27. Principle of Inheritance and Variation

1. Introduction

Genetics is the study of genes, genetic variation, and heredity in living organisms.

The father of genetics is Gregor Mendel, a late 19th-century scientist and Augustinian friar. Mendel studied "trait inheritance", patterns in the way traits are handed down from parents to offspring. Trait inheritance and molecular inheritance mechanisms of genes are still primary principles of genetics in the 21st century, but modern genetics has expanded beyond inheritance to study the function and behaviour of genes. Term genetics was given by Bateson for "the education of phenomena of heredity & variation.

2. Inheritance – Heredity and Variations

2.1 Heredity

It is the transmission of genetic characters from parents to the offsprings. It deals with the phenomenon of "like begets like", e.g., human babies are like human beings in overall characteristics. About 200 characters are found to be hereditary in man.

2.2 Variations

The progenies of the same parent are not identical to each other. Variation is the degree by which progeny differs from their parents. These are common in sexually reproducing organisms. The difference in characters of offspring mainly depend upon crossing over (occurs during meiosis) and fertilization. Asexually reproducing organisms are monoparental, hence shows no genetic variations.Heredity & variation can be studied in offspring or siblings (offspring at different births) of biparental sexual reproduction.

2.3 Branches of Genetics

(1) **Transmission genetics /Classical genetics :–** Study of Mendelian and non- Mendelian genetics.

(2) **Forward genetics :–** It is the identification of mutated gene using the mutated phenotype.

(3) **Reverse genetics :–** It is the study of genes whose protein products are unknown.

(4) **Cytogenetics :–** It is the study of various aspects of chromosomes.

(5) **Molecular/biochemical genetics :–** It is the study of structure and functions of genes.

(6) **Population or biometrical genetics :–** It is the study of the behaviour and effects of gene in population using mathematical tools.

3. Mendelian History

3.1 Gregor Johann Mendel

is father of genetics (proposed theory of inheritance). He was born on July 22, in 1822 in Silisian, a village in Heinzendorf. In 1843, he joined Augustinian monastery at Brunn (then in Austria, now Brno Czechoslovakia). He was made a priest (monk) in 1847 and a teacher in 1849. Gregor Johann Mendel was sent to study Botany and Physics at the University of Vienna from 1851-1853. Breeding experiments were performed between 1856–1863 (7 years) and collected the results. 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 Hybridisation". Mendel died due to kidney disorder in 1884 without getting any recognition during his life time.

3.2 Reasons for Non recognition of Mendel's Work

(1) Scientific world was busy at that time with Darwin's work (origin of species, Darwin.

(2) Mendel's ideas were ahead of his time.

(3) Local nature of the journal in which Mendel's work was published.

(4) Mendel failed to reproduce his conclusions on hawkweed.

4. Genetics Terminology

4.1 Character

Well defined morphological or physiological feature of organism, e.g., stem height.

4.2 Trait

It is the distinguishing feature of a character, e.g., tallness, dwarfness.

4.3 Mendelian Factor

Unit, which takes part in inheritance & expression of hereditary character.. The term factor was changed into gene by Johnnsen.

4.4 Alleles (Bateson)

They are various forms of a gene or Mendelian factor, which occur on the same locus on homologous chromosomes and control the same character. Alleles or allelomorphic pair controls different expressions or traits of same character (tallness and dwarfness in pea) but these days term is also used for similar form of Mendelian factor or gene present on two homologous chromosomes. Factors can be of two types:

(1) **Dominant factor :–** Allele, which is expresses in heterozygous as well as in homozygous state. It is denoted by capital letter (T or D for tallness in pea, R or W for round seeds in pea).

(2) **Recessive factor :–** Allele, which is unable to express in heterozygous or in presence of dominant allele. It expresses only in the homozygous state (e.g., tt, rr). It is denoted by small letter (t or d for dwarf ness in pea r or w for wrinkled seed in pea).

4.5 Homozygote (Bateson and Saunders)

An individual having identical mendelian factors or genes for a character (TT or tt), Homozygote or homozygous individual is always pure for the trait.

4.6 Heterozygote (Bateson and Saunders)

An individual having two contrasting factors or alleles for a character. Heterozygote or heterozygous individual is also called hybrid, e.g., Tt or Dd.

4.7 Hemizygous

If individual has only one gene of a pair then individual said to be hemizygous. Human male is always Hemizygous for sex linked gene.

4.8 Cross

Fertilization between male and female plants. It may be

(1) **Monohybrid cross :–** A cross made to study inheritance of a single pair of factors or genes.

(2) **Dihybrid cross :–** A cross made to study simultaneous inheritance of two pairs of factors or genes.

(3) **Selfing or cross :–** Fertilization with male gametes of the same individual.

(4) **Reciprocal crosses :–** A pair of crosses involving male of one type and female of second type and then vice versa (male of second type and female of first type). Reciprocal crosses are useful in knowing sex-linked characters and characteristic dominance.

(5) **Back Cross :–** Cross between hybrid and one of its parental genotype. It is of two types -

- **Out cross :–** Cross between F_1 and dominant organism (or parent).
- **Test cross :–** Cross between F_1 and a recessive organism to know homozygosity or heterozygosity of F_1.

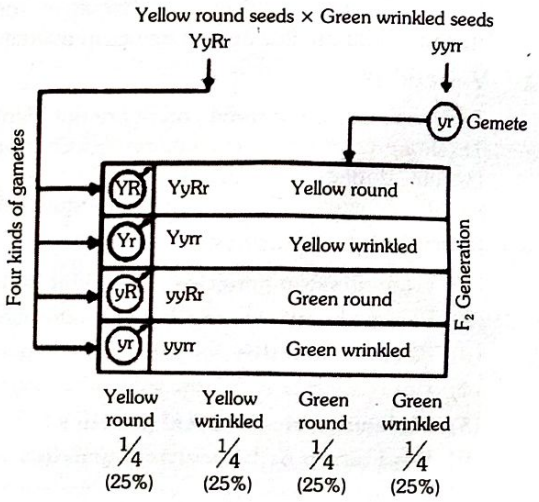


Figure :– Dihybrid test cross

Test cross yields a ratio is 1 : 1 for monohybrid test cross and 1 : 1 : 1 : 1. For dihybrid test cross.

4.9 Hybrid/Heterozygous

Individual obtained by crossing two genetically different organisms. Heterosis (Hybrid Vigour; Shull): Presence of superior qualities in hybrid than either of parents.

4.10 F_1 Generation

F_1 or first filial (sons & daughters) generation is derived from a cross between genetically different parents (generally homozygous) so that F_1 generation consists of hybrids). Checker board/Punnet Square (Punnet): Square used to shows probable gametes, phenotype and genotype of cross.

4.11 F_2 Generation

F_2 or second filial (sons and daughters) generation is the generation derived from self breeding or breeding between individuals of F_1 generation.

4.12 Pure line (Johannsen)

True breeding individuals, which have homozygous traits due to continued self breeding over 3-4 generations or doubling of chromosomes in haploid individual.

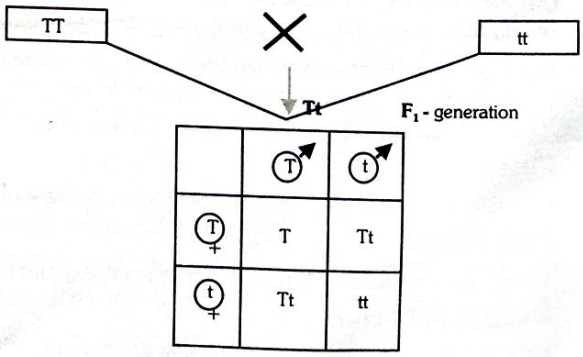
4.13 Genotype (Johannsen)

Genetic constitution of an individual with regard to one or more characters. e.g. TT (pure tall), Tt (hybrid tall), tt (dwarf).

F_2 genotypes ratio = $(1: 2 : 1)^n$. Where n = No of heterozygosity in F_1.

First time it was used by C punnett. The representation of generations to analyze in the form of symbols of squares male gametes lie horizontally and female gemetes lie vertically.

The ratio of characters (traits) appear/visible morphologically is phenotypic ration it is 3:1 Genetic constitution is called Genotype (using symbols for genes) it is 1:2:1.



TT = tall (dominant homozygous)

Tt = Tall (dominant heterozygous),

tt = Dwarf (recessive homozygous)

4.14 Phenotype (Johannsen)

The external, morphological or physiological expression of an individual. For recessive genes, phenotype and genotype are similar. For dominant genes, the phenotype is same for both homozygous and heterozygous states.

F_2 Phenotypes ratio = $(3 : 1)^n$.

Where n = No of heterozygosity in F_1.

Phenotype is influenced by environment as well as age. In primrose, dominant allele for red flower colour (A) does not express above 30°C so that flowers appear white (colour of recessive homozygote aa).

4.15 Phenocopy

If different genotypes are placed in different environmental condition then they produce same phenotype. These genotypes are said to be phenocopy of each other.

4.16 Number of Gamete types

Depends upon genotype of parents In bisexual plants both male and female gametes are formed with similar constitution. Segregation occurs before gamete formation. AA will form one type of gametes, A. AABB and AABBCC will also form one type of gametes-AB or ABC.

(No. of gametes = 2^n, n= No. of heterozygosity)

Aa has two types of gametes, A and a. AaBb parent produces aB, ab, Ab and aB gametes.

Aa Bb Cc parent develops eight types of gametes – ABC, AbC, Abc, aBC, aBC, aBc, abC, abc.

5. Mendelian Experiment

5.1 Rediscovery of Mendelism

In 1900, Hugo de Vries of Holland, Carl Correns of Germany and Tschermak 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 generalization of Mendel into principles or law of heredity.

5.2 Selection of Pisum sativum by Mendel:

(1) John Goss had already worked on Garden/Edible Pea/ Pisum sativum

(2) Pure varieties of Pea were easily available in garden.

(3) Life cycle of Pea plant is of a few months so that results can be seen quite early.

(4) Flowers are bisexual and flowering normally self pollinated.

(5) Flowers can be cross-pollinated only manually.

(6) Pea plant has 7 contrasting characters that explain genetic inheritance.

5.3 Reasons for success of Mendel

(1) Mendel 's breeding material had pure lines/ pure breed.

(2) He chose only those characters, which showed consistent results.

(3) He worked with 1-3 traits at one time.

(4) He kept pedigree record of every cross and subsequent generations

(5) He used statistical analysis

(6) He was lucky in choosing those characters, which showed complete independent assortment even then seven characters chosen by him being present on four chromosomes-1, 4, 5, & 7.

