PMT 1994; JIPMER 2002; BHU 2008]

- None of their daughters are colour blind
- All the sons are colour blind
- All the daughters are colour blind
- Half of their sons are colour blind

2. Given is : X is the chromosome with gene for haemophilia and X is the chromosome with normal gene. Which of the following individuals will act as carrier for haemophilia [MP PMT 1992]

- $X^hY$
- $XY$
- $X^hX^h$
- $X^hX$

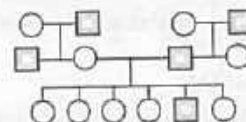
3. More men suffer from colour blindness than women because [MP PMT 1992; KCET 2011]

- Women are more resistant to disease than men
- The male sex hormone testosterone causes the disease
- The colour blind gene is carried on the 'Y' chromosome
- Men are hemizygous and one defective gene is enough to make them colour blind

4. Sex-linked characters are [CPMT 1993]

- Dominant
- Recessive
- Lethal
- Not inherited

5. This pedigree is of a rare trait, in which children have extra fingers and toes. Which one of the following patterns of inheritance is consistent with this pedigree [AIIMS 2009]

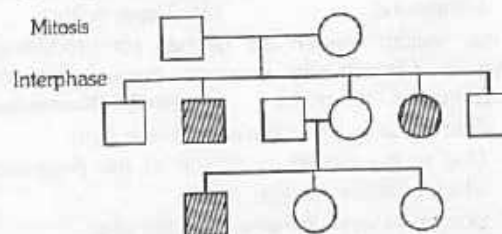


- Autosomal recessive
- Autosomal dominant
- Y-linkage
- Sex linked recessive

6. If mother is a 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]

- All the sons
- All the daughters
- 50% sons and 50% daughters (carrier)
- All the sons and not in daughters

7. Study the pedigree chart given below



What does it show [CBSE PMT 2009]

- Inheritance of a sex-linked inborn error of metabolism like phenylketonuria
- Inheritance of a condition like phenylketonuria as an autosomal recessive trait
- The pedigree chart is wrong as this is not possible
- Inheritance of a recessive sex-linked disease like haemophilia

8. A man can inherit his X chromosome from [Kerala PMT 2008]

- His maternal grand mother or maternal grand father
- His father
- His maternal grand father only
- His paternal grand father
- His paternal grand mother

9. In a pedigree analysis,  $\square = 0$  represents [Kerala PMT 2008; KCET 2015]

- Unrelated mating
- Consanguinous mating
- Affected parents
- Siblings
- Non-identical twins

10. Select the incorrect statement from the following [CBSE PMT 2009]

- Linkage is an exception to the principle of independent assortment in heredity
- Galactosemia is an inborn error of metabolism
- Small population size results in random genetic drift in a population
- Baldness is a sex-limited trait



101	c	102	b	103	b	104	a	105	a
106	a	107	b	108	d	109	a	110	d
111	c	112	a	113	d	114	c	115	b
116	b	117	d	118	c	119	a	120	d
121	a	122	b						

## Interaction of gene and cytoplasmic inheritance

1	c	2	d	3	a	4	a	5	b
6	c	7	d	8	c	9	a	10	d
11	b	12	b	13	b	14	c	15	c
16	d	17	b	18	d	19	a	20	b
21	b	22	a	23	b	24	b	25	b
26	a	27	d	28	a	29	d	30	c
31	a	32	b	33	c	34	d		

## Linkage and Crossing over

1	a	2	d	3	b	4	d	5	d
6	a	7	c	8	b	9	d	10	c
11	b	12	b	13	d	14	a	15	a
16	b	17	a	18	d	19	c	20	b
21	c	22	d	23	e	24	d	25	c
26	a	27	b	28	a	29	c	30	b
31	b	32	b	33	c	34	c		

## Chromosomes and Genes

1	d	2	a	3	c	4	c	5	d
6	c	7	d	8	e	9	b	10	d
11	b	12	d	13	b	14	d	15	d
16	a	17	b	18	d	19	b	20	b
21	a	22	b	23	a	24	a	25	a
26	b	27	a	28	a	29	c	30	c
31	c	32	c	33	c	34	c	35	e
36	a	37	a	38	c	39	c	40	b
41	c	42	a	43	b	44	d	45	a
46	d	47	b	48	c	49	d	50	a
51	b	52	d	53	a	54	a	55	d
56	b	57	c	58	e	59	d	60	a
61	b	62	a	63	c	64	a	65	a
66	b	67	c	68	d	69	a	70	c
71	a	72	c	73	a	74	b	75	b
76	b	77	b	78	c	79	b	80	a
81	b	82	c	83	c	84	a	85	d
86	b	87	b	88	a	89	d	90	d
91	a	92	d	93	c	94	c	95	d

96	a	97	d	98	c	99	c	100	d
101	d	102	a	103	c	104	b	105	d
106	d	107	d	108	d	109	d	110	d
111	c	112	b	113	d	114	c	115	c
116	b	117	b	118	a	119	d	120	c
121	d	122	c	123	b	124	c	125	a
126	d	127	c	128	c	129	b	130	a
131	a	132	a						

## Multiple allelism

1	c	2	c	3	a	4	b	5	d
6	c	7	d	8	d	9	e	10	c
11	d	12	d	13	c	14	c	15	d
16	d	17	d	18	b	19	c	20	b
21	c	22	c	23	c	24	d	25	d
26	a	27	c	28	c	29	c	30	b
31	d	32	c	33	b	34	a	35	a
36	b	37	b	38	d	39	d	40	b
41	b	42	d	43	a	44	a	45	d
46	d	47	d	48	a	49	a	50	b
51	d	52	a	53	a	54	c	55	d
56	d	57	c	58	b	59	b	60	a
61	c	62	c	63	b	64	c		

## Genetic variation

1	c	2	a	3	c	4	a	5	c
6	b	7	c	8	d	9	c	10	a
11	d	12	c	13	d	14	a	15	b
16	a	17	c	18	c	19	b	20	d
21	b	22	a	23	b	24	d	25	d
26	a	27	b	28	b	29	b	30	b
31	b	32	a	33	c	34	e	35	d
36	c	37	c	38	d	39	a	40	c
41	d	42	d	43	b	44	c	45	d
46	b	47	d	48	b	49	b	50	c
51	d	52	c	53	a	54	b	55	a
56	b	57	d	58	e	59	a	60	c
61	b	62	d	63	b	64	d	65	a
66	d	67	d	68	d	69	d	70	b
71	c	72	d	73	d	74	a	75	d
76	a	77	d	78	a	79	d	80	a
81	d	82	a	83	a	84	b	85	c
86	c	87	d	88	c	89	c	90	d

## 1242 Principles of Inheritance and Variation

91	c	92	a	93	c	94	c	95	a
96	a	97	a	98	a	99	b	100	b
101	a	102	d	103	b	104	b	105	a
106	c	107	a	108	b	109	d	110	c
111	d	112	d	113	d	114	c	115	a

## Sex determination

1	e	2	c	3	c	4	b	5	a
6	b	7	b	8	b	9	b	10	c
11	b	12	c	13	d	14	c	15	a
16	b	17	a	18	c	19	a	20	b
21	a	22	a	23	b	24	b	25	b
26	a	27	a	28	a	29	b	30	c
31	a	32	c	33	b	34	b	35	d
36	e	37	c	38	a	39	c		

## Sex linked Inheritance

1	d	2	d	3	d	4	b	5	c
6	c	7	b	8	a	9	b	10	d
11	c	12	e	13	c	14	c	15	d
16	b	17	d	18	c	19	d	20	a
21	b	22	b	23	b	24	a	25	a
26	d	27	d	28	d	29	a	30	b
31	c	32	c	33	d	34	c	35	c
36	b	37	b	38	c	39	a	40	d
41	b	42	b	43	b	44	b	45	a
46	c	47	b	48	d	49	d	50	c
51	d	52	a	53	d	54	c	55	a
56	d	57	a	58	a	59	b	60	a
61	c	62	a	63	c	64	a	65	a
66	b	67	b	68	d	69	d	70	c
71	b	72	d	73	a	74	b		

## Twins &amp; I.Q., Eugenics, Euthenics and Euphenics

1	d	2	c	3	d	4	c	5	d
6	a	7	a	8	d	9	b	10	a
11	d								

## NCERT Exemplar Questions

1	b	2	a	3	a	4	d	5	d
6	b	7	d	8	b	9	d	10	c
11	a	12	c	13	a	14	b	15	b
16	d	17	d	18	c				

## Critical Thinking Questions

1	d	2	a	3	b	4	b	5	b
6	b	7	b	8	c	9	c	10	d
11	a	12	c	13	a	14	c	15	e
16	a	17	b	18	b	19	b	20	d
21	d	22	d	23	e	24	d	25	c
26	d	27	c	28	a	29	a	30	a
31	a	32	b	33	a	34	d	35	a
36	a	37	a	38	b	39	e	40	b
41	d	42	d	43	a	44	c	45	d
46	a	47	d						

## Assertion and Reason

1	c	2	c	3	c	4	a	5	c
6	a	7	a	8	d	9	a	10	a
11	a	12	a	13	a	14	c	15	a
16	a	17	c	18	a	19	b	20	b
21	a	22	a						

