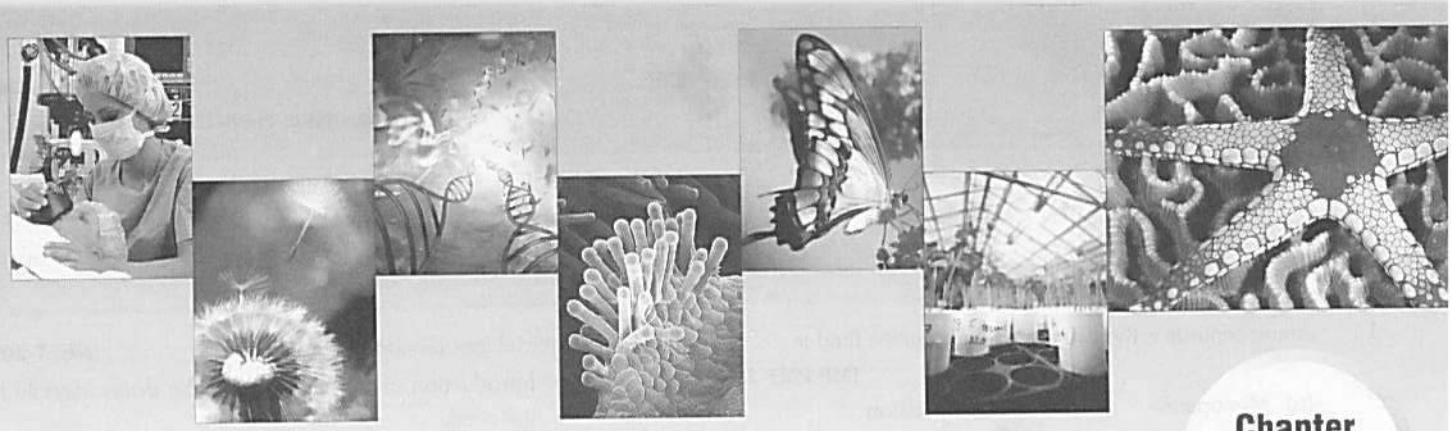


Chapter

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 twins9) 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 /*Drosophila* genetics	Morgan
Father of human & physiological genetics	Garrod
Father of quantitative inheritance	Kolreuter
Father of *Neurospora* 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

(i) Traits have two alternative forms.

(ii) 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_2 Generation (Bateson, 1905) : F_2 or second filial generation is the generation of individuals which arises as a result of inbreeding or interbreeding amongst individuals of F_1 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.

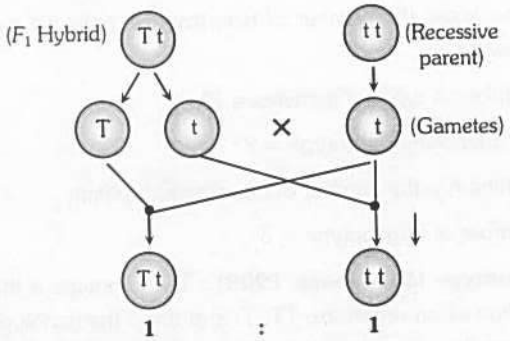


Fig : 7.1-1 Showing test cross

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 monestry 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.