(7) He left out some cases for which he could not provide explanation (flower & seed coat colour).

5.4 Mendel's experimentation

Mendel's experiments involved four steps- selection, hybridization, selfing and calculations. Mendel experimented on pure breeding plant of Pea (Pisum sativum) He selected 14 contrasting traits for 7 characters in pea for his experiments.It is now known that theses 7 characters are located on only 4 chromosomes of pea plant. Mendel used hybridization technique to conduct his experiments. Hybridisation is the crossing of two different individuals to produce an offspring which will have characters of both parents. He crossed parents with contrasting traits. Firstly he made monohybrid cross (i.e. cross between parent that differ from each other in one character) followed by dihybrid cross and finally trihybrid cross. The F1 hybrids were self crossed to give rise to F2- generation.

6. Monohybrid Cross (Inheritance of One Gene)

It is a cross involving single pair of contrasting traits of a character. All the contrasting traits used by Mendel and his results are given below. Mendel studied 7 characters or 7 pairs of contrasting traits

In a monohybrid cross, when a cross is made between pure tall and pure dwarf plant in F1 generation , all plants will be tall. When F1 plants are self-pollinated, than in F-2 generation both tall and dwarf plants are found in apporx. Ratio of 3:1.

F_2 ratio obtained in monohybrid cross by selfing of F_1individuals.

Phenotypic ratio is 3 : 1(dominant : recessive) but genotypic ratio is 1 : 2 : 1(pure dominant : hybrid dominant : recessive).

Actual data obtained by Mendel in F_2 progenies in garden pea				
S.No	Character	Dominant	Recessive	Ratio
1	Length of plant	787(tall) (T) or D	277(dwarf) (t) or(d)	2.84:1
2	Flower position	651(axial) (A) or T	207(terminal) (a) or (t)	3.14:1
3	Shape of pod	882(inflated) (I) or C	299(constricted) (i) or (c)	2.91:1
4	Colour of pod	428(green) (G) or Y	152(Yellow) (g) or (y)	2.82:1
5	Shape of seed	5,474(round) (R) or W	1.850(wrinkled) (r) or (w)	2.96:1
6	Cotyledon colour	6,022(yellow) (Y) or G	2,001(green) (y) or (g)	3.01:1
7	Colour of flower	705(violet) (V or R) or W	224(white) (v or r) or w	3.15:1
Average of all traits studied 2.98:(= 3:1)				

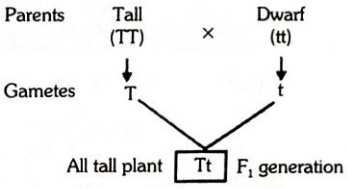
6.1 Mendel's Postulates based on monohybrid cross

(1) Postulate on factors of genetics :– A character is represented genetically in an individual by two unit factors present on same locus in the two homologous chromosomes.Two unit factors can give rise to three types of combinations – two of first trait (TT), two of second trait (tt) or two of both the traits (Tt) as in hybrid

(2) Postulates on Dominance and Recessiveness

(3) Postulate on Segregation

6.2 Mendel's laws of Inheritance based on monohybrid cross.



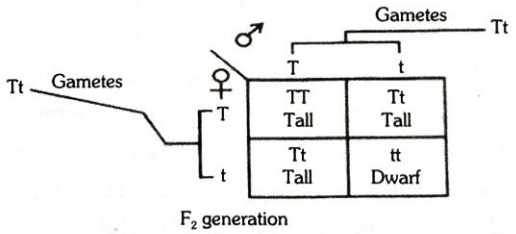
(1) Law of Dominance :– In a hybrid where both contrasting alleles are present. Only one unit factor/allele is able to express called dominant other is recessive remains suppressed. This law is not universally applicable. In a cross between pure breeding red flowered (RR) Pea plant and white flowered (rr) pea plant, F_1 generation is red flowered though it has received both the factors (R and r) because red is dominant over white colour. On selfing, recessive trait reappears in F_2 shows that it is suppressed in F_1 generation and not lost.

Phenotyoic ratio – 3 : 1

Genotypic ratio – 1:2:1

$\text{Tall} - \frac{3}{4}$, $\text{Dwarf} - \frac{1}{4}$

$\text{TT} - \frac{1}{4}$, $\text{Tt} - \frac{2}{4}$, $\text{tt} - \frac{1}{4}$



F_2 generation

- **Exception to law of Dominance :–**

(a) Incomplete Dominance/Blending Inheritance-(By Correns in Mirabilis jalapa) :– Phenomenon of neither of the two alleles being dominant so that expression in hybrid is an intermediate between two alleles. Incomplete dominance is not blending inheritance because parental characters reappear in F_2 generation. In snapdragon (Dog Flower, Antirrhinum) and 4 O'clock (Mirabilis jalapa) there are two types of pure breeding plants, red and white flowered. They produce hybrid F_1 plants with pink flower. On selfing them, F_2 generation has 1 red : 2 pink : 1 white flowered plants with phenotypic ratio being similar to genotypic ratio. Pink colour is due to incomplete dominance of red colour over white colour.

(b) Codominance :– Phenomenon of two alleles of a same gene, both expressing themselves in the organisms. Human RBC has two antigens, M and N. Alleles are codominant. Human can be MM, MN or NN. The codominant alleles are shown with same capital letter but with different superscripts like I^AI^B for alleles in human blood group AB or Hb^AHb^S for normal and sickle cell erthrocytes. In Andalusian fowls, crossing of black and white bird yields blue F_1 fowls but F_2 generation has 1 black : 2 blue : 1 white. Blue colour appears due to fine mixing of black and white feather parts. In short horned cattle, cross of red and white animals yield roan in F_1 and 1 red : 2 roan : 1 white in F_2.

Explanation of the concept of dominance :– Every gene contains information to express a particular trait. Diploid organisms have two copies of each gene, they are called alleles. These two alleles may be identical or non-identical. One of them may be different due to some changes that it has undergone which modifies the information that particular allele contains. Theoretically, the modified allele could be responsible for production of

(i) The normal/less efficient enzyme, or

(ii) A non-functional enzyme, or

(iii) No enzyme at all

In case (i), the modified allele is equivalent to the unmodified allele, i.e., it will produce the same phenotype/trait. But, if the allele produces a non-functional enzyme or no enzyme. In case (ii) and (iii), the phenotype may be effected. The unmodified (functioning) allele, which represents the original phenotype is the dominant allele and the modified allele is generally the recessive allele. Hence, the recessive trait is due to non-functional enzyme or because no enzyme is produced. If the mutated allele forms an altered but functional product, it behaves as incompletely or codominant allele.

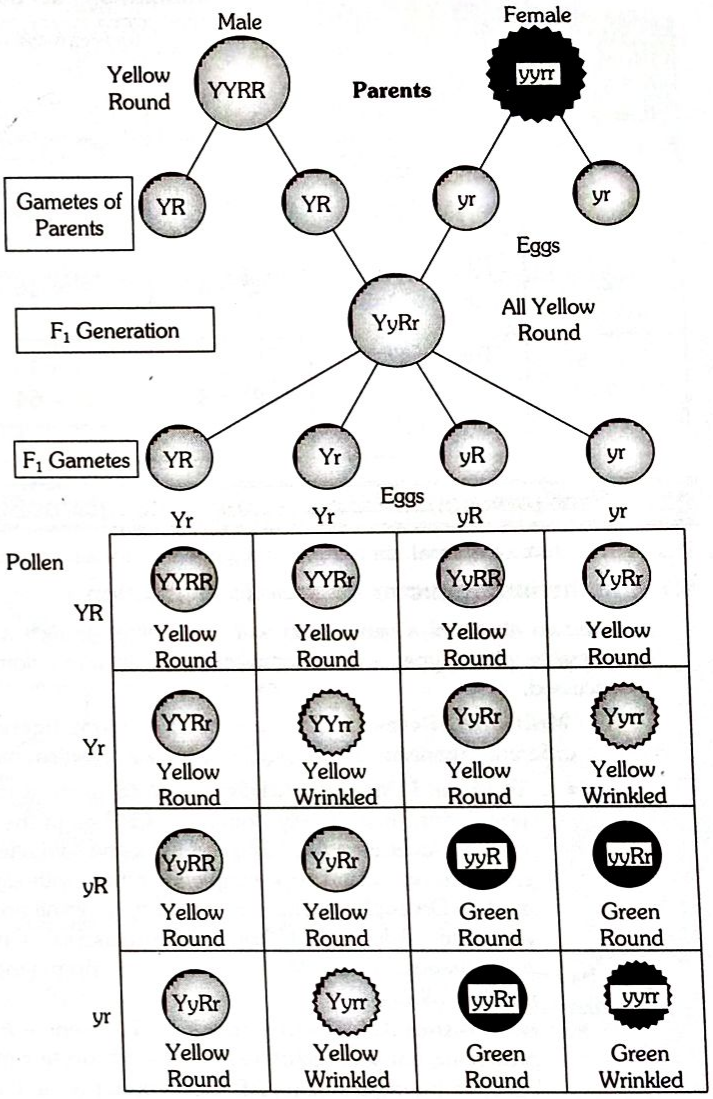
(2) Law of Segregation (Principal of purity of gametes) :– Alleles of a trait or particulate entities which keep their identity in the hybrid, separate out at the time of gametogenesis randomly passing to different gametes for random pairing during passage to offspring. This law is universally applicable. In a monohybrid cross, pure tall plant (TT) and dwarf plant (tt) forms F_1 or hybrids (Tt) are all tall. On self breeding them, both tall and dwarf appear in ratio of 3 : 1 (787 tall to 277 dwarf). Further self breeding shows that phenotypic monohybrid ratio of 3 tall : 1 dwarf is genotypically 1 tall : 2 hybrid tall : 1 dwarf. Since gamete carries only one factor of a trait, this is also c/d principle of purity of gametes.

7. Dihybrid Cross (Inheritance of two genes)

These crosses are made to study the inheritance of two pairs of Mendelian factors or genes. For example- When a cross is made between yellow-round and wrinkled-green seeds plants with only yellow round seeds are seen in F1 generation but in F2-generation, four types of combinations are observed. Two of these combinations are similar to the parental combinations and others are new combinations. These are round green and wrinkled yellow. The ratio of four combination in F2 generation is Phenotype ratio 9 : 3 : 3 : 1(both dominant : one dominant second recessive : one recessive second dominant : both recessive).

Genotypic dihybrid ratio is 1 : 2 : 1 : 2 : 4 : 2 : 1 : 2 : 1.