## AS Answers and Solutions

## Mendelism

- (a) Gregor Johann Mendel was the first to formulate clear cut laws of heredity.
- (c) Genetics is the study of principles and mechanism of heredity and variations.
- (c) Term genetics was first used by W. Bateson (1905).
- (b) Gregor Johann Mendel (1822–1884 Austria) is known as father of genetics, because he was the first to demonstrate the mechanism of transmission of character from one generation to the other.
- (c) Mendel born in 1822 and died in 1884.
- (d) He was an abbott (head) of Augustinian monastery of St. Thomas at Brunn, Austria in 1847.
- (a) Mendel died before his work could be appreciated by the rest of the scientific community. In 1900, three botanists, Correns of Germany, De Vries of the Netherlands and Tschermak of Austria rediscovered his work after reaching similar conclusions independently.
- (b) The cross of heterozygous dominant with its recessive parent is called test cross. The test cross gives 1 : 1 ratio in monohybrid condition whereas 1 : 1 : 1 : 1 in dihybrid condition.
- (c) Number of gene pair ( $n$ ) = 2  
The number of  $F_2$  genotype =  $3^n = 3^2 = 9$   
Number of kinds of gamete =  $2^n = 4$

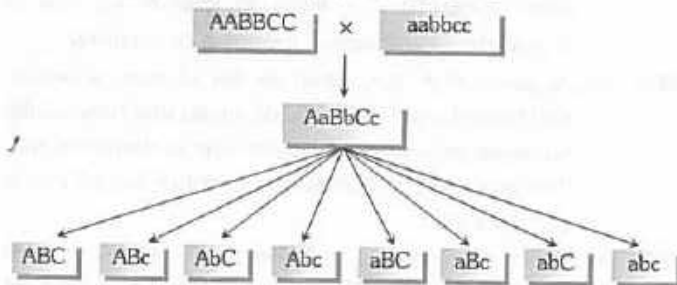
23. (b) Kinds of gametes may be calculated by following formula:

$$\text{Number of gametes} = (2)^n$$

$n$  is number of alleles

**Example :**  $D/d : E/e : F/f$  have trihybrid cross i.e.,  $n = 3$  then

$$\text{Kind of gametes} = (2)^3 = 2 \times 2 \times 2 = 8$$



24. (a) Pod length did not use by Mendel for his experiment.

28. (b) Mendel selected these 7 characters –

- (1) Stem length (2) Flower position
- (3) Pod shape (4) Pod colour
- (5) Seed shape (6) Seed colour (7) Seed coat colour

29. (a) According to Hardy Weinberg principle

$$p^2 + 2pq + q^2 = 1; (p + q)^2 = 1$$

(AA)  $p^2 = 360$  out of 1000 individual or  $p^2 = 36$  out of 100

$q^2 = 160$  out of 1000 or  $q^2 = 16$  out of 100

so,  $q = \sqrt{16} = .4$ . As  $p + q = 1$

so,  $p$  is 0.6.

32. (d) The genotype is the genetic constitution of an organism. Term 'gene' was given by Johannsen (1909) for any particle to which properties of Mendelian factor or determiner can be given.

35. (a)  $RrYy$

$$2 \times 2 = 4 \text{ gametes}$$

(In heterozygous condition 2 gametes and in homozygous condition 1 gamete is produced)

$$R \begin{cases} Y - RY \\ y - Ry \end{cases} ; r \begin{cases} Y - rY \\ y - ry \end{cases}$$

38. (b) Individuals having a pair of contrasting characters are known as allelomorph.

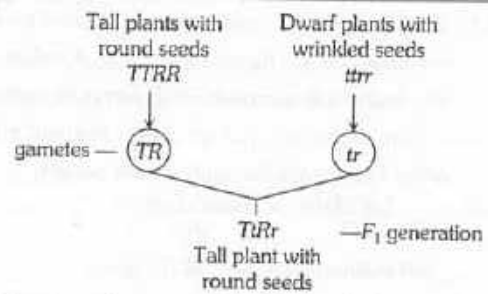
39. (a) Allele is an alternative form of a gene for example,  $T$  and  $t$  alleles represent tall and short stem height of pea plant.

40. (b) In codominance, both the genes of an allelomorph pair express themselves equally in  $F_1$  hybrids, 1:2:1 ratio both genotypically as well as phenotypically in  $F_2$  generation.

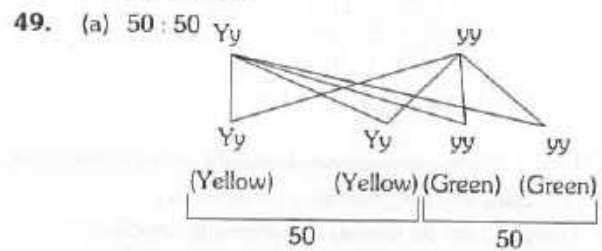
43. (b) Dominant factor : It is one of a pair of alleles which can express itself whether present in homozygous or heterozygous state.

45. (a) The homozygote is pure for the character and breeds true, that is, it gives rise to offspring having the same character on self breeding, e.g.,  $TT$  or  $tt$ .

48. (c)



Therefore, the proportion of dwarf plants with wrinkled seed is zero.



It is called test cross. Which is used to determine the genotype of given offspring.

50. (a) The factor of an allelic pair which is unable to express its effect in the presence of its contrasting factor in a heterozygote is called recessive factor.
54. (a) Genes controlling seven traits in pea studied by Mendel are located on 4 chromosome cotyledon and seed coat colour on chromosome-1; of pod form, flower position and stem length on chromosome 4; of pod colour on chromosome 5 and of seed form on chromosome 7.
55. (d) Yellow coloured pod and wrinkled seed is recessive character according to Mendel.
59. (c) Germplasm can be selected as seed or plantlets for their superior traits.
60. (b) He enunciated three major principles of inheritance i.e., Law of dominance, law of segregation and law of independent assortment.
62. (d) Mendel gave only two laws of genetics. First law is segregation and second law is Independent assortment.
63. (d) Linkage is the tendency of two or more genes to inherit together. Mendel's law are true only in absence of linkage and gene interaction.
65. (d) As allele pairs separate or segregate during gamete formation and the paired condition is restored by random fusion of gametes during fertilization.
67. (b) Because in meiosis chromosomes and DNA amount are transmitted to daughter cell from the parental cell.
68. (a) In this cross  $YYRR$  is responsible for dominant yellow and round seeds and  $yyrr$  for recessive green and wrinkled seeds. Thus in  $F_1$  generation yellow and round seeds are formed.
70. (a) Mendel in his experiment considered total 7 characters (3 characters of seed i.e., seed shape, seed colour, cotyledon colour, 2 characters of pod i.e., pod shape and pod colour and 2 characters of plant i.e., plant height and position of pods on the stem).
71. (c) Test cross include cross of  $F_1$  of the recessive parents i.e., ( $Tt \times tt$ )



73. (c) Test cross : It is a cross to know whether an individual is homozygous or heterozygous for dominant character. The individual is crossed with recessive parent.

78. (c) In plant breeding back cross is performed a few times in order to increase the traits of that parent.

80. (a) Tall Plant  $\times$  Dwarf plant  
 $TT \times tt$   
 (Self Pollination)  $Tt \rightarrow$  Tall ( $F_1$  gen)  

$\begin{matrix} \text{♀} \\ \text{♂} \end{matrix}$	$T$	$t$
$T$	$TT$	$Tt$
$t$	$Tt$	$tt$

 $F_2$  gen.  
 All Tall

81. (c) 1 : 2 : 1; one homozygous dominant, two heterozygous dominant and one homozygous recessive.

82. (d) There will not be formed any recessive combination.

$$Tt \times TT$$

	$T$	$T$
$T$	$TT$	$TT$
$t$	$Tt$	$Tt$

83. (c) A dihybrid test cross give a 1:1:1:1 ratio indicating that two pairs of factors are segregation and assorting independently.

$$TtRr \times ttrr$$

	$TR$	$Tr$	$tR$	$tr$
$tr$	$TtRr$	$Tttr$	$tRr$	$ttrr$

$TtRr$  – Tall Round

$Tttr$  – Tall wrinkle

$tRr$  – Dwarf Round

$ttrr$  – Dwarf wrinkle

1 : 1 : 1 : 1 Phenotypic ratio

84. (b) 9 : 3 : 3 : 1 is dihybrid phenotypic ratio of progeny.

87. (c) 1/8; because in  $F_2$  generation by dihybrid cross 16 hybrids are formed in which only 2 are homozygous for dominant and recessive character. The genotypic ratio for homozygous and heterozygous sets of gene is 2 : 14. It is 1/8 of the 16 hybrids.

88. (b)  $RR \times rr$   
 $\downarrow$   
 $F_1 - Rr$   
 Gametes –  $R, r$

90. (c)  $RR \times rr \rightarrow F_1 = Rr$   
 (Red flower) (White flower)

	$R$	$r$
$R$	$RR$	$Rr$
$r$	$rR$	$rr$

After selfing

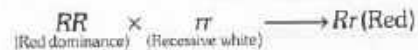
$RR$  = Homozygous for red flower

$Rr$  and  $rR$  = Heterozygous for red flower

$rr$  = homozygous for white flower

3 Red and 1 White = 3 : 1

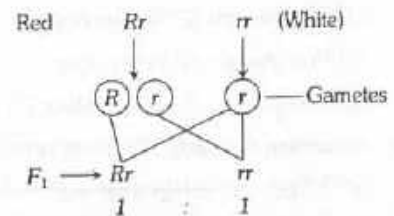
93. (b) All red flowered plants; according to Mendel's law of dominance.



94. (c) In this experiment, Mendel stated that each character is governed by a single gene and there is no linkage and gene interaction. He failed to explain his law of independent assortment in the presence of linkage.

95. (d) All plants of  $F_1$  generation are tall because gibberellic acid treated plant is genetically dwarf and behave like recessive individual and other one is dominant tall, thus according to Mendel's dominance law all plants of  $F_1$  are tall.