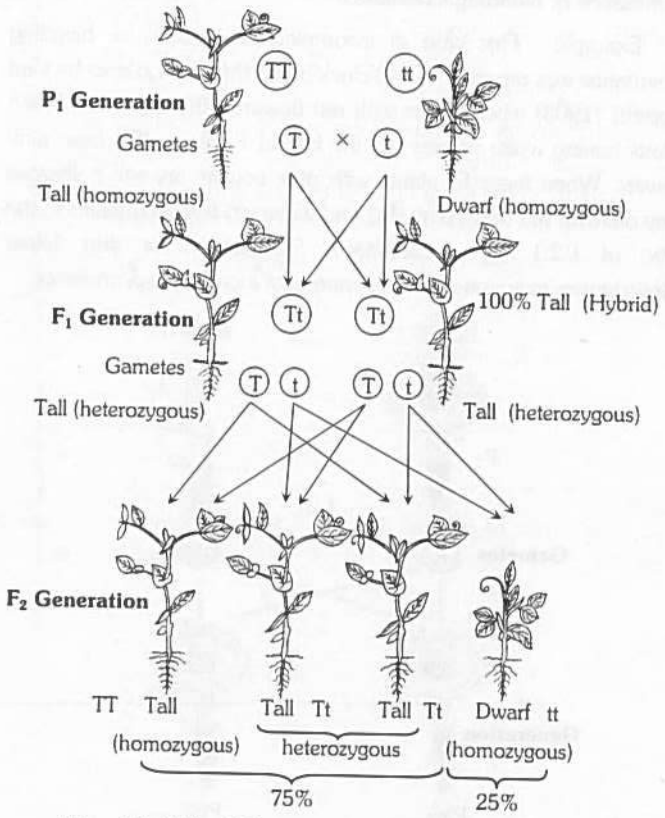


Fig : 7.1-2 Mendel's monohybrids crosses between tall and dwarf pea plants

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

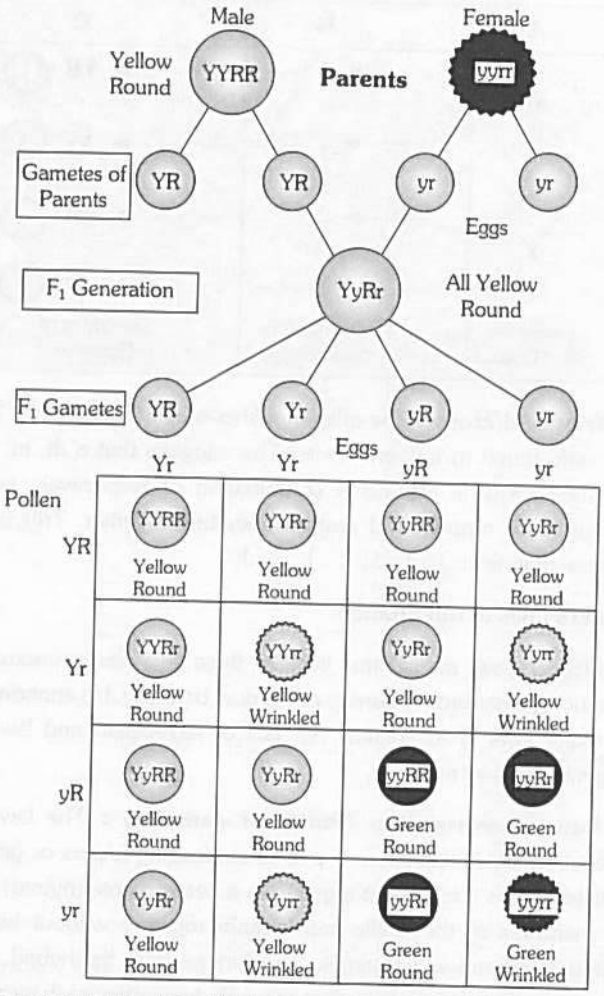


Fig : 7.1-3 Mendel's dihybrid cross between pea plants having yellow round seeds and green wrinkled seeds

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)
	r →	Yr (2)
y →	R →	yR (3)
	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.

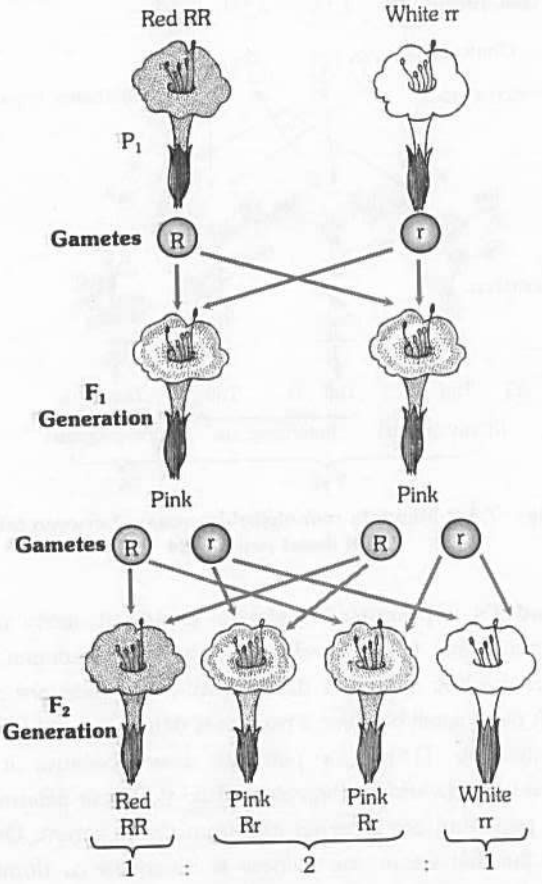


Fig : 7.1-4 Incomplete dominance of flower colour in *Mirabillis jalapa*

(ii) **Codominance (1:2:1 ratio) :** In codominance, both the genes of an allelomorphic 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.

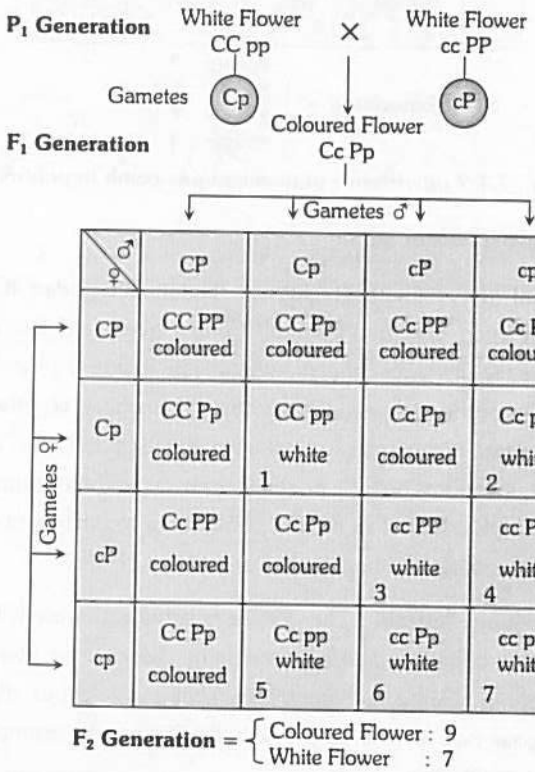


Fig : 7.1-5 The results of an experiment to show the operation of complimentary genes in the production of flower colour in sweet pea (*Lathyrus*)

(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.

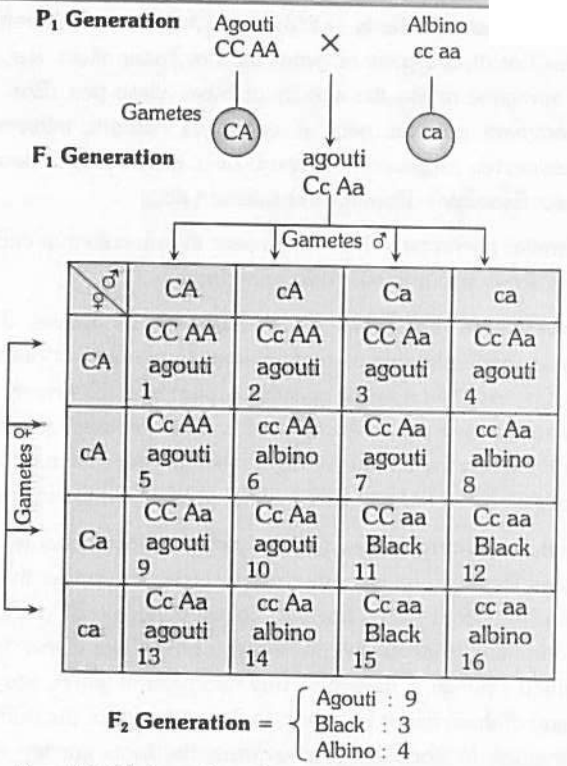


Fig : 7.1-6 Interaction of supplementary genes in mice for coat colour

(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.