It has 1 (both pure dominant) : 2 (one pure dominant, second hybrid dominant) : 2 (one hybrid, second pure dominant) : 4 (both hybrid dominant) : 2 (one hybrid dominant, second recessive) : 2 (one recessive, second hybrid dominant): 1 (one pure dominant, second recessive) : 1 (one recessive, second pure dominant) : 1 (both recessive).



Mendel's dihybrid cross between pea plants having yellow round seeds and green wrinkled seeds

7.1 Mendel's laws of Inheritance based on dihybrid cross

(1) Law of Independent assortment :– Two factors of each trait assort at random and independent of factors of other traits at the time of meiosis and get randomly as well as independently rearranged in offspring. Mendel crossed pure yellow round seeded (YYRR) Pea plant with green wrinkled seeded (yyrr) Pea plant. F_1 plants were all yellow and round seeded (RrYy). In a dihybrid cross if genes are to assort independently, F_1 organisms will produce four types of male (RY, Ry and rY and ry) and four types of female gametes with 16 types of gametic recombination (4×4). In F_2 generation four types of plants appeared – 315/556 or 9/16 yellow rounded, 101/556 or 3/16 yellow and wrinkled, 108/556 or 3/16 green rounded and 32/556 and 1/16 green wrinkled.

Important-

Differential ratio in F2 generation can also be arrived by law of probability, which states that when two independent events occur simultaneously, the combined probability of the two outcomes is equal to product of their individual probabilities of occurrence.

In case of seed colour, F2 generation has 3/4 yellow seeds and 1/4 green seeds. In case of seed shape, F2 generation has 3/4 round seeds and 1/4 wrinkled seeds. Their combined probabilities are Yellow round 3/4×3/4=9/16 Yellow wrinkled 3/4×1/4=3/16 Green round 1/4×3/4= 3/16 Green wrinkled 1/4 × 1/4=1/16. Law of independent assortment is applicable to only those genes, which are present on different chromosomes or showing regular crossing over.

8. Trihybrid Cross

Ratio obtained in F_2 generation raised in trihybrid cross is followed by selfing F_1. Phenotypic ratio is 27 (all three pure or hybrid dominant): 9 (one and two dominant, third recessive) : 9 (one and third dominant, second recessive) : 9 (one recessive two and three dominant) : 3 (one dominant, two and three recessive) : 3 (second dominant, one and three recessive) : 3 (one and two recessive, third dominant) : 1 (all three recessive).

Genotypic trihybrid ratio is 1 : 2 : 1: 2 : 4 : 2 : 1 : 2 : 1: 2 : 4 : 2 : 4 : 8 : 4 : 2 : 4 : 2 : 1 : 2 : 1 : 2 : 4 : 2 : 1 : 2 : 1

Summarised account of Mendel's experiments

Number of traits/ hybrid (n)	Experiment	Types of gametes (2^n)	Number of zygotes/ offspring $(gametes)^2$	Number of Phenotype (2^n)	Number of genotype (3^n)	Phenotypic ratio	Genotypic ratio
1.	Monohy-brid ross (Aa × Aa)	$2^n = 2^1 = 2$	$2^2 = 4$	$2^n = 2^1 = 2$	$3^1 = 3$	3 : 1	1 : 2 : 1
2.	Dihybrid cross (AaBA × AaBb)	$2^2 = 4$	$4^2 = 16$	$2^2 = 4$	$3^2 = 9$	$(3 : 1)^2$ = 9 : 3 : 3 : 1	$(1 : 2 : 1)^2$ = 2 : 4 : 2 : 1 : 2 : 1 : 1 : 2 : 1
3.	Trihybrid cross [AaBbCc × AaBbCc]	$2^3 = 8$	$8^2 = 64$	$2^3 = 8$	$3^3 = 27$	$(3 : 1)^3$	$(1 : 2 : 1)^3$

9. Post-mendelian discoveries/ gene interaction

It is modification of normal phenotype of a gene and nonallelic genes. Due to interaction of their alleles which is of two types-

9.1 In the intragenic or interallelic interaction

The two alleles of a gene, react with each other in such a way as to produce an expression different from the normal dominant – recessive phenotype, e.g., incomplete dominance, co dominance, multiple alleles.In this Incomplete and co-dominance are already discussed.

(1) Multiple Alleles :– They are multiple forms (more than two) of a gene, which occur on same gene locus, distributed in different organisms of a population, but an organism has only two alleles and a gamete has only one allele.

- **Wild and Mutant Alleles :–** Wild allele was originally presented in population and till now most widespread in population. It is usually dominant. Change in the wild allele gives rise to recessive and less common mutant allele. There may be several mutant alleles of the same wild allele. Symbol used for such an allele is represented by small letter for mostly recessive and using superscripts for others with sign of (+) for the wild type, e.g., w (white eyes), w^i, w^{bl}, w^c, w^h, w^+ (red eye) in Drosophila. There is a wild type dominant allele designated with a superscript of (+) or capital letter. Completely recessive alleles are without any superscript. Other alleles are mutants of either of the two. They show dominant – recessiveness, codominance, incomplete dominance. No. of genotypes in multiple allelism= n/2(n+1), Where n =No. of multiple alleles.
- **Inheritance Of Blood Groups In Human :–** ABO blood grouping of humans is determined by three alleles I^A,I^B and I^o producing antigen/agglutinogen on erythrocyte surface named A, B and nil respectively. I^A and I^B and show codominance between them producing AB blood group while they are both dominant to alleles I (L^O). Blood group O is produced when two recessive ii alleles are present. Coat pigment in Rabbit is expressed by four alleles, C, c^{ch}, c^h and c wild grey or agouti colour is due to dominant allele C, silver grey or chinchilla due to C^{ch}, C^{ch}, and albmo due to CC. In Himalayan from the fur is white with dark brown /black extremities (feet, tail, nose, pinnate). Leaf form in cotton, seed coat colour in Maize, self –sterility in plants, and red-white eye colour series in fruitfly Drosophila are all controlled by multiple alleles.

Phenotype (Blood group)	Genotype
A	I^AI^A or I^AI^O
B	I^BI^B or I^BI^O
C	I^AI^B
O	I^OI^O or ii

(2) Pleotropy :– They are genes, which influence more than one trait. It has one more evident effect on one trait called major effect and less evident effects on other traits called secondary effect. When a number of related changes are caused by a pleiotropic gene, the phenomenon is called syndrome, e.g. sickle cell anaemia. The later is an autosomal hereditary disorder due to Hb^s Instead of Hb^A (normal).Both are codominant. Hb^A produces normal haemoglobin while Hb^S forms hame-globin in which β-chain has amino acid valine instead of glutamic acid in 6th position. This

S.No.	Blood groups of parents	Possibilities of Children's blood group
1.	A × A	A and O
2.	A × B	A, b, AB and O
3.	A × AB	A, B and AB
4.	B × B	B and O
5.	A × O	A and B and AB
6.	B × O	B and O
7.	B × AB	A, B and AB
8.	AB × O	A and B
9.	AB × AB	A, B and AB
10.	O × O	O

is major effect. Under oxygen deficiency (at high altitude, vigorous exercise) 6-valine develops hydrophobic bonds with complementary sites on adjacent strands forming a helical polymer of upto 14 strands .It changes erythrocyte circular to sickle cell form (secondary effects).

In Garden pea a single gene control flower colour, colour of seed coat and red spots in the axils of leaves.

(3) Lethal Genes (2:1 instead of 1:2:1 or 3:1) :- Genes, which control some vital functions of organism and cause death of the organism in homozygous condition. Most striking example to explain lethal gene is sickle cell anaemia (Hb^SHb^S).

Cuenot first reported that inheritance in mouse body colour did not agree with Mendelian inheritance, because the dominant allele for yellow body colour is lethal in homozygous condition.

In plants, it was first reported in Antirrhinum majus by E. Baur, where yellow leaved or golden leaved or Aurea plant never bread true. Thus, the ratio comes out to be 2 : 1.

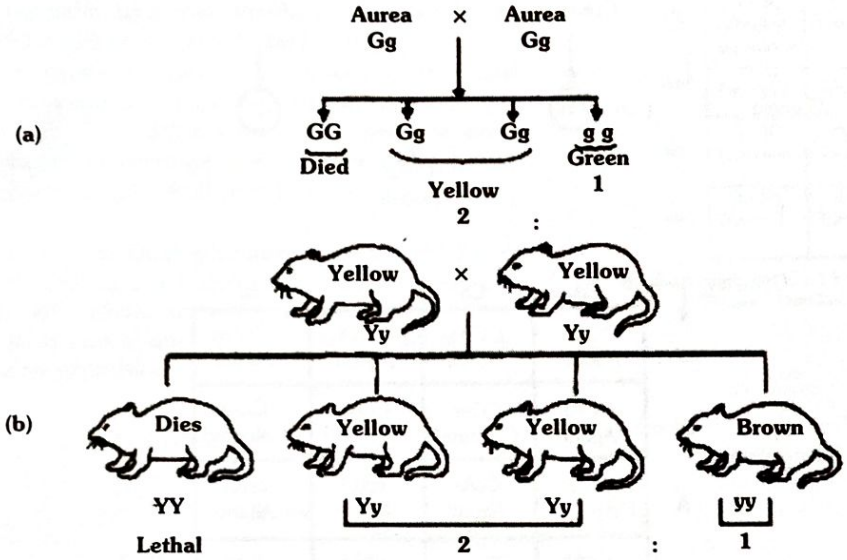


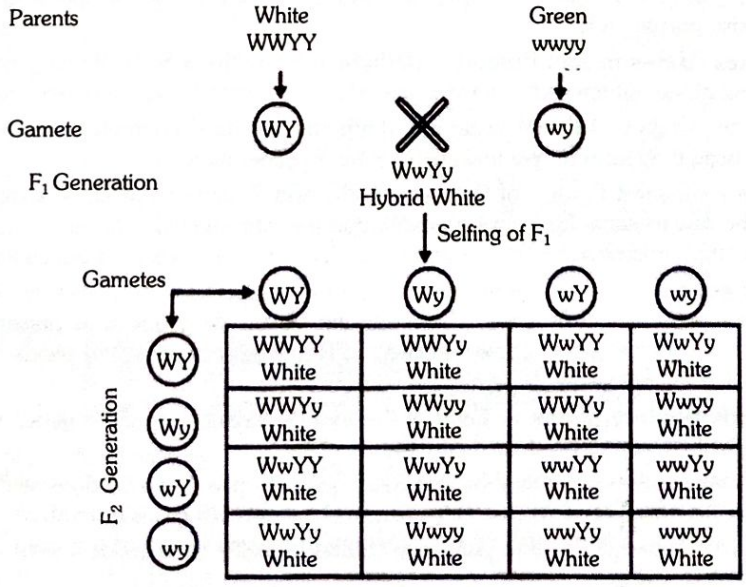
Figure :- Inherltance of lethal gene in (a) Antirrhinum and (b) in mice

The individual dies in embryonic state in absolute lethality (yellow fur in mice), before reproductive maturity in sublethality (e.g., sickle cell anaemia, brachypalangy) and after sexual maturity in delayed lethality, with the death of homozygous lethal the monohybrid ratio comes to 2:1.