98. (a) When heterozygous red (dominant) flower ( $Rr$ ) is crossed with white flower ( $rr$ ), red and white flowered plants will be produced in equal ratios. The cross can be given as -



99. (a) In  $F_2$  gen. of dihybrid cross, the expected genotypic proportion of individuals having both the dominant alleles in homozygous condition is 1/16 and genotypic proportion of individual having both recessive alleles in homozygous condition is 1/16.



$\downarrow$   
 $TtRr - F_1$

$\begin{matrix} \text{♂} \\ \text{♀} \end{matrix}$	$TR$	$Tr$	$tR$	$tr$
$TR$	$TTRR$	$TTRr$	$TtRR$	$TtRr$
$Tr$	$TTRr$	$TTrr$	$TtRr$	$Tttr$
$tR$	$TtRR$	$TtRr$	$ttRR$	$ttRr$
$tr$	$TtRr$	$Tttr$	$ttRr$	$ttrr$

In  $F_2$  generation ( $TTrr$  is 1/16)

100. (d)  $CcPp \times CcPp$   
 Purple White

$\begin{matrix} \text{♀} \\ \text{♂} \end{matrix}$	$CP$	$Cp$	$cP$	$cp$
$Cp$	$CCPp$ Purple	$CCpp$ White	$CcPp$ Purple	$Ccpp$ White
$cp$	$CcPp$ Purple	$Ccpp$ White	$ccPp$ White	$ccpp$ White

Phenotypic ratio– 3 purple : 5 white

103. (b)  $Tt \times Tt$   
Tall Tall

♀	♂	T	t
T	TT	Tt	
t	Tt	tt	

In  $F_2$  - 3 tall : 1 dwarf

This is the law of segregation.

1 Homozygous tall

1 Homozygous dwarf

2 Heterozygous tall

104. (a)  $TT$  is homozygous tall plant,  $Tt$  is heterozygous tall plant and  $tt$  is homozygous dwarf plant.

105. (a)  $AaBB \times aaBB$

Gametes for  $F_1 = AB, aB$  and  $aB, aB$

After crossing =  $AaBB, aaBB$

Ratio = 1 : 1

111. (c)  $AA \times aa - P$

↓  
 $Aa - F_1$

114. (c) The offsprings shows 27 : 9 : 9 : 9 : 3 : 3 : 3 : 1 ratio is found in trihybrid cross.

116. (b) It is the test cross.

117. (d) Which can transmit independently not linked to genes.

### Interaction of gene and cytoplasmic Inheritance

1. (c) The skin shade has to vary from very dark in  $AABBCC$  individual to very light in  $aabbcc$  individual.

2. (d) Dominant epistasis is the phenomenon of masking or suppressing the expression of a gene by a dominant non-allelic gene.

Eg, fruit colour in *Cucurbita pepo* (Summer squash)

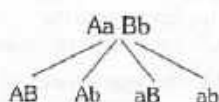
3. (a) The inheritance due to the genes found in cytoplasm (mitochondria and chloroplast) is called cytoplasmic inheritance or non-Mendelian inheritance. The leaves of *Mirabilis jalapa* may be green, white or variegated. This is due to plastid inheritance.

5. (b)  $CCpp \times ccPP$   
White ↓ White  
 $CcPp$   
Red

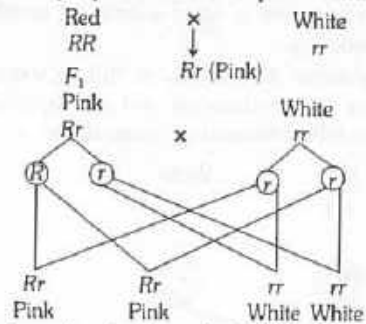
In complementary gene action, 9 : 7 ratio is obtained in  $F_2$  in which two dominant genes are responsible for red flower colour.

9. (a) By the rule of  $2^n$  where  $n$  = Number of Heterozygous pairs of genes

Thus  $2^2 = 4$  type of combinations are formed



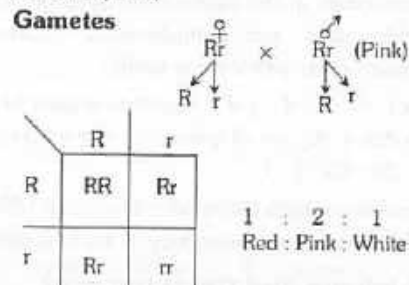
11. (b) *Mirabilis jalapa* shows incomplete dominance.



So, the ratio of pink and white flower will be 1 : 1.

14. (c) Here, both type of traits are transmitting in a single individuals. It means both are dominant at a time or codominant. e.g., blood groups of human being and roan colour in cattle.

15. (c) Parents (Pink)



16. (d) In epistasis, an allele of one gene suppressed or masked the phenotypic expression of a gene at another locus. The gene which is masked is called hypostatic gene.

17. (b) It is an example of Incomplete dominance.

18. (d) Pink colour is due to incomplete dominance.

19. (a) The complementary genes are two pairs of non allelic genes which interact to produce only one phenotypic trait, but neither of them if present alone produces the phenotypic trait in the absence of other.

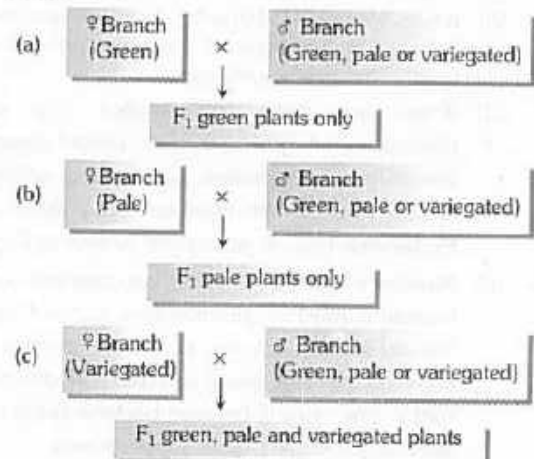
20. (b) Incomplete dominance is found in *Antirrhinum majus* (snapdragon) and *Mirabilis jalapa* (4 O'clock plant).

21. (b) *Mirabilis jalapa* shows incomplete dominance.

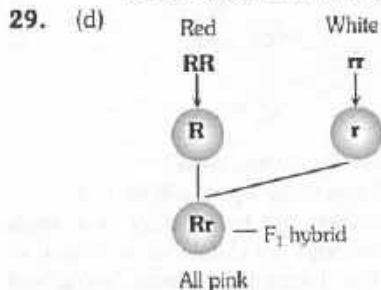
22. (a) The phenotypes of the offspring in this plant depend on the phenotype of the branch that contributes the female gametes.

23. (b) The transmission of traits from parents to offspring by means of plasmagones is known as cytoplasmic inheritance.

24. (b)



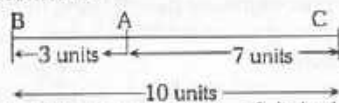
25. (b) Responsibility of a single gene for more than one phenotypic effect, often seemingly unrelated is known as pleiotropy.
27. (d) Cytoplasmic inheritance is due to extranuclear genes present in mitochondria and chloroplasts. These genes do not follow Mendelian inheritance.



30. (c) Certain genes present outside the nucleus or chromosomes in the semiautonomous organelles, such as chloroplast and mitochondria, located in the cytoplasm also control some traits.
32. (b) Ratio 1 : 4 : 6 : 4 : 1 it is due to two pairs of genes. But if it is due to 3 pairs of genes, the size ratio shall be 1 : 6 : 15 : 20 : 15 : 6 : 1.
33. (c) Plasmid is an extra chromosomal circular DNA molecule which replicates independently in the host chromosome.

### Linkage and Crossing over

1. (a) In *Neurospora* it produces 4 types of ascospores two of one parental type, two of other parental type, two of one recombinant and another two of second recombinant type.
3. (b) The tendency of parental combinations to remain together, which is expressed in terms of low frequency of recombinations is called linkage.
4. (d) From the recombination frequencies between different genes A, B and C, it is clear that distance between A and B genes is 3 units, between B and C 10 units and C and A 7 units. Hence linear order of genes on chromosomes is :



5. (d) Crossing over is separation of linked genes (T. H. Morgan).
6. (a) It was Morgan (1910) who clearly proved and defined linkage on the basis of his breeding experiments in fruitfly *Drosophila melanogaster*.
9. (d) When two genes are located very closely on chromosome they are called as "Linked genes" and they inherit together therefore two types of gametes will be formed in maximum numbers. Thus higher number of the parental type off springs will be seen in F<sub>2</sub> generation.
10. (c) Number of linkage groups in an organism is equal to its haploid number of chromosomes. e.g. in *Drosophila* the haploid number is  $n=4$ , so linkage group is 4. But the number of linkage group is prokaryotes (bacteria, like *E. coli*) is one. This is because bacteria being prokaryotes have only a single circular chromosome.

11. (b) Crossing over occurs at pachytene sub-stage of prophase I of meiosis.
12. (b) Mendel's law of independent assortment is not universally accepted and is applied only to genes present on different chromosomes and not to genes present on the same chromosome.
13. (d) Crossing over is a process by which chromatids of homologous exchange portions of genetic material, usually during tetrad formation of meiosis. Crossing over occurs at pachytene sub stage of prophase I of meiosis but visible at diplotene substage of prophase I.
14. (a) T.H. Morgan (1911) proposed the 'chromosome theory of linkage' along with W.E. Castle based on his experiments in *Drosophila*.
17. (a) Because distance between the linked genes is the major factor which controls the frequency of crossing over.
18. (d) During crossing over non-sister homologous chromosome exchange genes.
20. (b) In sweet Pea linkage was firstly discovered.
21. (c) Recon – Unit of recombination  
Muton – Unit of mutation  
Cistron – Unit of function
25. (c) The garden pea plant has seven pairs of chromosome and the same number of linkage groups.
27. (b) Homologous chromosomes moves apart they remain attached to one another at specific points called chiasmata.
30. (b) The synapsis, pairing of homologous chromosomes takes place during zygotene. Synapsis results in the formation of bivalents during zygotene. The formation of special proteinaceous structure called synaptonemal complex occur. After zygotene stage cell enters in pachytene stage in which the bivalents become spiralled, shortened and thickened.
31. (b) In maize  $n=10$ , hence linkage groups = 10.
33. (c) Morgan proposed that chiasmata lead to crossing over by breakage and reunion of homologous chromosomes.