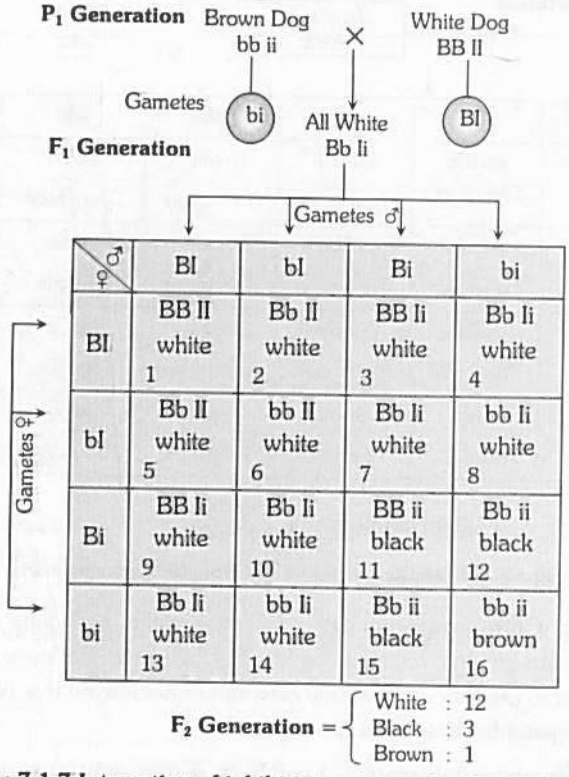


Fig : 7.1-7 Interaction of inhibiting genes in dog for coat colour showing dominant epistasis

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.

P_1 Generation Triangular AABB × Top Shaped aabb

F_1 Generation AaBb × Triangular AaBb

$\rightarrow F_2$

♀ / ♂	AB	Ab	aB	ab
AB	AABB Triangular	AABb Triangular	AaBB Triangular	AaBb Triangular
Ab	AABb Triangular	AAbb Triangular	AaBb Triangular	Aabb Triangular
aB	AaBB Triangular	AaBb Triangular	aaBB Triangular	aaBb Triangular
ab	AaBb Triangular	Aabb Triangular	aaBb Triangular	aabb Top shaped

F_2 Generation = Triangular : 15, Top shaped : 1

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.

P_1 Generation Rose Comb RR pp × Pea Comb rr PP

Gametes Rp, rP

Walnut comb Rr Pp

F_1 Generation

Gametes ♂ / Gametes ♀

♀ / ♂	RP	Rp	rP	rp
RP	RR PP walnut 1	RR Pp walnut 2	Rr PP walnut 3	Rr Pp walnut 4
Rp	RR Pp walnut 5	RR pp rose 6	Rr Pp walnut 7	Rr pp rose 8
rP	Rr PP walnut 9	Rr Pp walnut 10	rr PP pea 11	rr Pp pea 12
rp	Rr Pp walnut 13	Rr pp rose 14	rr Pp pea 15	rr pp single 16

F_2 Generation = Walnut : 9, Rose : 3, Pea : 3, Single : 1

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

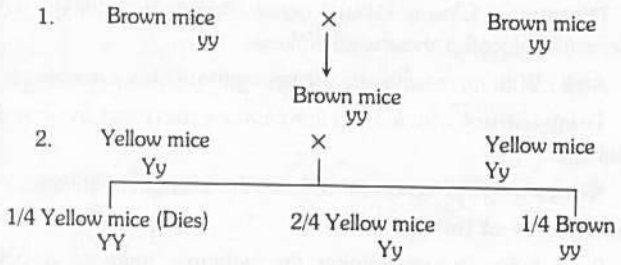
Pleiotropic effect of genes

Lethal genes : Lethal factor were first of all reported in mice body by French geneticst '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).

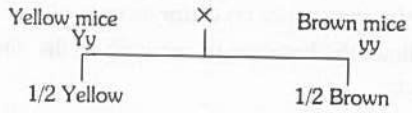
Dominant lethals : The dominant lethal genes are lethal in homozygous condition and produce some defective or abnormal phenotypes in heterozygous condition. Their most serious effect in heterozygous may also cause death. Following are the examples of dominant lethal genes.

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, Stiegleder concluded that yellow mice are heterozygous. The homozygous yellow ($1/4^{th}$ of the total offsprings) dies in the embryonic condition. When there 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.

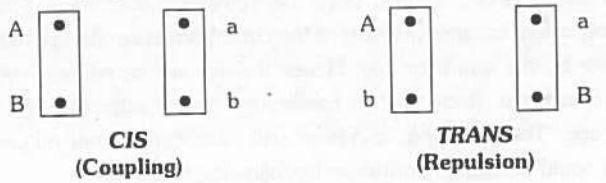
Theories of linkage

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 Punnet (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).



CIS (Coupling) *TRANS* (Repulsion)

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 *Drosophilla* $n=4$, 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 Cattle. 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.

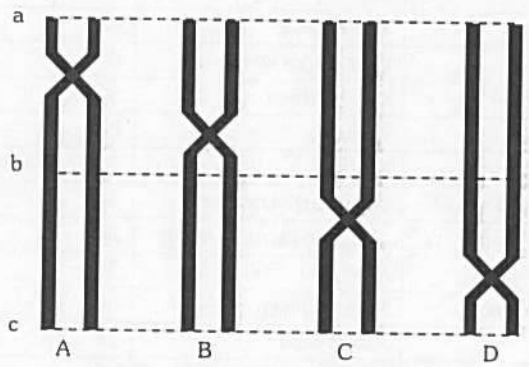


Fig : 7.1-11 Diagram showing possibilities of crossing over between genes at different distances

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. It's occurrence during the act of meiosis produces variations in the heritable characters of the gametes.

(v) This phenomenon has also found it's 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

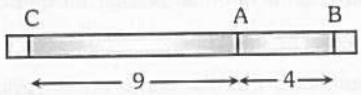
A linkage or genetic chromosome map is a linear graphic representation of the sequence and relative distances of the various genes present in a chromosome. A chromosome map is also called a linkage map or genetic map.

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.*, chiasma % = 2 (cross over %) or crossover %= 1/2 (chiasma %).