9.2 In intergenic or nonallelic interaction

Two or more independent genes of same or different chromosomes interact to form a different expression, e.g. complementary genes, supplementary genes, duplicate genes, epistasis, lethal genes.

(1) Epistasis (Bateson) :- Phenomenon of suppression of phenotypic expression of a gene by nonallelic gene which shows its own effect. Gene which masks effect of another is called epistatic gene while the one which is suppressed is termed hypostatic gene. It is of two types–dominant, recessive.



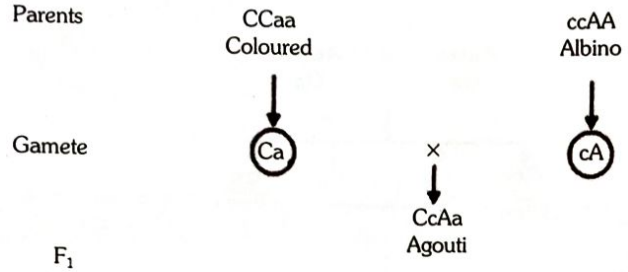
F_2 Phenotypic ratio : 12 White : 3 Yellow : 1 Green or 12 : 3 : 1

Figure : Dominant epistasis

- **Dominant Epistatis: (Dihybrid F2 ratio is 12:3:1) :–** A dominant epistatic allele suppresses expression of a nonallelic gene whether the later is dominant or recessive. Fruit colour of summer squash (Cucurbita pepo) is contributed by a gene, which produces yellow colour in dominant state (Y-) and green colour in recessive state (yy). There is non-pigment forming dominant epistatic gene, (W-).
- **Recessive Epistasis: (Dihybrid F2 ratio is 9:3:4) :–** Epistatic gene suppresses expression of nonallelic gene only when it is in homozygous recessive state.

(a) In onion, red (R) and yellow (r) bulbs are produced due to (I-R-) and (I-rr) state. When epistatic gene is homozygous recessive, bulb appear white (iiR, iirr).

(b) Coat colour in mice also shows recessive epitasis.



Gametes → / ↓	CA	Ca	cA	ca
CA	CCAA Agouti	CCAa Agouti	CcAA Agouti	CcAa Agouti
Ca	CCAa Agouti	CCaa Coloured	CcAa Agouti	Ccaa Coloured
A	CcAA Agouti	CcAa Agouti	ccAA Albino	ccAa Albino
ca	CcAa Agouti	Ccaa Coloured	ccAa Albino	ccaa Albino

F2 Generation

F_2 Phenotypic ratio : 9 (Agouti) : 3 (Coloured) : 4 (Albino)
Figure : Recessive epistasis

(2) Supplementary Genes (Dihybrid F2 ratio is 9:3:4) :– These are two nonallelic genes in which one type of genes produces its effect whether the other is present or not and second (supplementary) gene produces its effect only in the presence of the first usually forming a new trait. In cholam (Sorghum) a gene produces blackish purple glume colour in dominant state (P-) and brown colour in recessive state (pp)

A nonallelic supplementary gene Q-interacts with dominant P-gene to form reddish purple glume colour. It has no effect on the recessive nonallele (pp Q- is brown).

A cross between PPqq (Blackish Purple) and ppQQ(brown) yields reddish purple glume (PpQq) and F2 dihybrid ratio of 9 reddish purple : 3 blackish purple : 4 brown.

(3) Complementary Genes (Bateson and Punnet / (Dihybrid F2 ratio is 9:7)- If two genes present on different loci produce same effect when present alone but interact to form a new trait when present together, they are called Complementary Genes.

For example there are two white varieties of sweet pea (Lathyrus odoratus) controlled independently by two different genes (C-pp and ccP-). When crossed they form purple flowers (C-P-) in F_1 generation.

Selfing of hybrids gives a dihybrid F_2 ratio of 9 purple (C-P-) and 7 white (ccpp,cc- -, - -pp). Dominant *gene* C produces an enzyme that converts the raw material for flower pigmentation into chromagen. Dominant gene P produces another enzyme that oxidizes chromagen into purple anthocyanin. So, the dominant alleles of both genes are required for expression of flower colour.

(4) Suppressor/Inhibitor gene :– It is a non lethal gene without any expression non allelic gene. Example, in Rice I-gene inhibits the expression of dominant purple colour gene (P) so that the leaves are green in its presence (I-P-). Green leaves also occur when leaf colour gene is recessive (iipp). Cross between IIPP and iipp both green) yields hybrid greens (IiPp) which on self breeding form 3 purple to 13 green plants.

(5) In collaborative supplementary genes :– Each of the two dominant non allelic genes shows independent expression but when present together, they interact to produce a new trait.

In poultry formation of rose comb is controlled by dominant gene R-, pea comb by dominant gene P-, single comb by recessive pprr. When both dominant genes (P and R) occur together, walnut comb (P-R-) is formed.

A cross between pure pea-combed (PPrr) and pure rose combed (ppRR) birds yields walnut combed birds which on inbreeding form 9 walnut : 3pea : 3 rose: 1 single.

(6) Duplicate Genes (F2 ratio is 15:1) :– Two or more independent genes found on different chromosomes, which produces same or nearly similar phenotype in dominant state, producing same intensity of effect even when present together, so that dominant phenotype is most abundant.

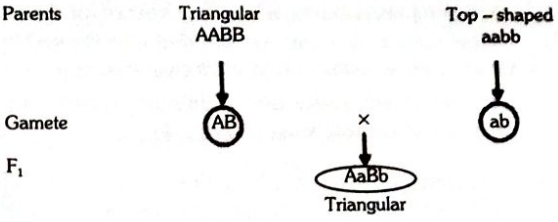
(i) Oat black grain colour is produced by B_1- - - or B_2 - - - and white by b_1 b $_1$b $_2$ b_2. when two pure breeding black grained varieties ($B_1B_1b_2b_2$ × $b_1b_1B_2B_2$) are crossed , F_1 are all black ($B_1b_1B_2b_2$). On self breeding they produces black and white grained plants in the ratios of 15:1.

(ii) In shepherd purse two independently genes influence fruit shape with similar phenotypic effect.

(7) Polymeric/Additive Genes :– They are duplicate genes with cumulative /additive effect i.e. two independent dominant genes (whether homo-zygous or heterozygous) having similar phenotypic effect individually but produces a new cumulative effect (similar in homozygous and heterozygous states), if present together.

In summer squash (cucurbita pepo), two independent genes produces circular (for spherical) shape in dominant state (S_1 . s_2s_2 and s_1s_1 . S_2 and long (or cylindrical) shape in recessive state ($s_1s_1s_2s_2$). When both the dominant genes are present (S_1-S_2 .), disc –shaped fruits are formed. F_2 dihybrid ratio is 9 discoid : 6 circular :1 long.

Qualitative Inheritance :– Qualitative inheritance is that type of inheritance in which one dominant alleles influences the complete traits so that two such alleles do not change the phenotype. Monogene is gene in which one dominant allele controls the complete or qualitative expression of a trait.

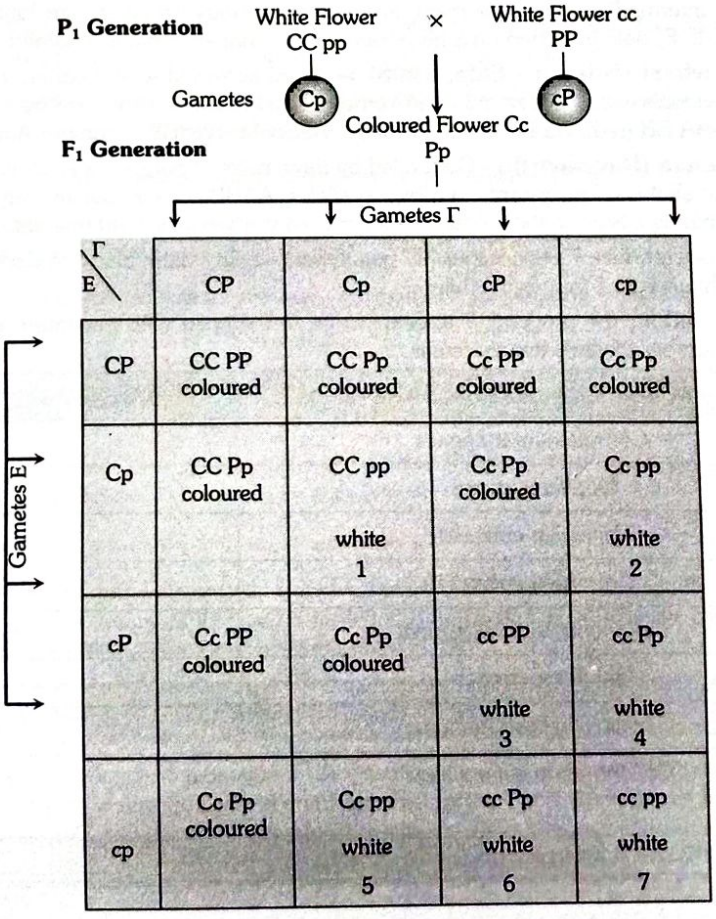


Selfing of F_1

Gametes →	AB	Ab	aB	ab
AB	AA BB Triangular	AA Bb Triangular	Aa BB Triangular	Aa Bb Triangular
Ab	Aa Bb Triangular	AA bb Triangular	Aa Bb Triangular	Aa bb Triangular
aB	Aa BB Triangular	Aa Bb Triangular	aa BB Triangular	aa Bb Triangular
ab	Aa Bb Triangular	Aa bb Triangular	aa Bb Triangular	aa bb Top shaped

(F_2 Generation)

F_2 Phenotypic ratio : 15 (Triangular) : 1 (Top shaped)
Figure : Duplicate genes interaction



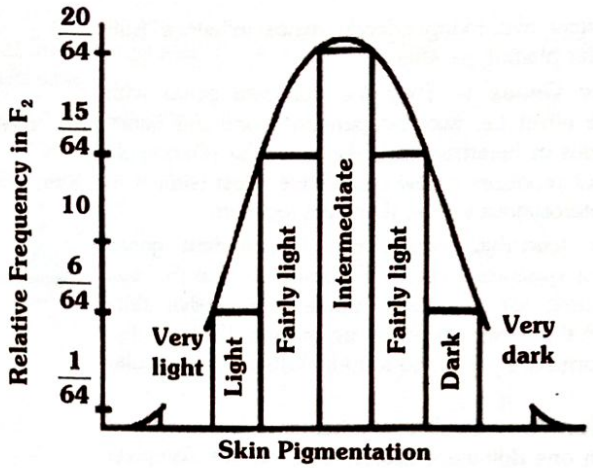
The results of an experiment to show the operation of complimentary genes in the production of flower colour in sweet pea (*Lathyrus*)

9.3 Quantitative Inheritance

The total phenotypic expression is the sum total/additive /cumulative effect of all the dominant alleles of genes. Polygenic inheritance is that type of inheritance in which the complete expression of a traits is controlled by two or more genes in which a dominant allele of

each gene contributes only a unit fraction of the trait. First scientific study of polygenic inheritance was carried out by Nilsson–Ehle on Kernel colour in wheat. Before that also studied by Korleuter on Tobacco and Galton on human. Mertic (quantitative) trait is a trait, which can be measured in units (weights, size, number, intensity of colour).