### Chromosomes and Genes

3. (c) Polytene chromosome were discovered by Balbiani (1881) in salivary gland cells of larvae of *Chironomus tentans*.
4. (c) Telocentric are rod shaped chromosomes having sub-terminal centromere. Its terminal end is called telomere.
6. (c) Centromere joins the microtubules of chromosomal spindle fibres and helps in the movement of chromosome during cell division. Centromere also plays an important role in the polymerization of tubulin protein used in the formation of microtubules. Hence it is helpful in attaching the chromosome with spindle fibres.
9. (b) Sperm is haploid structure in which twenty two autosome and one X or Y chromosome are present.
12. (d) Unfertilized human egg is haploid structure in which 22 autosome and one X chromosome are present.



13. (b) Centromere is a region of chromosomes which holds sister chromatids together and the area where chromosome are attached to spindle fibres during cell division.
18. (d) The gene pool can be defined, "as the aggregation of all gene in a mendelian population". Mendelian population is an inter breeding group of population which occurs as a community in an area.
19. (b) The cross over percentage differ for different pairs of genes because the amount of crossing over depend on the frequency with which breaks in the chromatids occur between the two genes. This frequency further depends specially on the distance of these genes which are apart on the chromatids.
22. (b) Polytene chromosomes contain several dark stained regions called bands separated by lighter or less stained area called interbands. These dark bands are heterochromatin (genetically inert) area.
23. (a) In eukaryotes DNA is tightly bound to histones which form a DNA protein particles called nucleosome. Each nucleosome contains 2 copies of each  $H_{2A}$ ,  $H_{2B}$ ,  $H_3$  and  $H_4$  histone protein bounded on 146 BP of DNA. Each nucleosome bead is linked to next nucleosome bead by linker DNA.
24. (a) Each chromosome has a centromere (primary constriction) but in some cases secondary constriction is also present, more specifically called as 'secondary constriction-II'. Its position is constant for particular chromosome heterochromatin is condensed in this region. In man 'secondary constriction-II' is present in chromosome number 1, 10, 13, 16 and Y. Nuclear organizer is called 'secondary constriction-I'.
25. (a) The behaviour of homologous chromosomes during meiosis (their, segregation and independent assortment) and their reunion in fertilization confirmed that the genetic material occurs in the chromosomes and formed the basis of **chromosomal theory of inheritance**.
28. (a) Tizo and Levan (plant cytologists) in 1956 enabled the human geneticists to ascertain the correct chromosome number.
29. (c) Depending upon the position of centromere and relative length of two arms, human chromosomes are of three types- metacentric, submetacentric and acrocentric. The photograph of chromosomes are artificially arranged in the order of descending length is seven group (A to G).
38. (c) Aleurone is triploid and root tip is diploid.
39. (c) The genes present on the differential part of Y chromosome are passed directly from father to son and are called as Holandric genes.
40. (b) The crossing overs occurs in the homologous chromosomes only during the four stranded or tetrad stage in between pachytene and diplotene phase of meiosis.
42. (a) Polytene chromosomes were first observed in the salivary glands of *Chironomous* larva and hence called salivary gland chrom.
48. (c) Idiogram is a composite photograph or diagram of metaphasic chromosomes of haploid or diploid set of an organism arranged in a series of decreasing size, thickness position of centromere and shape.
50. (a) In metacentric chromosome centromere is present in the middle so, these chromosomes are isobrachial. In anaphase the chromosome appear V shaped.
52. (d) The character of the chromosomal complement with reference to the comparative size, shape and morphology of different chromosomes.
59. (d) Polytene chromosomes are multistranded giant chromosomes which are formed by somatic pairing of homologous chromosomes and their repeated replication or endoduplication to form 1000 (e.g. salivary of gland of *Drosophila*) to 16000 (e.g. chironomous) chromonemata.
61. (b) Linkage is the pattern of assortment of genes that are located on the same chromosome. It is important because, if the genes are located relatively far apart, crossing over is more likely to occur between them than in they are located close together. In sex linkage, the speciality is criss-cross inheritance.
63. (c) The lampbrush chromosomes are found in oocytes of many vertebrates (Amphibian, reptiles birds) and some invertebrates (insects). They also occur in the giant nucleus of the unicellular algae *Acetabularia*.
64. (a) Euchromatin or dynamic chromatin is relatively expended and open. It has the potential of being actively transcribed.
65. (a) Number of linkage groups in an organism is equal to the haploid number of chromosomes. In *Drosophila melanogaster*,  $2n = 8$ , hence  $n = 4$  linkage groups.
66. (b) Centromere (Kinetochore) lies in the region of primary constriction where two chromatids are connected at the centromere.
67. (c) Polytene chromosomes was described by Kollar (1882) and first reported by Balbiani (1881). They are found in salivary glands of insects (*Drosophila*) and called as salivary gland chromosomes.
68. (d) Carmine is a dye extracted from the cochineal insect (*coccusacti*).
70. (c) Crossing over always takes place between nonsister chromatids of two homologous chromosomes.
71. (a) The reduction of DNA content does not occur in meiosis-I. Truly haploid nuclei in terms of DNA contents as well as chromosomes number are formed in meiosis-II. When the chromatids of each chromosome are separated into different nuclei. Thus meiosis-II is necessary.
72. (c) Human male has 44 autosomes and two different sex chromosomes viz X & Y.
73. (a) DNA is the hereditary material, which is located inside the chromosome.
74. (b) Man is heterogametic because it has X and Y chromosomes.
75. (b) Caspersen (1970), stained chromosomes with a fluorescence dye it gives different banding patterns helpful to know various types of chromosomal aberrations.
76. (b) E. strasburger (1875) discovered these distinct structures during cell division.

77. (b) DNA is known as genetic material.
79. (b) The actual distance between two genes is said to be equivalent to the percentage of crossing over between these genes i.e. 66%. Crossing over chances between y and b genes suggest that these are to be placed on the chromosome at a distance of 66 units.
80. (a) Chromosome number is fixed for a species. The lowest number is seen in *Haplopappas gracilis* i.e.  $2n = 4$  and maximum in ophioglossum species i.e.  $2n = 1656$ .
82. (c) Because males have only one X chromosome, Y being without alleles. Therefore, even single recessive allele expresses its effect in males.
87. (b) Allele frequency is the relative proportion of a particular allele among individuals of a population. According to Hardy-weinberg equation, the frequency of dominant and recessive alleles in a population will remain constant from generation to generation if there is no mutation, selection, random drift and migration. As per Hardy-weinberg equation.
- $$p^2 + 2pq + q^2 = 1$$
- $p$  = dominant allele frequency  
 $q$  = recessive allele frequency  
 $p^2$  = homozygous dominant genotype  
 $2pq$  = heterozygous genotype  
 $q^2$  = homozygous recessive genotype
- Here,  $p = 0.6$  and  $q = 0.4$   
 Therefore, heterozygotes frequency is  
 $= 2pq = 2 \times 0.6 \times 0.4 = 0.48$
93. (c) Hofmeister discovered nuclear filaments in the nuclei of the pollen mother cells of the plant, *Tradescantia*, in 1848.
94. (c) Multiple alleles being located on the same locus do not show crossing over.
95. (d) Jumping genes or transposons genes are found in both prokaryotes and eukaryotes. These were discovered by Mc clintock in case of maize.
98. (c) Wood cock (1973) observed the structure of chromatin under electron microscope. He termed each beaded structure on chromosome as nucleosome.
99. (c) Chimeras are the individual having the different genotypes in its different parts.
103. (c) Gene control protein synthesis through controlling the synthesis of a specific protein and it controls heredity through transmission of heredity characters from one generation to another.
104. (b) Gene is capable of duplication of its genetic material by faithful replication and its precise distribution among new cells by cell division.
105. (d) These are small proteins responsible for coiling DNA in to nucleosome.
106. (d) Histones are main structural protein found in eukaryotic cells.
109. (d) Woodcock termed each beaded structure on chromosome as nucleosome. So nucleosome are units of chromosomes.
112. (b) Holandric genes are the Y-linked genes. The Y-linked type sex-linked inheritance is performed by those genes which are localized in the non-homologous sections of Y chromosome and that have no alleles in X-chromosome.
113. (d) *Neurospora* (ascomycetes fungus) which is also called *Drosophila* of plant kingdom.
114. (c) In viruses of animals, DNA is present and in viruses of plants, RNA is present as genetic material.
116. (b) Both the chromatids move towards opposite poles due to repulsive force called anaphasic movement.
117. (b) A gene is a segment of DNA which contains the information for one enzyme or one polypeptide chain coded in the language of nitrogenous bases or the nucleotides.
118. (a) This theory was given by Beadle and Tatum (1958), while they were working on red mould or neurospora.
120. (c) Jumping genes are also called 'Transposons'.
121. (d) Cistron, muton and recon were given by Seymour Benzer to explain the relation between DNA length and gene.
123. (b) Genes determine the physical as well as physiological characteristics. These are transmitted from parents to the offsprings generation after generation.
124. (c) DNA and proteins (Polynucleotides and histone proteins).
128. (c) Unwanted portion (introns) of genes is spliced off and remaining part i.e., exons get joined by ligase.
129. (b) According to this theory one gene controls the synthesis of one polypeptide chain and not of the complete enzyme or protein molecule.
131. (a) Based on the location of centromere the chromosomes are categorised in to (i) Telocentric (ii) Acrocentric (iii) Submetacentric (iv) Metacentric.