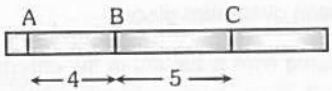
Accordingly, Sturtevant, 1911 prepared the first chromosomal map. Infact this map is a line representation of a chromosome where the location of genes has been plotted as points at specific distances. These distances are proportional to their crossing over percentage. Suppose there are three genes on 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 (*Aulocantha*) 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	*Amoeba proteus*	500
Man	*Homo sapiens*	46
Gorilla	*Maccaca mulatta*	48
Pig	*Sas scrofa*	40
Sheep	*Ovis aries*	54
Cat	*Felis maniculata*	38
Dog	*Canis familiaris*	78
Rat	*Rattus rattus*	42
Rabbit	*Oryctolagus cuniculus*	44
Honey bee	*Apis mellifera*	32, 16
Mosquito	*Culex sp*	6
Grasshopper	*Gryllus*	23(M), 24(F)
Pink bread mould	Neurospora crassa	14
Baker's yeast	*Saccharomyces cerevisiae*	34
Broad bean	*Vicia faba*	12
Garden pea	*Pisum sativum*	14
Onion	*Allium cepa*	16
Maize	*Zea mays*	20
Potato	*Solanum tuberosum*	48

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

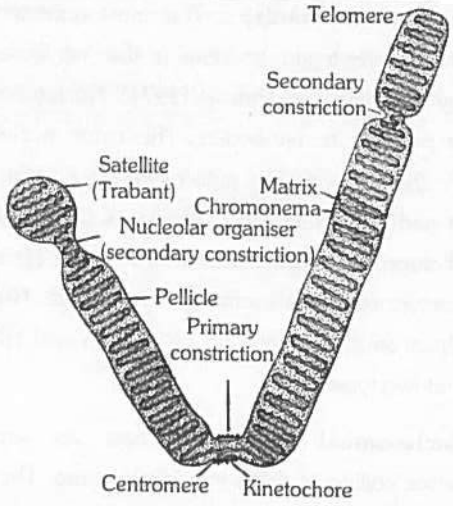


Fig : 7.1-12 Structure of chromosome

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.

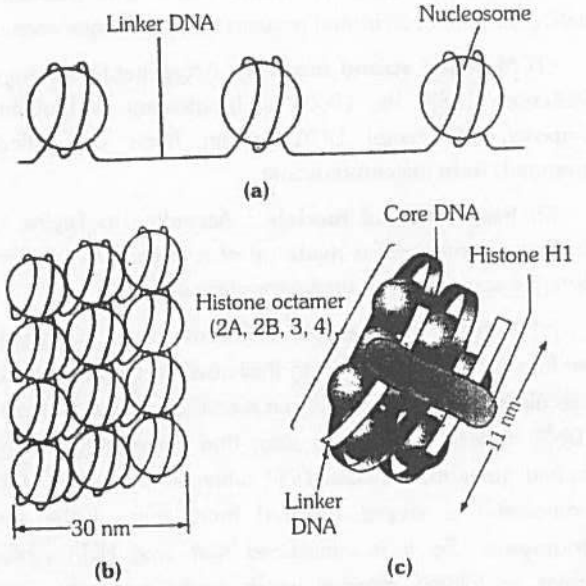


Fig : 7.1-13 Nucleosomes (a) 3 Nucleosomes (b) Nucleosomes coiled to form a solenoid (c) Basic structure of a nucleosome

(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
Q (quinacrine) banding	Chromosomes exposed to quinacrine mustard (acridine dye) which preferentially binds to AT-rich DNA. Other fluorescent dyes used are *DAPI or Hoeschst* 33258.	UV fluorescence reveals fluorescencing *Q* bands which correspond to G-bands. DNA of Q/G bands contains more closely spaced SARs, giving tighter loops (Q loops).
G (Giemsa) banding	Chromosomes treated with alkaline solution and subjected to controlled trypsin digestion before staining with Geimsa, a DNA banding chemical dye. Relatively permanent stain.	Dark bands are called G bands and pale bands are G-negative. G bands are presumed to be *AT-rich*. They are late replicating and contain highly condensed chromatin.
R (reverse) banding	Chromosomes treated with heated saline or restrictase to denature AT-rich DNA and stained with *Giemsa*. GC-specific chromomycin dyes, *e.g*, chromomycin A, olivomycin or mithracin give the same pattern.	R-banding pattern is essentially the reverse of the *G*-banding pattern. R bands are Q negative. They generally replicate in the S-phase and have less condensed chromatin.
T (telomeric) banding	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 inetnsely staining. They are especially concentrated at the telomeres.
C (centromere) banding	Chromosomes pre-treated with sodium hydroxide or barium hydroxide and stained with Giemsa.	Preferred darkening of constitutive centromeric heterochromatin. Rest of the chromosome show Q banding pattern.

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 23rd 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 phytohaemaglutinin (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.

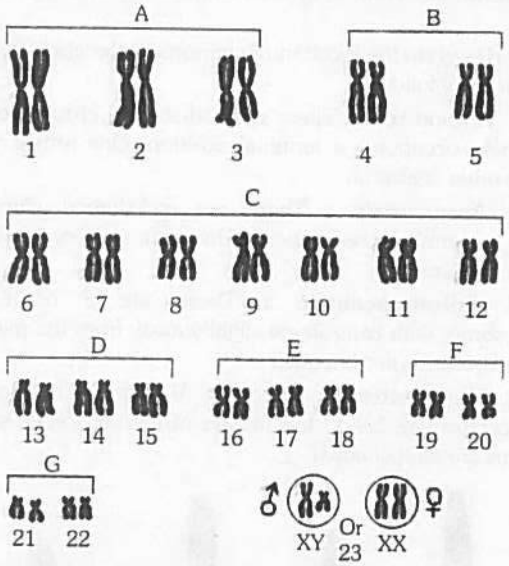


Fig : 7.1-14 Human karyotype

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.