Only dominant alleles are contributing alleles as they take part in produces a number of phenotypes in F_2 generation – Number of phenotype for polygenes = $2n + 1$



(1) 1 : 2 : 1 (3 phenotypes) in case of a pair of alleles.

(2) 1: 4: 6: 4:1 (5 phenotypes) in case two pairs.

(3) 1: 6 : 15 : 20 : 15 : 6 : 1 (7 phenotypes) when there are three pairs of alleles or polygenes.

As result the number of intermediate types are many and like continuous variations are found in the progeny .The number of parental types are fewer. If F_2 data is plotted on a histogram , a bell-shaped curve is obtained.

(1) Kernel colour in wheat (Nilsson - Ehle, 1908) :– A red kernelled wheat variety crossed with white kernelled wheat variety yields F_1individuals with medium coloured kernel. Kernel colour is controlled by two polygenes with dominant alleles contributing colour–AA BB (red), Aa BB or AABb (dark), AaBb/Aabb/aaBB(medium), Aabb/aaBb(light) and aabb (white).

(2) Skin colour in Human (Davenport) :– Controlled by three pairs of polygenes A, B and C. Negro /very dark/black colour is due to presence of all the six dominant contributing alleles AABBCC. Caucasian /very light/white colour is due to all six recessive non-contributing alleles aabbcc. A marriage between the two yields intermediate or mulatto(AaBbCc) children .

Marriage b/w mulattoes produce 7 phenotypes (27 genotypes) –1 very dark /black : 6 dark : 15 fairly dark : 20 intermediate / mulatto :15 fairly light : 6 light 1 very light/white.

When a graph is plotted for the progeny, it appears to be bell-shaped with maximum number of intermediate types and progressively lesser number towards two extremes.

S.No.	Types of non-allelic genetic interactions	Dihybrid phenotypic ratios in F_2 generation
1.	Complementary genes	9 : 7
2.	Duplicate genes	15 : 1
3.	Recessive epistasis	9 : 3 : 4
4.	Dominant epistasis	12 : 3 : 1
5.	Polymeric/Additive genes	9 : 6: 1
6.	Inhibitory genes	13 : 3
7.	Supplementary genes	9 : 3 : 4
8.	Collaborative gene action	9 : 3 : 3 : 1

10. Chromosomal basis of Inheritance

10.1 History

DNA is chemical basis, gene is physical basis, chromosome is mechanical basis of heredity. Chromosomal theory of inheritance was proposed independently by Sutton and Boveri. The two workers found a close similarity between transmission of hereditary traits and behaviour of chromosomes while passing from one generation to the next through the agency of gametes. Sutton and Boveri noted that the behaviour of chromosomes is parallel to the behaviour of Mendelian factors (Genes).

10.2 Salient features of chromosomal theory of inheritance

(1) Like the hereditary traits the chromosomes retain their number, structure and individuality throughout the life of and organism and from generation to generation. The two neither get lost nor mixed up. They behave as units.

(2) Both chromosomes as well as genes occur in pairs in the somatic or diploid cells. The two alleles of a gene pair are located on homologous sites on homologous chromosomes.

(3) A gamete contains only one chromosome of a type and only one of the two alleles of a trait.

(4) Paired condition of both chromosomes as well as Mendelian factors is restored during fertilization.

(5) Homologous chromosomes synapse during meiosis and then separate or segregate independently into different cells, which establishes the quantitative basis for segregation and independent assortment of hereditary factors.

These features explain parallelism between mendelian factors and chromosomal inheritance. Sutton united the knowledge of chromosomal segregation with Mendelian principles and called it the chromosomal theory of inheritance.

Following the synthesis of ideas, experimental verification of the chromosomal theory of inheritance by T.H. Morgan and his colleagues, led to discovery of the basis for the variations, that sexual reproduction produced.

A comparison between the behavior of chromosome and genes

A	B
Occur in pairs	Occur in pairs
Segregate at the time of gamete formation such that only one of each pair is transmitted to a gamete	Segregate at gamete formation and only one of each pair is transmitted to a gamete
Independent pairs segregate independently of each other	One pair segregate independently of another pair

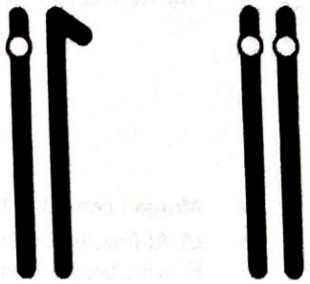
10.3 Experimental proof of Chromosomal Theory/Drosophila Genetics

Thomas Hunt Morgan (1866–1945) is known as father of experimental genetics. He was awarded Nobel Prize of physiology in 1933 for his pioneer work in experimental genetics.

(1) **Drosophila melanogaster as material for experimental Genetics** - Drosophila is more suitable than pea as experimental material because of following reasons:

- It is easily available over rotten fruits (feeding over yeast present over fruit surface).
- It is small sized (2mm) and can be cultured inside bottles having yeast culture over medium with cream of wheat, molasses & agar- agar.
- A new generation can be raised within 2 weeks. Single mating producing hundreds of flies.
- Female is distinguishable from male by its larger size and ovipositor
- It has a smaller number (4 pairs) of morphologically distinct chromosomes.
- Polytene chromosomes occur in the salivary glands of larva. Polytene chromosomes can be used to study different types of chromosome aberrations.
- It has heteromorphic (XY) sex chromosomes in the male.

The sex chromosomes are placed at first position in karyotype.



Male (XY) **female (XX)**

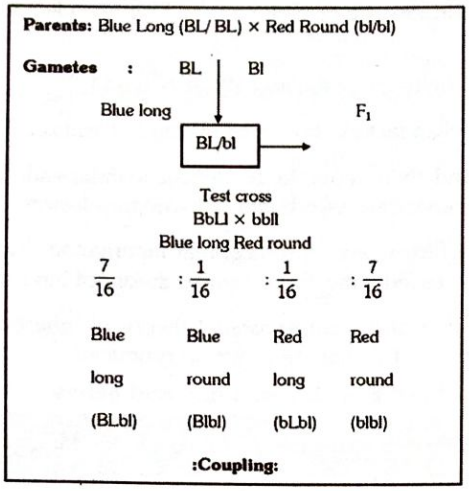
Figure :– Sex chromosomes of Drosophila

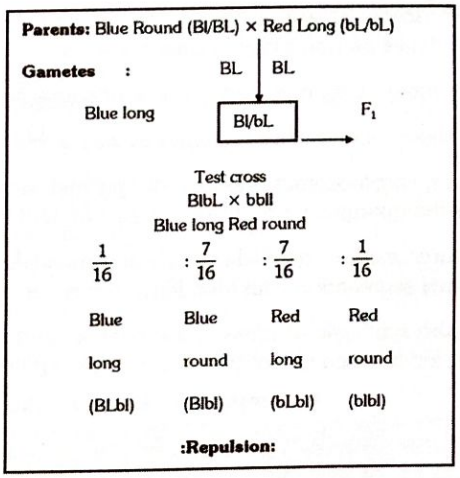
(2) **Morgan - Thomas Hunt Morgan** is popularly called father of experimental genetics. He employed Drosophila for experiment and discovered- chromosomal theory of linkage, chromosome mapping, crossing over, criss-cross inheritance and mutability of genes. Morgan was awarded Nobel Prize for physiology in 1933.

11. Linkage (Exception to Law of Independent Assortment)

According to Mendel's law of independent assortment, the gene controlling different characters get assorted independent to each other. It is correct if the genes are present on two different chromosomes, but if these genes are present on same chromosome they may or may not show independent assortment. If crossing over takes place between these two genes then the genes get segregated and they will assort independent to each other. But if there is no crossing over between these two genes there is no segregation, hence only parental combination will be found in gametes.

- The tendency of some of the genes to inherit together (en block) is known as linkage.
- Bateson and Punnet crossed two varieties of sweet pea (*Lathyrus odoratùs*) and observed that the results do not agree with the Mendel's law of independent assortment.
- They formulated the hypothesis of coupling and repulsion to explain the unexpected F_2 results of a dihybrid cross between a homozygous sweet pea having dominant alleles for blue flowers (BB) and long pollen grains (LL) with another homozygous double recessive plant with red flowers and round pollen grains (bbll).





- Test cross ratio 7 : 1 : 1 : 7 indicated that there was a tendency of the dominant allele to remain together. Similar was the case with recessive alleles. It was called as gametic coupling by Bateson and Punnet. Two dominant genes from one parent entered the same zygote more frequently than expected or the tendency of two dominant genes to remain together in the process of inheritance is called coupling (AABB × aabb).
- In another cross took a sweet pea plant with blue flowers & round pollens (BBll) and another plant with red flowers & long pollens (bbLL) and obtained ratio 1 : 7 : 7 : 1 in test cross.
- When two dominant or recessive genes comes from different parents, they tend to remain separate. This was called as repulsion or the tendency of two dominant genes from different parents to enter different zygotes was called as repulsion (AAbb × aaBB).
- T.H. Morgan in 1910 showed that coupling and repulsion are two aspects of linkage. He suggested that the two genes present on the same chromosome, are in coupling phase and when present on two different homologous chromosomes are in repulsion phase. The genes, which remain together on the same chromosome are called linked genes. The genes, which are found on different chromosomes are known as unlinked genes.

$$\frac{A \quad B}{a \quad b}$$ Coupling phase (cis arrangement)

$$\frac{A \quad b}{a \quad B}$$ Repuling phase (Trans arrangement)

- Morgan carried out several dihybrid crosses in Drosophila to study genes that were sex-linked.
- (A)At first, he crossed yellow bodies (y) and white eyed (w) female with brown bodied (y^+) red eyed (w^+) male which produced F_1 with brown bodied red eyed female and yellow bodied white eyed male. In F_2 generation, obtained by intercrossing of F_1 hybrids, the ratio deviated significantly from expected. He found 98.7% to be parental and 1.3% as recombinants.