### Multiple allelism

3. (a) The skin colour of a person is the result of an interaction between two pairs of genes.
4. (b) The difference between Rh positive and Rh negative depend on a single pair of genes ( $r$ ) with the gene responsible for the Rh positive condition dominant (RR, Rr)
6. (c) ABO system is one of the most important human "blood group systems. The system is based on the presence or absence of antigens A and B on the surface of red blood cells and antibodies against these in blood serum. A person whose blood contains either or both of these antibodies cannot receive a transfusion of blood containing the corresponding antigens as this would cause the red cells to clump.

Phenotypes and genotypes of ABO blood groups are

Phenotype	Genotype
A	$I^A I^A$ or $I^A i$
B	$I^B I^B$ or $I^B i$
AB	$I^A I^B$
O	$ii$

9. (e) ABO blood groups are controlled by a series of three multiple alleles. Different combination of these alleles produce four blood types A, B, AB and O.
15. (d) Antisera D (also called D Antigen) is used for testing to the blood group of Rh, because Antisera D containing antibody against protein D.
17. (d) Karl Landsteiner of USA. He was awarded Nobel prize for the same in 1930.
19. (c) The process of mating among closely related individuals is known as inbreeding.
21. (c) Alleles for blood group B (of child) are absent in father (blood group A), therefore they should come from mother and they are present in persons having blood group B or AB.
23. (c) Person with blood group AB has both A and B antigens on RBC but no antibodies in the plasma.
24. (d) Erythroblastosis foetalis disease is related to the birth of a child related with Rh factor. It cause the death of the foetus within the womb or just after birth.
25. (d) Heterozygous  $I^A I^B$  individual are blood group AB. Both the A antigen (Product of the  $I^A$  alleles) and the B antigen (Product of the  $I^B$  alleles) are produced by them.
28. (c) Rhesus monkey (*Macaca rhesus*).
29. (c) A person with blood group AB possess both the antigen A and B but their blood plasma does not possess any of the antibodies.
30. (b)  $Rh^-$  woman married with  $Rh^+$  man, become sensitized simply by carrying a  $Rh^+$  child within her body. Some of the cells from the embryo may mix into her own blood stream during development. The first child of the parents with this genetic background is nearly always normal.
31. (d) O individuals produce neither A or B antigen, but have both type of antibodies. So their blood can be transfused into any recipient.
37. (b) Agglutinin A and B is a specific antigen present normally at surface of R.B.C.
38. (d) The person with blood group O is said to universal donor, because in this, there are no antigens on the surface of RBC.
42. (d) Since the blood groups of parents are A and B, therefore their genotypes are AO and BO so the possible genotypes of their children are AO, BO, AB and OO i.e. the possible blood groups are A, B, AB and O.
44. (a) When the blood group of one parent is AB and that of other is O then the child may have blood group either 'A' or 'B' as blood group 'O' has no dominant genes for antigens.
45. (d) Since the blood group of father is AB (genotype AB), therefore one of the two antigens i.e. A or B has to be present in child's blood i.e. he can have any other blood group but not 'O' (genotype OO).
47. (d) Stage I of blood clotting is concerned with the formation of thromboplastin released from damaged tissue or platelets. Thromboplastin helps in the formation of the enzyme thrombokinase.
51. (d) Rh factor was first of all reported in RBCs of *Macaca rhesus* (rhesus monkey) by Landsteiner and Wiener in 1940. It is found only in man and rhesus monkey and is not reported from other animals.
52. (a) The Rh factor causes erythroblastosis foetalis, when a woman who is  $Rh^-$  develops antibodies against her foetus, which is  $Rh^+$ . The symptoms of the disease are caused by agglutination of the baby's red blood cells.
57. (c) IgG is the most abundant long-acting antibody representing about 80 percent of the antibody. It is able to pass across the placenta from mother of child.
58. (b) Codominance is a condition in which heterozygote where both members of an allelic pair contribute to phenotype, which is then a mixture of the phenotypic traits produced in either homozygous condition. In cattle the cross of red X white produces roan offspring whose coat consist of both red and white hair.
59. (b) In erythroblastosis foetalis, the antibodies in the blood stream of the mother, pass through the placenta and cause damage to the red cells of the foetus.
60. (a) Blood group in human are inheritable trait and are inherited from parents to offsprings on the basis of mendel's law.
62. (c) If a man with blood B, marries a woman with blood group A then, genotype will be of B blood group child.

♀ \ ♂	$I^A$	$I^B$	$I^O$
$I^A$	$I^A I^A$	$I^A I^B$	$I^A I^O$
$I^B$	$I^B I^A$	$I^B I^B$	$I^B I^O$
$I^O$	$I^O I^A$	$I^O I^B$	$I^O I^O$

63. (b)

♀ gametes	♂ gametes	
	$I^A$	$I^B$
$I^A$	$I^A I^A$	$I^A I^B$
$I^B$	$I^B I^A$	$I^B I^B$

### Genetic variation

5. (c) Albinism is an autosomal recessive trait. The genotype of such mother would be 'Aa' i.e. one recessive gene 'a' responsible for the absence of melanin and one dominant gene responsible for the presence of melanin would be present. So the mother would be heterozygotic carrier.
9. (c) Chromosomal aberrations, genetic drift and recombination all play role in bringing genetic variations but recombination is more common.
10. (a) Because germplasm DNA is inherited from one generation to another.
16. (a) Phenylketonuria is homozygous autosomal recessive disorder.
18. (c) Mutation are sudden stable inheritable/transmissible discontinuous variations which appear in organism due to permanent change in their genotypes.
20. (d) Individuals with Turner syndrome have one X chromosome ( $HH + XO$ ) due to non-disjunction of sex chromosome in their parents.



22. (a) The chromosome number in Turner's syndrome is  $2n = 45$  due to fusion of (ZZ + O) ovum with gynospem (ZZ + X).
26. (a) In transition, purine replaces purine and pyrimidine replaces pyrimidine and in transversion, a purine is replaced by pyrimidine or vice versa.
29. (b) The individuals with this syndrome are trisomic for chromosome 21. Chromosomal complement is  $46 + \text{additional chromosome } 21 = 47$ .
31. (b) Phenotypically these individuals are males, but they can show some female secondary sexual characteristics and are usually sterile.
33. (c) There is a large scale possibility of autosomal aneuploidy in human beings.
40. (c) Meiotic non-disjunction.
42. (d) De Vries observed a number of mutation in his experimental plant evening primrose, *Oenothera lamarckiana*.
43. (b) Because father can give only Y-chromosome to their son.
45. (d) White eye mutation in drosophila, result in depigmentation in many other parts of the body giving a pleiotropic effect.
47. (d) Sickle cell Anaemia is a hereditary disorder of autosomal nature which is caused by mutation of the gene controlling  $\beta$ -chain of haemoglobin.
49. (b) Because AIDS is caused by Virus HIV. HIV also known as LAV = Lymphadenopathy associated virus.
51. (d) Natural selection will preserve the adaptive mutations.
52. (c) In man, enzyme tyrosinase (also inhibited) by excess phenylalanine) is absent due to homozygous recessive autosomal alleles. Melanin or pigment formation from dihydrophenylalanine is stopped. There is lack of pigment in skin, hair and iris.
55. (a) The idea of mutations was brought forth by Hugo de Vries, who worked on evening primrose.
56. (b) It has an abnormal karyotype of  $10 \times 2n + 1 (18) = 47$
60. (c) Continuous variation are small and graded these include variation in colour, shape size, weight and structure of body parts, Height. These variations may be somatogenic or blastogenic.
64. (d) Change in single base pair of DNA is also a type of mutations called point mutations
69. (d) Multivalents is an association of more than two homologous chromosomes. The number of multivalents depends upon the degree of synapsis and chiasmata formation among similar chromosomes.
74. (a) Down's syndrome is due to trisomy of 21<sup>st</sup> chromosome was first reported in 1866 by Langdon Down.
75. (d) Mutation induced by a mutagen are called as induced mutations.
76. (a) Mutation causes discontinuous variations. Gene duplication is a type of chromosomal mutation and it has been noticed in giant chromosomes of salivary glands of *Drosophila*.
77. (d) Epicanthus is the symptom of Down's syndrome characterised by folding of skin (epicanthus) at the inner part of the eyes.
79. (d) Sickle cell trait is a heterozygous condition in which an organism may have one gene for normal haemoglobin and other gene for sickle cell haemoglobin.
80. (a) Colchicine inhibits the cell division or mitosis, but duplication of chromosomes is continue, as a result, diploid becomes tetraploid.
81. (d) Suppressor mutation totally or partially restores a function lost by a primary mutation and is located at a genetic site different from the primary mutation.
82. (a) Polydactyly or extra fingers is caused due to autosomal dominant gene.
83. (a) Klinefelter's syndrome is a human sex abnormality, arises due to non-disjunction of sex chromosomes during meiosis. It is characterised by the chromosome number.  $2n = 47$ , the chromosomal formula is  $44A + XXY$ .
- Klinefelter's syndrome is a male in general appearance; testes are underdeveloped, enlarged breast (gynaecomastia), Mentally defective and abnormally tall. Mental retardation increases with X complement. The karyotypes of extreme Klinefelter's syndrome are  $44A + XXXY$ ,  $44A + XXXXY$ ,  $44A + XXXYY$  etc.
- In Turner syndrome, chromosome number is  $2n = 45$ . The chromosomal formula is  $44A + XO$ . She is a sterile female.
- $22A + XY$  is chromosome constitution of normal male individuals which are heterogametic.
- $22A + XX$  is chromosome constitution of normal female individuals, are homogenetic.
84. (b) Philadelphia chromosome is 22<sup>nd</sup> chromosome. It is so called because in 1960 investigation in philadelphia found that patients with chronic myeloid leukaemia have an abnormal chromosome later found to be 22<sup>nd</sup>.
86. (c) Polyploidy is the phenomenon of having more than two genomes or set of chromosomes e.g.  $3n$ ,  $4n$ ,  $5n$  etc.
89. (c) In inversion a piece of chromosome is removed and rejoined in reverse order.
92. (a) They occur in 1/700 births in women aged 25 years or under. The frequency increases with age to about 1/100 for women of age 40 and 1/10 for women of age 45.
96. (a) The condition of chromosomal change which involve the loss or gain of single chromosome is called aneuploidy. Genotype "22 pairs + XXY males" show the condition of Klinefelter's syndrome.
97. (a) "Cri-du-chat" syndrome condition is due to a deletion in the short arm of the chromosome number 5.
100. (b) Turner's syndrome is 23 monosomic (Aneuploidy) conditions, in which only one X chromosome is present no Y chromosome.
103. (b) On the basis of principles of simple recessive inheritance, the probability that their second child will also be an albino is 100%.