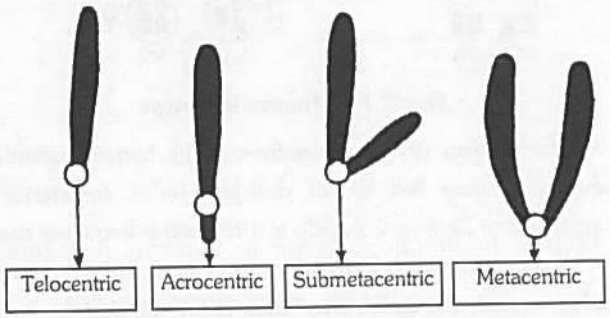


Fig : 7.1-15 Types of chromosomes

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 2000μm.

The chromosome is formed by somatic pairs between homologous chromosomes and repeated replication or endomitosis of chromonemata. These are attached to chromocentre. It has pericentromeric heterochromatin. Polytene chromosomes show a large number of various sized intensity bands when stained.

The lighter area between dark bands are called interbands. They have puffs bearing *Balbiani rings*. Balbani rings produce a number of *m*-RNA, which may remain stored temporarily in the puffs. These also occur in Malpighian tubules, rectum, gut, foot pads, fat bodies, ovarian nurse cells etc.

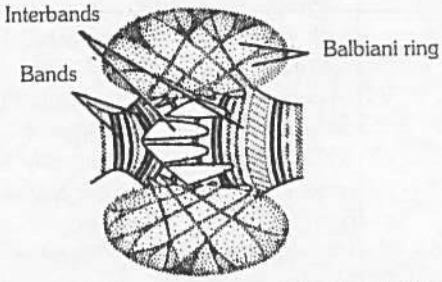


Fig : 7.1-16 Polytene chromosome showing balbiani ring

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 5900μm. These are found in pairs consisting of homologous chromosomes jointed at chiasmata (meiotic prophase-I). The chromosome has double main axis due to two elongated chromatids. Each chromosome has rows of large number of chromatid giving out lateral loops, which are uncoiled parts of chromomere with one-many transcriptional units and are involved in rapid transcription of *m*RNA meant for synthesis of yolk and other substances required for growth and development of meiocytes. Some *m*RNA produced by lampbrush chromosome is also stored as informosomes *i.e.*, *m*RNA coated by protein for producing biochemicals during the early development of embryo. Length of loop may vary between 5-100μm.

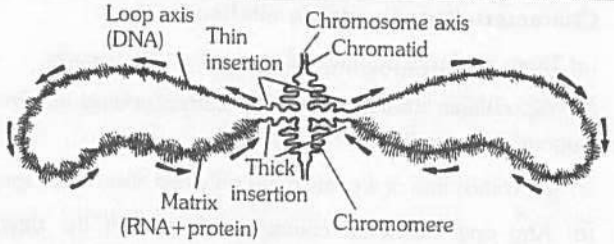


Fig : 7.1-17 A part of main axis with a pair of lateral loops of a lampbrush chromosome showing synthesis of RNA

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 synapses 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.

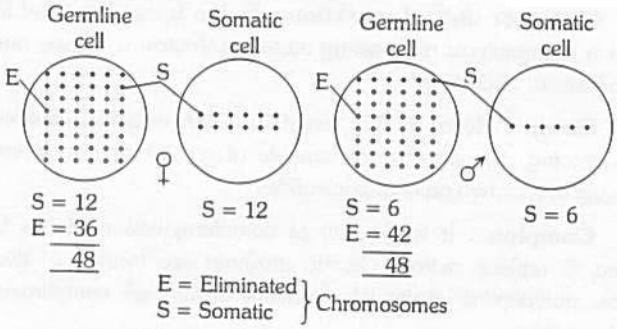


Fig : 7.1-18 Schematic representation of the S and E chromosomes of the gall insect *Miastor*

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 cistrom 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 suppresive.

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.

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

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

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

Table : 7.1-9 Genotype of blood groups in man

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

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

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

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

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

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	$I^o I^o \times I^o I^o$	O	A, B, AB
O and A	$I^o I^o \times I^a I^o$	O, A	B, AB
A and A	$I^a I^o \times I^a I^o$	O, A	B, AB
O and B	$I^o I^o \times I^b I^o$	O, B	A, AB
B and B	$I^b I^o \times I^b I^o$	O, B	A, AB
A and B	$I^a I^a \times I^b I^b$ $I^a I^a \times I^b I^o$ $I^a I^o \times I^b I^o$	O, A, B, AB	None
O and AB	$I^o I^o \times I^a I^b$	A, B	O, AB
A and AB	$I^a I^o \times I^a I^b$	A, B, AB	O
B and AB	$I^b I^o \times I^a I^b$	A, B, AB	O
AB and AB	$I^a I^b \times I^a I^b$	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^oI^o	O	A, B	AB
		A	O, B	
		B	O, A	
A	I^AI^o, I^AI^A	O, B	A, AB	O, B
B	I^BI^o, I^BI^B	O, A	B, AB	O, A
		A	B, AB	O, A
AB	I^AI^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 recipient gene (r). People having this antigen with genotype (RR or Rr) are called Rh positive (Rh^+) and those whose blood is devoid of it with genotype (rr) are Rh negative (Rh^-). About 85% human beings in Europe and 97% in India are Rh^+.

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

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 alongwith the mother's blood on reaching the foetal circulation cause clamping of foetal RBCs or agglutination reaction. The first child is some how born normal because by that time the number of antibodies in mother's blood remain lesser but they increase with successive pregnancies.

Thus the foetus following the first child dies either within the womb or just after its birth. This condition is known as erythroblastosis foetalis. So a marriage between Rh^+ boy and Rh^- girl is considered biologically incompatible.

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.