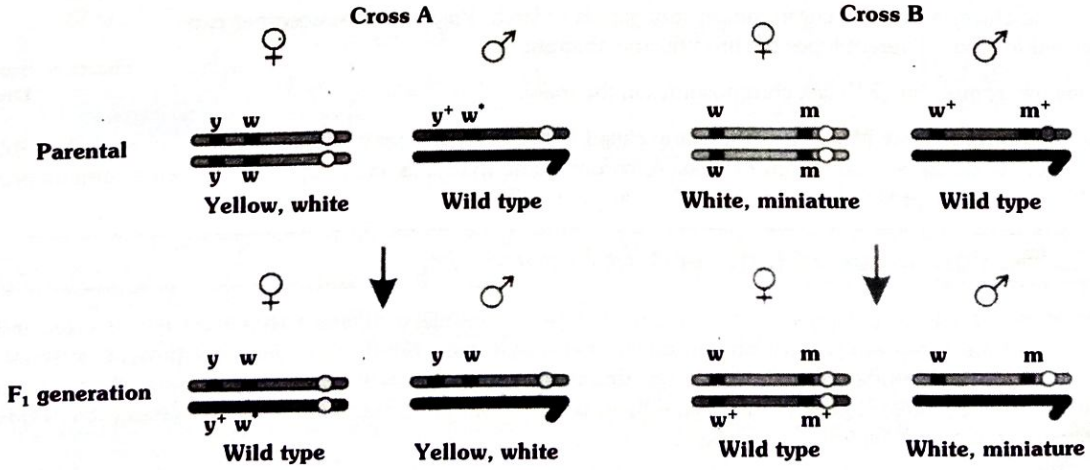


Fig : Results of two dihybrid crosses conducted by Morgan on the basis of results we can say that strength of linkage between y and w is higher than w and m

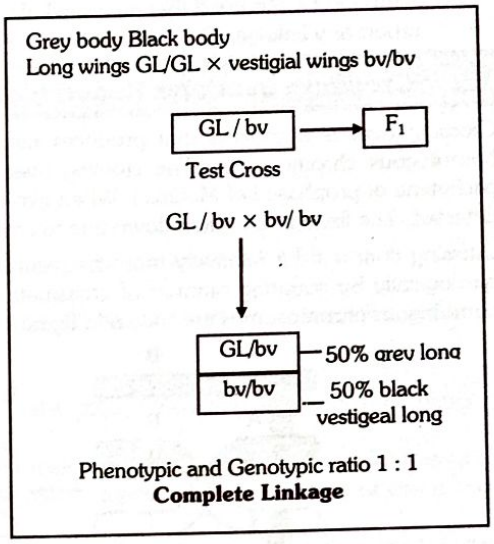
- (B) In a second cross between white eyed and miniature winged female (wwmm) with wild red eyed (w^+) normal winged male (m^+), the F_1 generation included red eyed normal winged female and while eyed miniature winged male. After intercrossing the F_1 progeny, he found 62.8% parental and 37.2% recombinants.
- According to Morgan, the degree or the strength of linkage depends upon the distance between the linked genes in the chromosome.

- Linkage, therefore, may be defined as "The tendency of two genes of the same chromosome to remain together in the process of inheritance".
- Unlinked genes (independently assorting) show dihybrid ratio of 9 : 3 : 3 : 1 and test cross ratio of 1 : 1 : 1 ; 1 while linked genes show a dihybrid ratio of 3 : 1 and test cross ratio of 1 : 1.
- Linkage is of two types, complete and incomplete.

11.1 Complete Linkage (Morgan)

Linkage of genes on a chromosome, which is not altered and is inherited as such from generation to generation without any cross over. In it, genes are closely associated and tend to remain together.

It is rare, found in male Drosophila, female Silkworm Moth and a few others. 100% parental combinations indicated that the gene for grey body colour is completely linked with long wings. In this dihybrid, F_2 phenotypic ratio is 3 : 1 and test cross ratio is 1 : 1 (like a monohybrid).



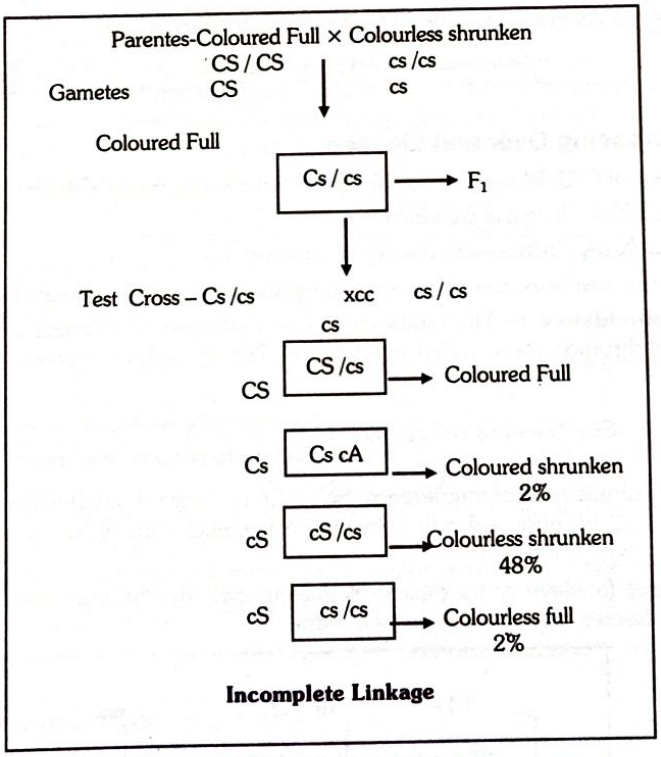
11.2 Incomplete Linkage

The linked genes do not always stay together because homologous non sister chromatids may exchange segments of varying length with one another during meiosis.

This is known as crossing over. The linked genes may have chances of separation by crossing over, are called incompletely linked genes and the phenomenon of their inheritance is called incomplete linkage. It produces both parental and recombinant types in variable ratio.

It is tendency of linked genes to separate to form recombinant type due to crossing over. In Maize, Hutchinson obtained dihybrids (CcSs) from a cross of homozygous dominant coloured full grained (CCSS) plant with colourless shrunken grained plant (ccss).

Hybrids were test crossed with recessive parent (ccss). Parental types (coloured full, colourless shrunken) occurred in 96.4% while recombinant types (coloured shrunken, colourless full) appeared in 3.6% cases. F2 ratio is 27 : 1 : 1 : 27



11.3 Chromosome Theory of Linkage : (By Morgan & Castle)

(1) Genes present on same chromosome, don't assort -independently but stay together during transmission (linkage).

(2) Genes present linearly over chromosome,

(3) Strength of linkage is inversely proportional to distance between two linked genes.

11.4 Linkage Group

A linkage group is a group of linearly arranged linked genes on a chromosome, which are inherited together as a single unit.

Number of linkage group in an organism is equal to 'n' number of chromosome, e.g., it is 12 in rice, 7 in Pisum, while in human beings, the number of linkage group is 23 in human female and 24 in human male and in Drosophila it is 4.

11.5 Mendel vs. Linkage

Mendel studied 7 character/gene /pairs of alleles, located on four out of 7 chromosome. Two of the chosen genes are located on first chromosome. They are so distantly located that they undergo regular crossing over. It results in 50% recombinations, which is equal to independent assortment. Chromosome 4 had three characters picked up by Mendel. Two of the three were distantly located and resulted in 50% recombination or no linkage.

Although he observed that grey seed always produces red flowered plant and white seed produces white flowered plant yet he did not report any linkage. They are not mentioned by Mendel. Probably, he did not make such cross & did not detected linkage.

12. Crossing over and Recombination:

Crossing over is a process that produces new combination of genes by interchanging of segments between non sister chromatids of homologous chromosomes. The crossing over occurs in between the homologous chromosome at four stranded or tetrad stage during pachytene of prophase I of Meiosis I. When two genes are located very close to each other in chromosomes, hardly any crossing over can be detected. The linkage is broken down due to crossing over.

Crossing over will be relatively more frequent if the distance between two genes is more. Frequency of crossing over can be determined cytologically by counting number of chiasmata. The details of the crossing over for two genes A and B and their alleles a and b on the homologous chromosomes are shown in figure.

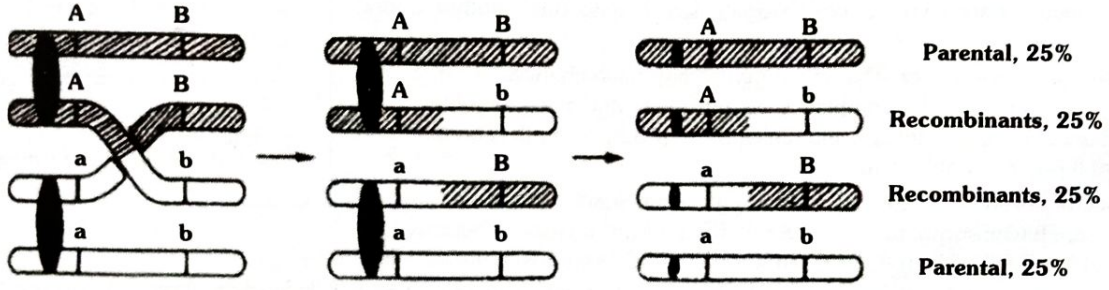


Figure :– Crossing over showing 50 per cent parental and 50 percent recombinants

The crossing over as shown above results in the formation of following four types of cells:

Parental combinations (Non–cross overs) $\left.\begin{matrix}AB\\ab\end{matrix}\right\}$ 50% Recombinants or Non–parental combinations (Cross overs) $\left.\begin{matrix}Ab\\aB\end{matrix}\right\}$ 50%

12.1 Factors Influencing Crossing Over and Linkage

(1) Distance :– Frequency of C.O. increases and strength of linkage decreases with increase in distance between genes.

(2) Temperature :– Fluctuations increase crossing over.