109. (d) Turner's syndrome is caused due to the absence of one of the X chromosomes i.e. 45 with XO (or 44 + XO).

112. (d) The sixth amino acid in  $\beta$ -chain of normal haemoglobin is glutamic acid. In sickle cell haemoglobin this amino acid is replaced by valine.

### Sex determination

2. (c) The female contain two X chromosomes. The eggs are produced by the meiosis i.e., reduction division. So the egg contains one X chromosome when released from ovary. After fertilization the diploid phase is restored.
3. (c) 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.
4. (b) Also called sex chromosome or heterosomes.
5. (a) Most flowering plants are monoecious and so do not have sex chromosomes. Sex chromosome has been reported in two plant species namely *Melandrium* (*Lychnis*) and *Sphaerocarpus*.
7. (b) The barr body is present in the neutrophil (polymorphonuclear leucocytes) of 3 to 5% cells in females, but not in males.
8. (b) The ratio of X and A chromosomes responsible for determination of sex in drosophila e.g. Superfemale – Number of X chromosome XXX/Set of autosome AA.  
Sex index ratio  $(X/A) = \frac{3}{2} = 1.5$  Super female.
11. (b) Haplodiploidy is a type of sex determination in which the male is haploid while female is diploid. It occurs in some insects like honey bees, ants and wasps.
14. (c) Super or meta females have 47 (44 + XXX), 48 (44 + XXXX) or 49 (44 + XXXXX) chromosomes.
16. (b) More than one X-chromosome in females is transformed into Barr bodies and drumsticks.
19. (a) Amniocentesis is the most widely used method for prenatal detection of many genetic disorders.
20. (b) Sex is determined at fertilization by the nature of the sperm that fertilizes the egg.
23. (b) Y-bearing sperms produce male embryo X-bearing sperms produce female.
24. (b) Barr body is nothing but 'X' chromosome which has become heterochromatic, thus appear as deeply stained body. Of the two 'X' chromosomes are remains normal while the other appears as a Barr body.
25. (b) Down's syndrome is a chromosomal abnormality in humans and is associated with an extra chromosome 21 (i.e. chromosomal formula is  $45 + XX = 47$ ), while one of the two X-chromosomes becomes genetically inert and heteropycnotic and forms the Barr body. Hence human female suffering from Down's syndrome contains  $(2X - 1 = 1)$  one Barr body.
26. (a) In *Drosophila*, occasionally flies are obtained in which a part of the body exhibits female characters and the other part exhibits male as gynandromorphs. These are formed due to misdivision of chromosomes and start as female with  $2A + 2X$  chromosomes.

28. (a) Gynandromorphs 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  $2A + X$ .

30. (c) Barr body is characteristic feature of female.

34. (b) Human male has  $44 + XY$  and without barrbody; It is associated with X chromosome of female, if male has one barrbody then it has XXY.

35. (d) Chromosomal theory of sex determination was proposed by an american Mc clung (1902) based on chromosome study of grasshopper.

37. (c) Random genetic drift in a population probably result from interbreeding within the small isolated population.

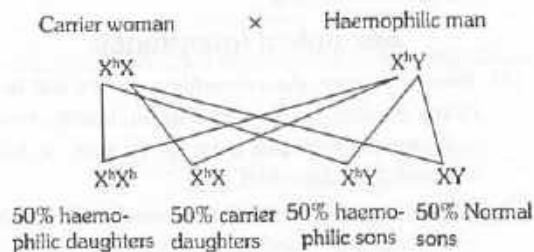
38. (a) In *Melandrium* (the garden pink) a variety of garden flower, sex is determined by XX-XY type or lygaeus type of chromosomes just as in animals.

39. (c) X/A ratio determines sex in *Drosophila*; ♀ is heterogametic (ZW) in birds. In 'XO type' the 'O' determines maleness.

### Sex linked inheritance

1. (d) Since daughter of a colourblind person will be a carrier of the disease and only one of the two X chromosomes will bear the recessive gene for disease, so half of their sons will be colourblind.
2. (d) When both the chromosomes bear genes for disease i.e. in homozygous condition, only then the character will be expressed otherwise the individual will act as a carrier.
3. (d) Because colour blindness is a recessive sex-linked disease, genes of which are present exclusively on non-homologous segment of X chromosome, so males need only one affected chromosome to express the character whereas females need both the affected chromosomes to exhibit the same.
4. (b) Genes for sex linked characters occurs in both segments of X and Y-chromosomes. Many sex linked characters (about 120) are found in man. Such character are mostly recessive.
5. (c) Y-linked disorders are caused by mutations on the Y-chromosome. Because males inherit a Y-chromosome from their fathers, every son of an affected father will be affected. Because females inherit an X-chromosome from their fathers, female offspring of affected fathers are never affected.
6. (c) Carrier mother is heterogametic for recessive colour blindness gene.
15. (d) A person whose father is colourblind i.e. person is normal because colour blind genes located on X-chromosome of father and it is transferred into only its daughters. This person marry with a lady whose mother is daughter of colour blind father i.e. lady is carrier for colour blind. Thus normal man marries with a carrier lady and their sons will be some normal and some colour blind.

18. (c) Haemophilia is more prevalent in men as compared to women. Its recessive gene is located on X-chromosome. This gene suppresses synthesis of factor VIII, necessary for the normal blood clotting.
19. (d) Both are recessive sex linked diseases.
22. (b) Since woman is normal and her father was a bleeder means she is a carrier and as she marries a victim of haemophilia so only half of their children will be diseased.
29. (a) Cystic fibrosis, Thalassaemia and haemophilia are the hereditary disease.
30. (b) Chance of a boy inheriting haemophilia solely depends upon the mother's nature irrespective of his father's nature. If mother is heterozygous for disease, chances are 50% and if she is homozygous similarly, then all his sons will inherit the disease.



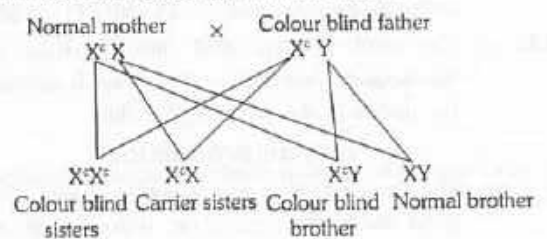
31. (c)

XX Normal women		X <sup>c</sup> Y Colourblind man	
$\begin{array}{c} \text{♀} \\ \text{X} \end{array}$	$\begin{array}{c} \text{♂} \\ \text{X} \end{array}$	X <sup>c</sup>	Y
X	X	XX <sup>c</sup>	XY
X	X	XX <sup>c</sup>	XY

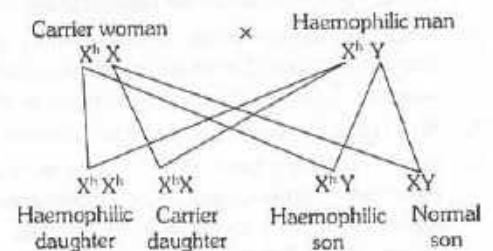
The daughters of this couple will normal eye sight and carrier if one of the carrier daughter marries with normal eyed man.

32. (c) Sickle-cell anaemia is a genetic disease reported from negroes due to a molecular mutation of gene  $Hb^A$  on chromosome 11 which produces the  $\beta$  chain of adult haemoglobin.
33. (d) The concept of sex-linked inheritance was introduced by Tomas H. Morgan in 1910, while working on *Drosophila melanogaster*.
37. (b) All the sons of a colourblind mother are colourblind.
38. (c) Sex influenced genes and sex limited genes are located on autosomes. While sex linked genes are located on X and Y chromosomes.

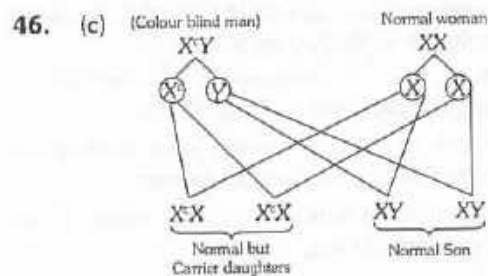
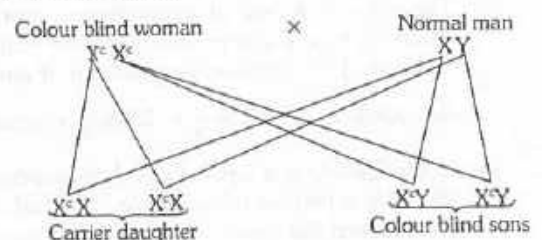
39. (a) Father colour blind and mother normal.