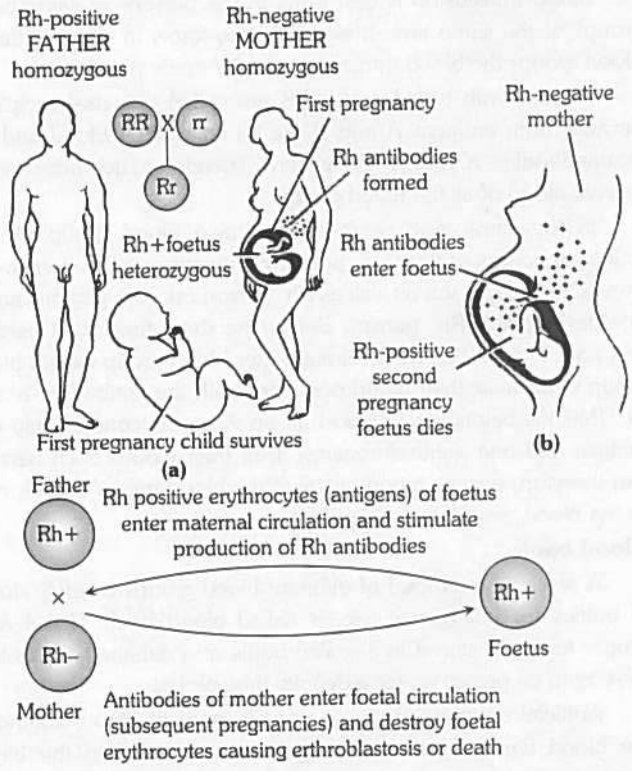


Fig : 7.1-19 Foetal death in the womb due to erythroblastosis foetalis

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^z, r', r", r^y and r. The alleles denoted by capital letter give rise to Rh^+ condition while those denoted by small letter to Rh^- condition. Rh^+ condition is dominant over Rh^- condition. Thus Rh^+ person may be homozygous (RR) or heterozygous (Rr) while Rh^- persons are always homozygous(rr). Hereditary trait for Rh^- factor is inherited according to Mendelian principle.

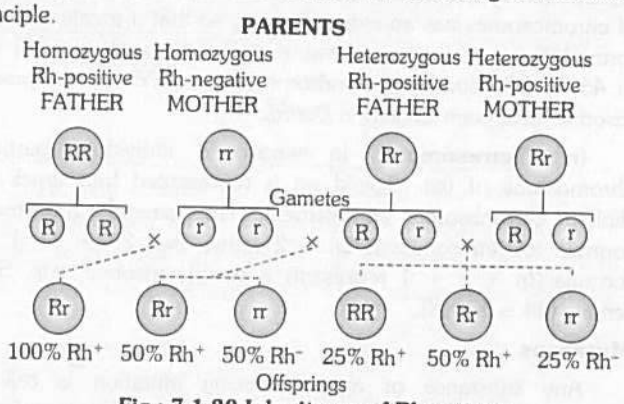


Fig : 7.1-20 Inheritance of Rh antigen

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.

GC → AT or AT → GC

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

GC → CG or TA , AT →TA or CG

(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.

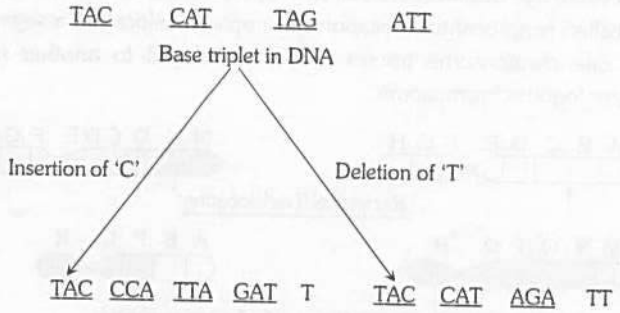


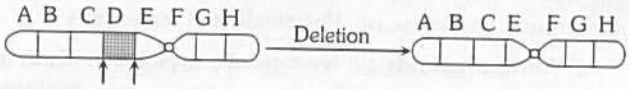
Fig : 7.1-21 Frame shift mutations

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**

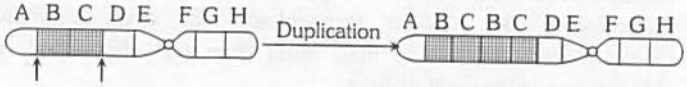
Deletion or deficiency : Sometimes a segment of chromosome break off and get lost. If a terminal segment of a chromosome is lost, it is called deficiency. Deficiency generally proves lethal or semilethal. If intercalary segment is lost it is termed deletion.



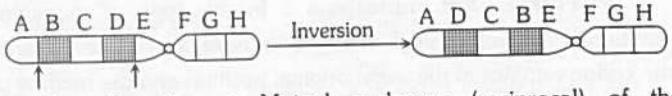
Deletion occurs during pairing in meiosis. For example in human babies deletion of a segment of chromosome number 5 causes a disease called *cri-du-chat* syndrome (the baby cries like a cat and is mentally retarted with small head).

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.

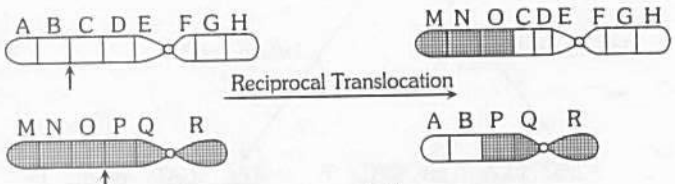
Duplication : In this mutation deleted chromosomal segment is attached to its normal homologous chromosome. Here a gene or many genes are repeated twice or more times in the same chromosome.



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

(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 tobacum*. 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 tuberoseus*.

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 chromsome 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 2*n* + 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) :** α-particles, β-rays, protons and neutrons.

(ii) **Ionizing (non particulate) :** X-rays, r-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 β 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 β chain of adult haemoglobin. The mutated gene Hb^S produces sickle-cell haemoglobin. The sixth amino acid in β chain of normal haemoglobin is glutamic acid. In sickle-cell haemoglobin this amino acid is replaced by valine. The children homozygous (Hb^SHb^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^AHb^A genotype are normal, those with the Hb^SHb^S genotype have sickle-cell disease, and those with the Hb^AHb^S genotypes have the sickle-cell trait. 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 β chain. Their haemoglobin contains δ 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 estrogen. 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 $2n + 1 (21^{st}) = 47$.

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 turcicus 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 sinuta* and *Vallisneria spiralis*.