(3) X-rays :– Exposure' to X-rays increases frequency of crossing over.

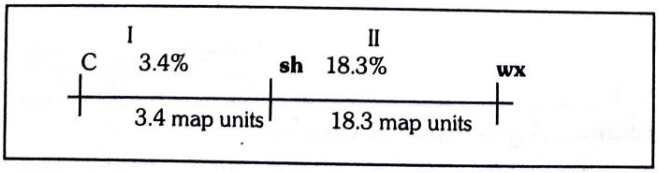
(4) Sex :– In heterogametic organisms, crossing over is sometimes very little, e.g., male Drosophila..

(5) Interference and coincidence :– The tendency of one cross over to interfere with the other cross over in an immediately adjacent region of the chromosome is called interference. The strength of interference is expressed in terms of a coefficient of coincidence.

$$\text{Coefficient of coincidence} = \frac{\%\,\text{observed double cross over}}{\%\,\text{expected double cross over}}$$

The coincidence is the complement of interference, So – (Coincidence + Interference = 1). When interference is complete or 1, no double cross overs will be observed and coincidence becomes zero. When interference decreases, coincidence increases. Coincidence values vary between 0 and 1.

Example: Map distances for three genes c (colourless aleurone), sh (shrunken grains) and wx (waxy endosperm) of corn and observed crossing over frequencies between the genes are.



Crossing over frequencies between genes C, sh and wx of corn

Region	Genes	% Cross overs	Map distances
I	C-sh	3.4	3.4
II	Sh-wx	18.3	18.3
Double cross over	c-sh-wx	0.1	

If crossing over in region I and II were independent, we should predict 0.034 × 0.183 = 0.06 or 6% double cross overs, where as only 0.1% was observed.

So coincidence $= \frac{0.1}{0.6} = 0.167$

Interference $= 1 - 0.167 = 0.823$ or 82%

12.2 Types of crossing over

(1) **Single Crossing Over :–** At one point , resulting in two cross-overs and two parental types

(2) **Double Crossing Over :–** At two points in a tetrad of chromatids

(3) **Reciprocal :–** At two points between the same nonsister chromatids.

(4) **Complementary :–** Three or all the four chromatids are involved in double crossing over.

(5) **Multiple Crossing Overs :–** At three or more places in the same tetrad of chromatids.

(6) **Somatic Crossing Over :–** Between homologous chromosomes that occurs in somatic cells without meiosis. Chromatids often become unequal, e.g., Aspergillus, Penicillium.

12.3 Independent Assortment and 50 % Recombinations

Independent assortment of genes/factors occur under two conditions.

(1) Occurrence of genes on different/non-homologous chromosomes.

(2) Genes found on same chromosome but at a distance, which allows one crossing over between them at every meiosis.

12.4 Crossing Over and Recombination Frequencies:

One crossing over produces 50% recombinant types. So, frequency of crossing over would be double the frequency of recombinants. 1% recombinants (1% cross-overs) means crossing over in 2% meiocytes.

12.5 Recombinations

They are genetic variations, which appear due to, reshuffling of genes in linkage groups resulting in change of genotypes. Recombinations can occur due to

(1) **Independent Assortment of Chromosomes :–** This occurs at the time of meiosis/gametogenesis. Independent assortment can produce 2^n recombinations (n= no. of pairs of chromosome).e.g. 2^{23} or 86 million types in case of 23 pairs of chromosomes in both male and female gametes.

(2) **Random Fertilization :–** Any of the possible recombination in male gamete can pair with any of the possible recombination in female gamete so that the chance of chromosome recombination multiplies. e.g. $2^{23} \times 2^{23}$ (8.6 million X 8.6 million or 7×10^{12}) in case of 23 pairs of chromosomes in human.

(3) **Crossing Over :** Exchange of segments between nonsister chromatids.

12.6 Chromosomal Mapping

Graphic representation of relative positions/order and relative distances of genes in a chromosome in the form of line like a linear road map depicting different places and their relative distances without giving exact mileage.

Crossing over is important in locating the genes on chromosome. The genes are arranged linearly on the chromosome. This sequence and the relative distances between various genes is graphically represented in terms of recombination.

frequencies or cross over values (COV). This is known as linkage map of chromosome. Distance or cross over units are called centimorgan (cM) or map unit. Term centimorgan is used in eukaryotic genetics and map unit in prokaryotic genetics. Cross-over value (COV)/ recombination frequency is measured by test cross. It is multiplied by one hundred to obtain percentage recombination frequency or cross-over units. Cross-over units are also called centimorgans (cM) or map units. 10% crossing over is called decimorgan (dM) while 100% crossing over is known as Morgan (M).

$$\text{Recombination frequency or cross over value} = \frac{\text{Number of recombinants}}{\text{Total number of offsprings}} \times 100$$

The recombination frequency depends upon the distance between the genes. If the distance between the genes is lesser the chances of crossing over is less and hence recombination frequency is also lesser and vice versa.

(1) **Map unit or centimorgan :–** It is equivalent to 1% recombination between two genes. So, recombination frequency is directly proportional to distance between genes. In any cross, if recombination frequency is 15%. It means the distance between the genes is 5 map unit. A.H. Sturtevant suggested that these recombination frequencies can be utilized in predicting the sequence of genes on the chromosome. On the basis of recombination frequency, he prepared first chromosomal map or genetic map for Drosophila.

13. Sex Determination

The factors which control the sex of an organism are under genetic control. Various mechanism which led to sex determination can be classified into following four categories.

(1) Environmental mechanism of sex determination.

(2) Non – Allosomic genetic sex determination – Fertility factor (plasmid) in bacteria.

(3) Chromosomal mechanism of sex determination.

(4) Genic balance mechanism or X/A balance.

13.1 Environmental Determination of Sexes

Environment determines sex in some potentially hermaphrodite animals. Crepidula becomes female when developed alone but produces male in company of a female. Bonellia turns female in isolation and develops into male if get attached to female.

13.2 Non-allosomic Genetic Determination of Sex

In bacteria 'sex' determination is controlled by fertility factor or plasmid. Sex is determined by a single gene in Chlamydomonas. Maize has two genes, one for male inflorescence and second for female inflorescence.

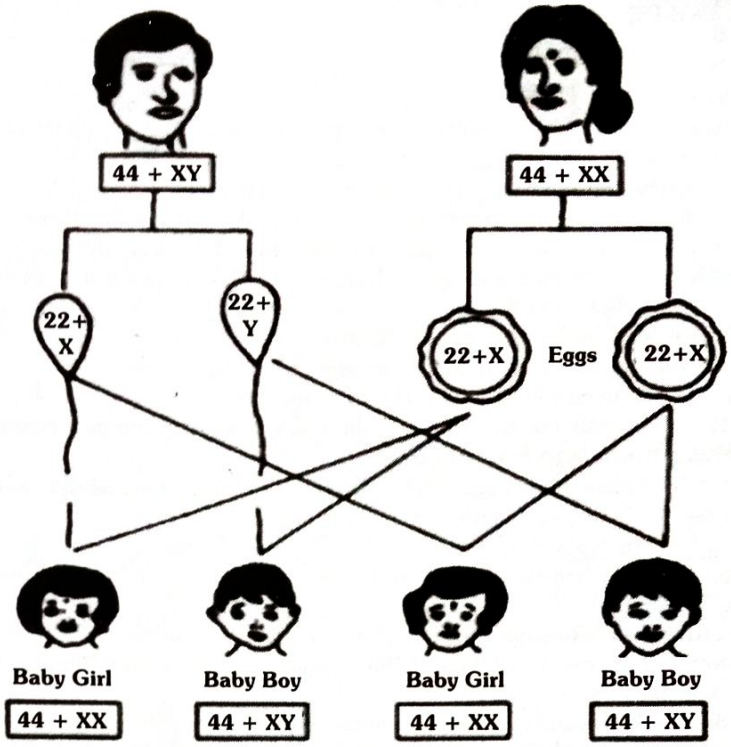


Figure :– Sex-determination in man.

Note that all the eggs carry X-chromosome but one-half of the sperms carry an X-chromosome and one-half carry a Y-chromosomnee.

13.3 Allosomic Determination of Sex

In most animals and some plants, sex is determined by sex chromosomes. Sex is decided at the time of fertilization, called syngametic sex determination.X-body or X-chromosome was discovered by Henking, when he found that in the testes of male bug one chromosome has no homologue. Stevens discovered Y-chromosome.

Mc Clung observed that in Squash Bug (Anas), male has 21 chromosomes while female has 22 chromosomes. He stressed that X-chromosome has a role in sex determination.

Wilson and Stevens gave "chromosome theory of sex" with X and Y-chromosomes being named as sex chromosomes.

Correns put forward the theory of heterogamesis. According to this theory one of the two sexes has different types (heteromorphic) of sex chromosomes. It produces two different types of gametes (androgamete and gynogamete). The other sex has homomorphic sex chromosomes. It produces one type of gametes or homogametes.

Sex of the offspring is determined by the type of heterogamete fusing with the homogamete.

(1) XY Method (XX-XY) :– It occurs in mammals and many insects, females having homomorphic XX sex chromosomes and males having heteromorphic XY-chromosomes. In human beings the Y-chromosome is acrocentric. It is shortest. X- chromosome is metacentric. Although differ in morphology yet XY chromosomes pairs during zygotene.

They have homologous and differential parts. Homologous parts take part in synapsis They carry similar genes, called X-Y linked genes, e.g. xeroderma pigmentosum, epidermolysis bullosa. The differential or non-homologous region of Y-chromosome is mostly heterochromatic.

Euchromatic part of differential region of Y- chromosome has holandric genes like TDF (testis determining factor) or SRY (sex determining region), ZFY (sperm formation), P□2V, hypertrichosis (excessive hairiness), webbed toes.

SRY/TDF is smallest gene (14 basepair). These are transferred directly from father to son.

X-chromosome carries some female determining genes like Sxl They are inhibited when SRY becomes functional.

Genes present on differential region of X-chromosome (sex-linked traits) express in males whether they are recessive or dominant because males are hemizygous e.g., red-green colour blindness, haemophilia.

Male is heterogametic. Two types of male gametes are called androsperms (22 AA+ Y) and gymnosperms (22 AA+ X). While female is homogametic (22 AA + X in human).

Y-chromosome of father is directly transferred to male offspring while its X chromosome is passed to the female offspring. Male offspring receives its X-chromosome from mother.

Female offspring receives its two XX-chromosomes from the two parents, one from each.

Because Y-chromosome determines the male sex, it is also called androsome XY sex determination is found in many bryophytes (females A + X, males A + Y}.