41. (b) Half Haemophilic and half carriers.



44. (b) The marriage between normal visioned man and colourblind woman will produce colour blind sons and carrier daughters.

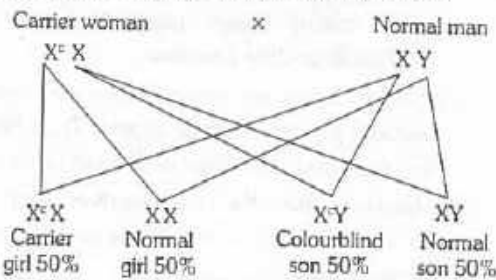


So all sons in the progeny will be normal.

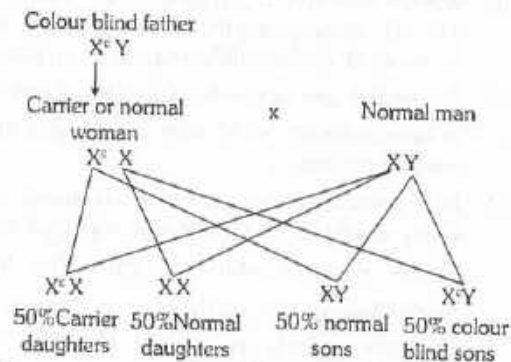
47. (b) Infact the girl with normal vision is carrier because her father is colourblind (daughter of colourblind father are either colourblind or carrier) and when she marries a normal man the possibility of their sons being colourblind is 50% because the genotype of parents is  $X^c X$  and  $XY$ , so only half of the possible combinations of  $XY$  have the X-linked recessive genes which exhibit the disease.
49. (d) Sex linkage is the transmission of characters and their determining genes alongwith sex determining genes which are found on the sex chromosomes. Y chromosome of male carries a few genes but X chromosome which is common to male and female carries a number of genes.



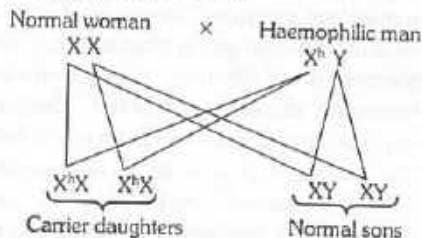
51. (d) A woman whose father was colourblind is actually normal phenotypically but carrier of colourblindness if this woman marrying with normal man. Then



55. (a) Daughters normal, 50% of sons colour blind.



56. (d) For a woman to be colourblind both her X chromosomes should bear genes for the disease; which is possible only when her father is colourblind and her mother is either colourblind or a carrier of disease.
57. (a) Brachydactyly is a disease characterized by small sized finger and is due to dominant gene on the autosome.
59. (b) All offsprings will be normal.



63. (c) Colour blindness is X linked recessive disease, influencing man in heterozygous condition and woman in homozygous condition.
64. (a) Colour blind person is unable to distinguish primary colour viz red and green.
65. (a)  $aa \times Aa$
67. (b) Gene for colour blindness is located on the nonhomologous part of X-chromosome. It is a sex linked recessive disease.
70. (c) Sex limited genes express their effects in only one sex and their action is clearly related to the sex hormones. For e.g. beard and moustaches development in human beings is a sex limited character.
71. (b) Colourblindness is known as X linked recessive disease.
72. (d) Haemophilia is X linked recessive disease, clotting factor of blood is mutated and found in Royal family of England.

73. (a) In the pedigree analysis the inheritance of a particular trait is represented in the family tree over generation.

### Twins & I.Q. Eugenics, Euthenics and Euphenics

- (d) They are also known as dizygotic twins.
- (c) Identical twins are produced when one fertilized egg divides into two blastomeres and both give rise to young ones.
- (d) Homozygotic individuals are 'true-breeder' when crossed with each other they produce only one type of gametes because they possess two of the same alleles for a particular trait.
- (c) 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 etc.
- (d) The defective or undesirable persons who possess inherited defects or diseases (haemophilia, colourblindness etc. and epilepsies, feeble, idiocy etc.) should not be permitted to reproduce so that the unwanted genes are gradually eliminated from the gene pool of human population.
- (a) Eugenics is the study of possibility of improving humanity by altering its genetic composition by encouraging breeding of those presumed to have desirable genes and discouraging breeding of those presumed to have undesirable genes.
- (a) Eugenics is the branch of science which deals with improvement of human race genetically.
- (d) Conjoined or siamese twins arise by incomplete separation of embryo at about 15 days or more after zygote formation.
- (b) Identical or monozygotic twins arise from a single zygote formed by fertilization of a single egg with a single sperm. Such twins are members of a clone and have the identical genotype. These arise by the separation of two or more blastomeres derived by mitotic divisions of original zygote.
- (d) Dizygotic also known as fraternal in which twins are formed by simultaneous fertilization of two different ova by two different sperms.

### Critical Thinking Questions

2. (a) Anaphase divides the paired hereditary factors into two equal and similar halves.

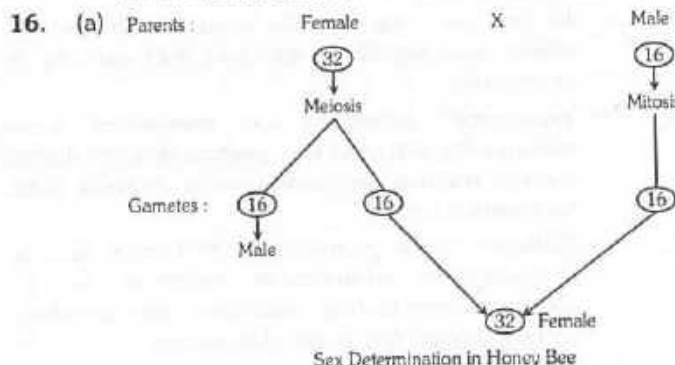
6. (b)  $XX \times Xx$  ..... Parents

↓  
 $XX Xx Xx xx$  .....  $F_1$  hybrid

In  $F_1$  generation half of the total offsprings represent parental genotype i.e.,  $Xx$ . Therefore out of 1200 seedling 600 will have parental genotype.

9. (c) Incomplete dominance shows the 1 : 2 : 1 phenotypic and genotypic ratio.
10. (d) Because mitochondrial genes also show maternal inheritance because all the mitochondria a zygote has come from the cytoplasm of the ovum.
11. (a) 20 chromosomes; because in meiosis cell division chromosome number becomes halved.

12. (c) Haploid in germinal cell, diploid in somatic cell and triploid in endosperm cells.
13. (a) Since a typical green plant is diploid, therefore it has two sets of chromosome. So the number of genome will be two because genome is a set of chromosomes.



18. (b) Cat-cry or cri-du chat syndrome is caused by the deletion of short arm of chromosome number 5 ( $5p^-$ ). So, the genotype of affected individual is  $46, XX, 5p^-$  in female and  $46, XY, 5p^-$  in males.
19. (b) Frequency of crossing over is proportional to the distance of gene.
20. (d) Sir Archibald Edward Garrod was an English physician, who pioneered the field of inborn errors of metabolism. He was born on November 25, 1857, in London and died on March 28, 1936, in Cambridge.
21. (d) When a cluster of gene show linkage behaviour they do not show independent assortment because they are located very close to each other.
22. (d) During embryonic development, the primary organizer signals the development of another organ or tissue by secreting chemical factors.
24. (d) Female is larger. Many offsprings are produced from single mating.
25. (c) Translocation is a kind of chromosomal rearrangement in which a block of genes from one linkage group is transferred to another linkage group.
26. (d) Reduction division takes place during spermatogenesis. Spermatids and sperms will have haploid number of chromosomes.
27. (c) According to Radial loop model. Each chromosome has one or two interconnected scaffolds made of non histone chromosomal proteins. The scaffold bears a large member of lateral loop all over it. 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.
30. (a) The extra Y-chromosome is strongly male determining. The extra Y-chromosome leads to over production of male hormone, which causes over aggressiveness. So XYY men are prone to violence, criminality and antisocial behaviour.
31. (a) Recessive characters are expressed in the subsequent generation only when present on the X chromosome of male.

32. (b) Marriages between close relatives and cousins is not advisable because recessive lethals are carried in heterozygous conditions and express themselves only when mating takes place between two carrier individuals or close relatives.
37. (a) Identical twins are formed when one sperm fertilizes one egg to form a single zygote. They have the same genotype and phenotype and are of same sex.
38. (b) Mutation alter the configuration and position of nucleotides which is irreversible process except reverse or back mutation.
40. (b) When a man with blood group "AB" marries a woman with "O" blood group then blood group of children will be 'A' or 'B' which is differ from parental blood group.
42. (d) The mother can be true but father is not exactly true.
44. (c) In human beings, curly hairs is dominant and straight hair is recessive.
45. (d) In transition, a nitrogen base is replaced by another of its type i.e. one purine is replaced by another purine ( $A = G$ ) while one pyrimidine by another pyrimidine ( $C = T$  or  $U$ ).
46. (a) The term euploidy ( $eu = \text{good} + \text{ploid} = \text{multiple}$ ) is applied to organisms with chromosome numbers that are multiples of some basic number. e.g.,  $x, 2x, 3x, 4x, 5x, 6x$ , etc.