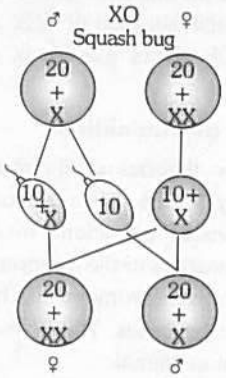


Fig : 7.1-22 Protenor type of sex determination in Grasshopper

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.

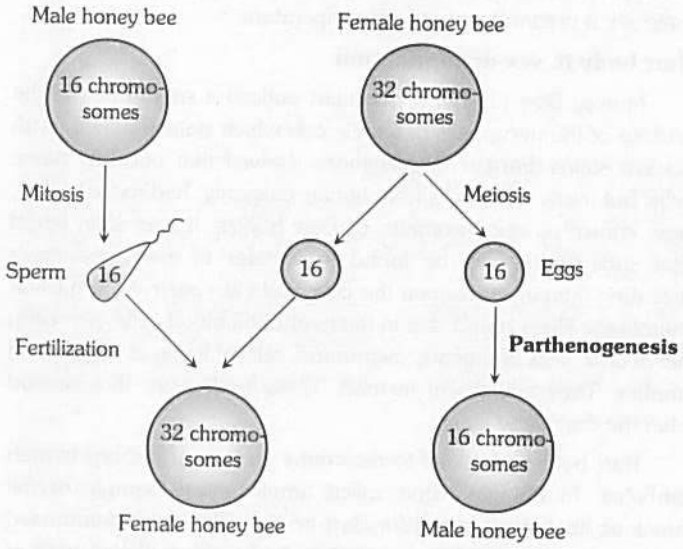


Fig : 7.1-23 Haploid-diploid mechanism of sex determination in honeybee

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

Organisms	Heterogametic sex	Gamete		Zygotes	
		Sperms	Eggs	F	M
Drosophila, man etc.	Male	X and Y	All X	XX	XY
Protenor(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 – Dilpoid 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.

Gyandromorphs in Dorsophila and ratio theory of sex determination : In *Drosophila* occasionally flies are obtained in which a part of the body exhibits female characters and the other part exhibits male characters. Such flies are known as gynandromorphs. These are formed due to misdivision of chromosomes and start as female with 2A+2X-chromosomes. 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.

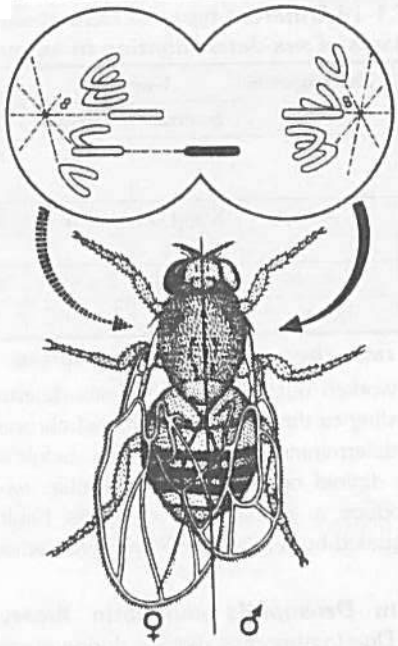


Fig : 7.1-24 Gynandromorph of Drosophila in which right half is male and left half is female

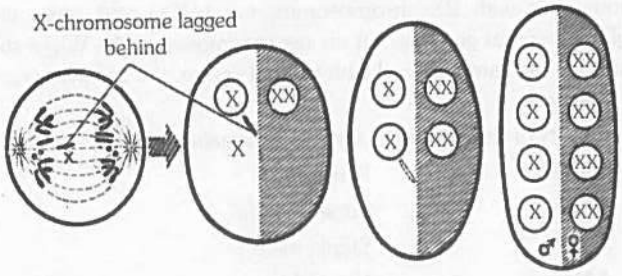


Fig : 7.1-25 Diagram to show origin of gynandromorphs

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			
Tetraploid	XXXX (4)	AAAA (4)	4/4 = 1.0
Triploid	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 Cris-cross inheritance. As the genes for most sex linked characters are located in X chromosome, they are called X-linked characters *e.g.*, colour blindness and haemophilia in man and eye colour in *Drosophila*.

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 Haemophila 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 deutoranopia. X-chromosome possesses a normal gene which control the formation of colour sensitive cells in the retina. Its recessive allele fails to do its job properly and results in colour blindness. These alleles are present in X chromosome.

Table : 7.1-17 Inheritance of colourblindness

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

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 antihaemophilic 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 diaster 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.

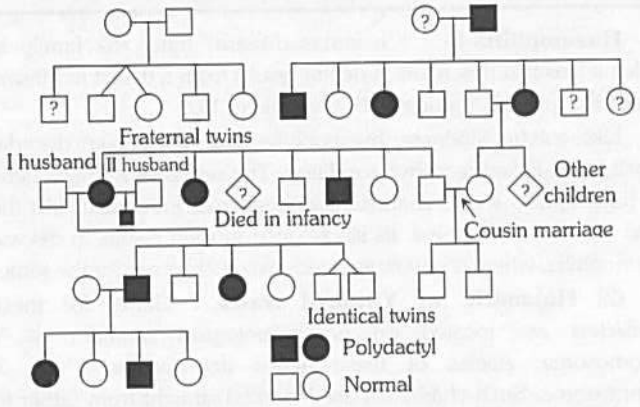


Fig : 7.1-26 Commonly used symbols in pedigree chart

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.

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

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

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

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

Eugenics is the branch of science which deals with improvement of human race genetically. 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 β 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 × tt → 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

1. Mendelism is related with [BVP 2003]
   (a) Heredity in living beings
   (b) Meiosis during sexual reproduction
   (c) Mutation in living organisms
   (d) None of the above

2. The branch of botany dealing with heredity and variation is called [MP PMT 1998]
   (a) Geobotany (b) Sericulture
   (c) Genetics (d) Evolution

3. Term '*genetics*' was given by [CPMT 1994, 97; MP PMT 2007]
   (a) Mendel (b) Morgen
   (c) Bateson (d) Boveri

4. The first great "geneticist" was [CBSE PMT 1991]
   Or
   Who is considered as father of genetics [NCERT]
   (a) Engler (b) Mendel
   (c) Schwann (d) Miller

5. Mendel was born in [MP PMT 1999]
   (a) 17^{th} century (b) 18^{th} century
   (c) 19^{th} century (d) 8^{th} century

6. Mendel was the native of [CPMT 1993]
   (a) France (b) Sweden
   (c) India (d) Austria

7. Organism with two different allele is [Odisha JEE 2008]
   (a) Heterozygous and homozygous
   (b) Heterozygous for the allele
   (c) Homozyous for the allele
   (d) None of these

8. In the first step of Monohybrid cross experiment, Mendel selected pea plants which were [MHCET 2015]
   (a) Pure tall as male and pure dwarf as female
   (b) Pure tall as female and pure dwarf as male
   (c) Heterozygous tall as male and pure dwarf as female
   (d) Heterozygous tall as female and pure dwarf as male

9. Which one of the following cannot be explained on the basis of Mendel's Law of Dominance [CBSE PMT (Pre.) 2010]
   (a) Factor occur in pairs
   (b) The discrete unit controlling a particular character is called a factor
   (c) Out of one pair of factor one is dominant, and the other recessive
   (d) Alleles do not show any blending and both the characters recover as such in F_2 generation

10. A man having the genotype EEFfGgHH can produce P number of genetically different sperms, and a woman of genotype IiLLMmNn can generate Q number of genetically different eggs. Determine the value of P and Q [WB JEE 2012]
    (a) P=4, Q=4 (b) P=4, Q=8
    (c) P=8, Q=4 (d) P=8, Q=8

11. How many types of gametes will be produced by an individual having genotype AaBbcc [MHCET 2015]
    (a) Four (b) Three
    (c) Two (d) One

12. In 1900 A.D. three biologists independently discovered Mendel's principles. They are [RPMT 1997; MP PMT 2002]
    (a) De Vries, Correns and Tschermak
    (b) Sutton, Morgan and Bridges
    (c) Avery, McLeod and McCarthy
    (d) Bateson, Punnet and Bridges

13. When a dihybird cross is fit into a punnett square with 16 boxes, the maximum number of different phenotypes available are [Kerala PMT 2008]
    (a) 8 (b) 4
    (c) 2 (d) 16
    (e) 12

14. In a monohybrid cross between two heterozygous individuals, the number of pure homozygous individuals obtained in F_1 generation is [Odisha JEE 2012]
    (a) 2 (b) 4
    (c) 6 (d) 8

15. In Mendel's experiment how many different kinds of seeds are produced from a short plant with wrinkled seeds (ttrr) [Odisha JEE 2009]
    (a) 9 (b) 4
    (c) 2 (d) 1

16. 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]
    (a) $\frac{1}{4}$ (b) $\frac{1}{8}$
    (c) $\frac{1}{16}$ (d) $\frac{3}{16}$

17. 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]
    (a) TT and tt (b) Tt and Tt
    (c) Tt and tt (d) TT and TT

18. 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]
    (a) Rrr_0 (b) $RrRr_0$
    (c) Rrr_0r_0 (d) rrR_0r_0

19. 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]
    (a) 3 : 1 (b) 1 : 1
    (c) 1 : 2 : 1 (d) 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 × WW (b) Ww × Ww

(c) Ww × ww (d) WW × WW

(e) ww × 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 geneotype 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 × 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 choosen 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 hybridieation, Tt X tt (F_1 hybrid and a recessive parent) give rise to the progency 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; WBJEE 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* × *rr pp* (b) *RR PP* × *rr pp*
(c) *RR pp* × *rr pp* (d) *RR Pp* × *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 TTrr 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* × *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 crinckled 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* × *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) *rrYy*, *rrYY* (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, phyenotypically a
(c) Genotypically Aa, phyenotypically A
(d) Genotypically aa, phyenotypically 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 of 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 if 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) Pleotropy (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 tumafaciens*
(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 Punnet

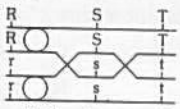
15. Which one of the following is the most suitable medium for culture of *Drosophila melanogaster* **[CBSE PMT 2006]**
(a) Ripe banana (b) Cow dung
(c) Moist bread (d) Agar agar

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%

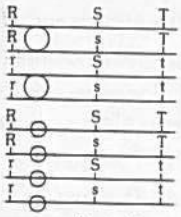
17. Linkage decreases the frequency of **[CPMT 1998]**
(a) Hybrid (b) Dominant allele
(c) Recessive allele (d) Both (a) and (b)

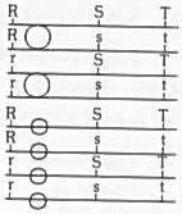
18. Crossing over in diploid organism is responsible for **[NCERT; CBSE PMT 1991, 98; MP PMT 2010]**
(a) Dominance of genes
(b) Linkage between genes
(c) Segregation of alleles (genes)
(d) Recombination of linked allele (genes)

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]**

(a) 

(b) 

(c)

(d)

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) Muton
(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 charaters in pea plants were studied by mendal 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 chaismata 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. Polytene 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 → Cistron → Muton → Recon
(b) Gene → Muton → Cistron → Recon
(c) Gene → Recon → Cistron → Muton
(d) Gene → Cistron → Recon → 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) Centrochrome (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.	*Ophioglossum*	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) Kollikar 1888 (d) Waldeyer 1888

16. The theory of recombination of linked gene due to crossing over of chromosome during zygotene of meiosis was put forwarded 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 electorn 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 construction–I (b) Secondary construction–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. Tizo 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) Lysosmes
(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 elgans* 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.	*Escherichia coli*
B.	4000 genes	2.	*Drosophila melanogaster*
C.	13,000 genes	3.	*Mycoplasma*
D.	32,000 to 50,000 genes	4.	*Homo sapiens*
E.	35,000 to 45,000 genes	5.	*Oryza sativa*

[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 posses 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) Tschermark (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) Heterochroatin
(c) Heptan (d) Cistron

65. *Drosophila melanogastor* 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 attatched 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 careers of hereditary material are **[MP PMT 2002]**

(a) Chromosomes (b) Gene
(c) Gametes (d) Gametocytes