### Assertion and Reason

1. (c) The cells of the plants regenerated from cell cultures (callus) show heritable variation for both qualitative and quantitative traits, such a variation is known as somaclonal variation. These variations arise as a result of structural changes in chromosome, gene mutations, plasmagene mutations, gene amplification, altered expression of multigene families, transposable elements or mitotic crossing over. So, (A) is true but (R) is false.
2. (c) The sex of the child to be born is initially governed by the chromosomal contribution of the father. All chromosomes are paired, and the Y is dominant over the X chromosome when combined with it. A child conceived will therefore be subject during development thereafter to a predisposition towards femaleness if receiving an X chromosome from the mother and an X chromosome from the father (XX), or towards maleness if receiving an X chromosome from the mother but a Y chromosome from the father (XY). Therefore sex in human is a monogenic not polygenic.
- Polygenic traits or continuous traits are those traits that are determined by the combined effect of more than one pair of genes. An example of this is human stature. The combined size of all of the body parts from head to foot determines the height of an individual. There is an additive effect. The sizes of all of these body parts are, in turn, determined by numerous genes. Human skin, hair, and eye color are also polygenic traits because they are influenced by more than one allele at different loci. The result is the perception of continuous gradation in the expression of these traits.

3. (c) Haemophilia is a blood disorder where the blood does not clot normally. Haemophilia A is the most common form, caused by a deficiency of blood clotting factor VIII. Haemophilia B is due a deficiency of blood clotting factor IX. Haemophilia is an hereditary disorder. In almost all cases, it is males that suffer the condition, although it is passed on by both females and males carrying the gene. Queen Victoria was a carrier of haemophilia A. It is also known as classical haemophilia and factor VIII deficiency haemophilia. Haemophilia B is also known as Factor IX haemophilia, and Christmas disease after the first patient diagnosed with it. Prothrombin producing platelets in such persons are not found in very low concentration.
4. (a) The first chemical mutagen discovered was mustard gas. C. Auerbach and her associates first discovered the mutagenic effects of mustard gas and related compounds during World war II. These compounds are examples of a large class of chemical mutagens that transfer alkyl ( $\text{CH}_3$ ,  $\text{CH}_3\text{CH}_2$  etc.) groups to the bases in DNA, thus are called alkylating agents.
5. (c)
6. (a) Carolus Linnaeus called humans as *Homo sapiens* or wise men and placed them along with apes and monkeys. There are certain similarities between human and chimpanzee. These are –
  - (i) RNA content of diploid cells is similar.
  - (ii) DNA matching shows that human similarity is 100% with chimpanzee and
  - (iii) Banding pattern of chromosomes shows very little difference in chromosomes 3 and 6 between humans and chimpanzee. Thus it can be concluded that among the primates, chimpanzee is the closest relative of the present day humans.
7. (a) Pollen is formed by meiosis in pollen mother cells. Hence, chromosome number will be reduced to half in pollens.
8. (d) Clones are asexually produced. These have same genotype and phenotype like parent.
9. (a) Back cross is a cross which is performed between hybrid and one of its parents. In plant breeding, back cross is performed a few times in order to increase the traits of that parent.  
For example, a crop plant is crossed with a wild variety in order to obtain its disease resistance. In the process most good traits of the crop plant get diluted. The hybrid is, therefore, repeatedly crossed with parent crop plant in order to transfer the good traits back into it.
10. (a) According to Mendel's concept of inheritance, each gene had two alternative forms or allelomorphs, one being dominant and the other recessive. Practically, the wild form can mutate in several ways. The mutant form can also mutate once again to give rise to another mutant form. Therefore, a gene can have more than two allelomorphs. These allelomorphs make a series of multiple alleles.
11. (a) Phenylketonuria is due to deficiency of liver enzyme Phenylalanine hydroxylase which converts phenylalanine into tyrosine. It occurs in person who are homozygous recessive. It results with a high level of phenylalanine in blood, tissue fluids and urine.
12. (a) Genotype is the hereditary properties of an organism and genetic complement of organism is called genotype.
13. (a) Holandric genes are always found on Y chromosomes of male. Hence, their inheritance is always from father to son.
14. (c) Haemophilia never occurs in women because it is a recessive gene disease if both the sex chromosome  $\text{X}^h\text{X}^h$ , it will be lethal i.e. such female die before birth. Its gene is never found on chromosome Y.
15. (a) Mutations may be either recessive or dominant. In haploid organisms like viruses and bacteria, both recessive and dominant mutations can be recognized by their effects on the phenotype of the organisms in which they originated. Therefore, in several crops desirable mutants have been isolated among haploids derived in culture.
16. (a) The position of centromere is fixed for a chromosome. It may lie at the centre (metacentric chromosome), near the centre (submetacentric) subterminal (acrocentric chromosome) or terminal (telocentric chromosome) position. During anaphase the chromosome bends in the region of centromere. Depending upon the position of centromere, an anaphasic chromosome can have V-shape, L-shape, J-shape or I-shape.
17. (c) Genetic analyses indicate that heterochromatin is largely genetically inactive. Most of the genes of eukaryotes that have been extensively characterized are located in euchromatic regions of the chromosomes. The earlier belief that no genes are found in heterochromatic regions is not correct, because genes could be located in heterochromatic regions in several cases like *Drosophila* and tomato. The genes in heterochromatic region perhaps become active for a short period.
18. (a) The surface of centromere bears a special trilaminar plate called kinetochore. Kinetochore has points for attachment of microtubules. Microtubules produce chromosome fibres or tactile fibrils require for the movement of chromosomes during anaphase.
19. (b)
20. (b) The lampbrush chromosomes are highly elongated special kind of synapsed mid-prophase or diplotene chromosome bivalents which have already undergone crossing over. They occur in diplotene stage of most animal oocytes, spermatocytes of many and even giant nucleus of unicellular alga *Acetabularia*. The number of pairs of loops gradually increase in meiosis till it reaches maximum is diplotene.
21. (a) Most sex – linked genes in male heterogametic animals are present on the X- chromosome. Some animals, however, may carry a few genes on the Y chromosome that produce visible effects on the phenotype of the organism. Y linkage is very rare in higher animal, particularly mammals. X linkage, on the other hand, is very common in all mammals that have been studied, the mammalian X chromosome contains a larger number of genes with major effects on phenotype.
22. (a) Chromosomes are stained with special fluorescent dyes that have differential affinity for different parts of the chromosomes. It brings about specific banding pattern. Bands are segments of stained chromosomes that appear lighter, darker or stained as compared to adjacent parts. Banding technique of chromosome staining is highly useful in knowing various types of chromosomal aberrations or abnormalities like additions, deletions and inversions.



## Principles of Inheritance and Variation

## SET Self Evaluation Test

- 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]
  - Migration
  - Mutations
  - Random drift
  - Selection
- Identify the correct order of organisation of genetic material from largest to smallest [AIPMT 2015]
  - Genome, chromosome, nucleotide, gene
  - Genome, chromosome, gene, nucleotide
  - Chromosome, genome, nucleotide, gene
  - Chromosome, gene, genome, nucleotide
- Electroporation procedure involves [AIIMS 2005; DUMET 2009]
  - Fast passes of food through sieve pores in phloem elements with the help of electric stimulation
  - Opening of stomatal pores during night by artificial light
  - Making transient pores in the cell membrane to introduce gene constructs
  - Purification of saline water with the help of a membrane system
- The total hereditary material outside the chromosome is called as [MP PMT 2011]
  - Plasmagene
  - Plasmon
  - Muton
  - Recon
- Freemartin is an example of [MP PMT 1997, 2000]
  - Hormonal control of sex
  - Sex reversal by gene
  - Environmental control of sex
  - None of the above
- Phenotype of an organism is the result of [CBSE PMT 2006]
  - Environmental changes and sexual dimorphism
  - Genotype and environment interactions
  - Mutations and linkages
  - Cytoplasmic effects and nutrition
- Which one of the following blood group systems is determined by genes on the X chromosome [MP PMT 1997]
  - Yt
  - ABO
  - Xg
  - MNSs
- Phenylketonuria (PKU) is an inherited disease which refers to
  - Decrease in phenylalanine in tissue and blood
  - Increase in phenyl pyruvic acid in tissue and blood
  - Elimination of sugar in urine
  - Elimination of gentisic acid in urine
- Antigen - A and antibody - B are present in which blood group [CPMT 1995; KCET 1999]
  - B
  - A
  - AB
  - O
- Which one of the following statements about the particular entity is true [CBSE PMT (Mains) 2010]
  - Centromere is found in animal cells, which produces aster during cell division
  - The gene for producing insulin is present in every body cell
  - Nucleosome is formed of nucleotides
  - DNA consists of a core of eight histones
- In *Drosophila* female has a pair of chromosomes [MP PMT 2011]
  - ZZ
  - XX
  - XY
  - ZW

## AS Answers and Solutions

1	c	2	b	3	c	4	b	5	a
6	b	7	c	8	b	9	b	10	b
11	b								

- (c) The random changes in gene frequencies occurring by chance and not under the control of natural selection are called genetic drift.
- (c) Electroporation method are used for introduction of recombinant DNA into the host. In this procedure temporary pores are formed in the plasma membrane of the host cell. These pores permit entry of foreign DNA.
- (a) Free martin is an examples of early influence of hormone on sex determination. Lillie and others found that when twins of opposite sex (One male and other female) are form, the male is normal but female is sterile with many male characteristics. Such sterile female is known as free martin.
- (b) Phenylketonuria is the genetically inherited disease. It occurs due to deficiency of an enzyme phenylalanine hydroxylase which catalyses the conversion of amino acid phenylalanine into tyrosin. This results in the increased level of phenylalanine in the blood at the same time it starts depositing in various body tissue.
- (b) 'Insulin' gene is found in every body cell but is not expressed in all cells.