Amongst angiosperms XY sex determination is found in Mclandrium (Lychnis), Coccmia, Salix etc.

(2) **ZW Method (ZW-ZZ or WZ-WW)** :– In birds, some reptiles and fishes, females heterogametic (A + Z, A + W). Males are homomorphic sex chromosomes (ZZ) & hence homogametic (A+ Z). Fragana elelior is the plant having ZW sex determination.

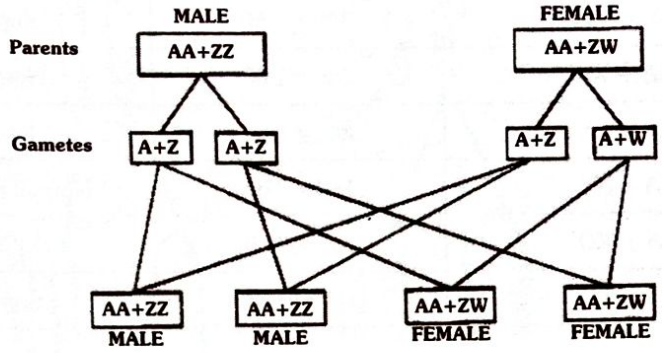


Figure :– ZW – ZZ determination of sex in chicken

(3) **XO Method (XX -XO)** :– In roundworms and certain insects (true bugs, grasshoppers, cock-roaches) the females have two sex chromosomes, XX while the males possess a single sex chromosome X. Females are homogametic (A-X). The males are heterogametic producing androsperms (A + Zero) and gynosperms (A + X). Amongst angiosperms, Dioscorea smuata has XX - XO sex complement.

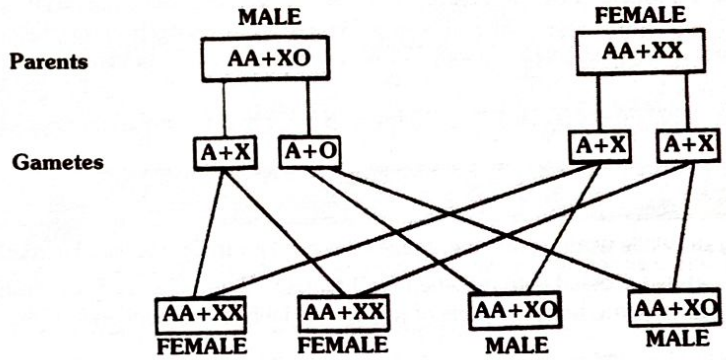


Figure :– XX – XO determination of sex in Cockroach / Grasshopper

(4) **ZO Method (ZO-ZZ)** :– In moths and butterflies, females have odd sex chromosomes (AA + ZO) while males have two homomorphic sex chromosomes (AA + ZZ). Females are heterogametic (A+ Z, A+Zero) and male homogametic (A+ Z).

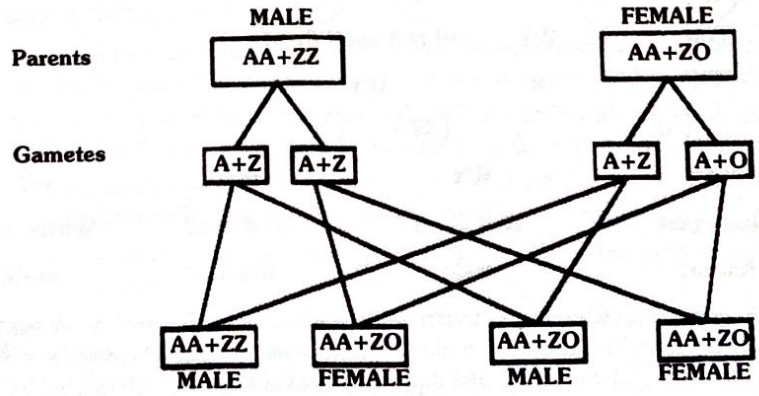


Figure :– ZO – ZZ determination of sex in butterfly

(5) **Haplodiploidy (Honey Bee Method) Arrhenotoky Mechanism**: In this one sex is haploid while other is diploid. In queen bee unfertilised eggs develop into drones or males (arrhenotoky or parthenogenesis.) Workers and queen bee develop from fertilised eggs and are sexually female.

13.4 Genic Balance or X/A Balance theory of Sex Determination

Given by Bridges. According to him, Y chromosome plays no role in sex determination of Drosophila and it is ratio between number of X-chromosome and set of autosomes which determines sex of fly.

Gynandromorphs :– It is a sex mosaic (an individual with one half of the body male and the other half female). These are common in Silk moth and Drosophila. Gynandromorphism is developed due to accidental loss of X-chromosome from a 2A + XX cell during mitosis.

13.5 Sex determination in plants

Monoecious plants have no sex chromosomes. Unisexual flowering plants have XX-XY type of sex chromosomal for sex determination, e.g. *Melandrium* (*Lychnis*) and *Sphaerocarpos*.

Chromosome Constitution	X/A ratio	Sex Index
AA + XXX	3/2 = 1.50	Super ♀
AA + XX	2/2 = 1.00	Normal ♀
AAA + XXY	2/3 = 0.67	Intersex
AA + XY	1/2 = 0.50	Normal ♂ (Fertile)
AA + XO	1/2 = 0.50	♂ (Sterile)
AAA + XY	1/3 = 0.33	Super ♂

Important-

Barr Body and Y Spots

Barr and Bertram (1949) found a small darkly stained chromatin body (heterochromatic) adhered to the nuclear membrane of nerve cells of female cats but not in male cats. Later on, Barr (1960) observed consistent presence of these chromatin bodies in epidermal cells of buccal and vaginal mucosa, skin cells etc. in females.

Lyon (1962) suggested that one X chromosome of maternal side becomes coiled, inert and hetero-pycnotic and forms a barr body. Number of barr bodies is always one less than the total number of X chromosomes in female. Normal male has no barr body.

In about 5% of polymorphonuclear leucocytes (neutrophils) in women, the heteropycnotic X chromosome occurs as a round pedunculated body attached to the nucleus. It is called drum stick (Barr body). A normal female has 1 drum stick per nucleus of neutrophil. In neutrophils of males drum stick is absent.

Y spots: Males are identified by number of Y spots (Zech 1970). Number of Y spots are equal to number of Y Chromosomes.

14. Sex Linked Inheritance

Sex linkage was discovered by **Morgan,** while working on inheritance of eye colour in Drosophila. He made three types of crosses :

Cross – 1 :– The white eyed male (w) was crossed with red eyed (w^+) female. All the flies of F_1 generation were found to be red eyed. F_1 flies were allowed to self breed. In F_2 generation, both the traits of red eye and white eye appeared in the ratio 3 : 1 showing that white eye trait is recessive to red eye trait.

Parents WW × wY

(Red eyed female) (White male eyed)

Gametes (W) (w) (Y)

F1 Ww WY (all red eyed flies)

F2 Ww × WY

Gametes (W) (w) (W) (y)

Progeny	WW	WY	Ww	wY
	Red eyed female	Red eyed male	Red eyed female	White eyed male

Cross – 2 :– Red eyed females of F_1 generation were crossed with white eyed male. **It is similar to test cross** where hybrids are cross bred with recessive parents. He obtained red and white eyed female as well as male in equal proportions – 1 red eyed female : 1 white eyed female : 1 red eyed male : 1 white eyed male. Test cross indicated that white eye colour was not restricted to male fly.

Parents Ww × wY

(Heterozygous female) (White eyed hemizygous male)

Gametes (W) (w) (w) (Y)

Progeny	Ww	WY	ww	wY
	Red eyed female	Red eyed male	White eyed female	White eyed male

Cross – 3 :– White eyed females were crossed with red eyed males. It was a reciprocal of cross 1 and should give the similar results as obtained by Mendel. However, Morgan obtained a surprising result. All the males were white eyed while all the females were red eyed.

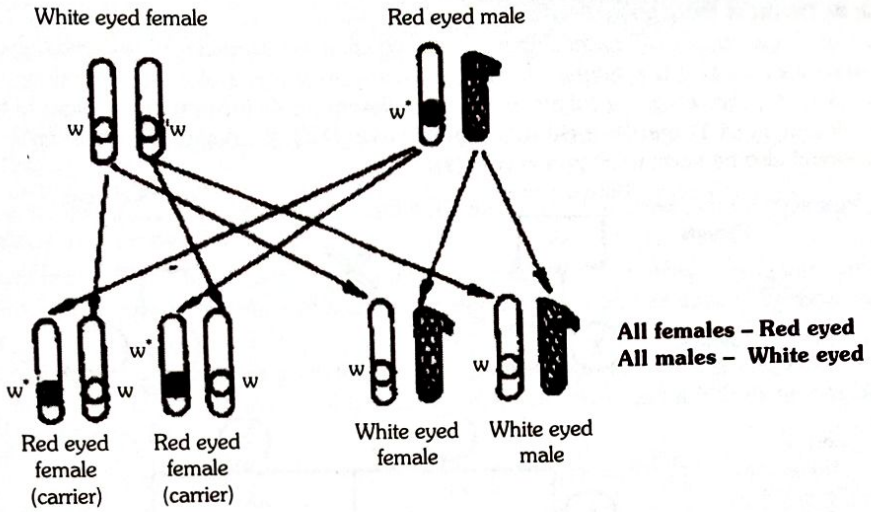


Figure :– Cross 3 of Morgan – Involving white eyed female and red eyed male showing criss-cross inheritance in *Drosophila*

Conclusion

Taking all the crosses into consideration, Morgan came to the conclusion that eye colour gene is linked to sex and is present on the X-chromosome. X-chromosome does not pass directly from one parent to the offspring of same sex but follows a criss-cross inheritance, i.e., it is transferred from one sex to the offspring of the opposite sex. Although according to Mendel, all offspring should have been red-eyed. But presence of white eye colour in F1 males shows that they must be carrying a single gene in males and double in females. Y-chromosome of males does not carry this gene. From this, Morgan predicted Red eye colour is dominant X- linked trait, criss-cross inheritance of sex-linked genes—father to daughter and then to grandson.

14.1 Criss-Cross Inheritance (Morgan)

It is a type of sex-linked inheritance where the genes of one parent are transferred to the grand children through children of opposite sex. Criss-cross inheritance is of two types :

(1) Diagynic :– Transfer of trait from male parent to grandson through daughter.

(2) Diandric :– In diandric criss-cross inheritance there is transfer of sex-linked trait from mother to grand daughter through her son.

14.2 Non-Criss-Cross Inheritance

It is autosomal as well as sex-linked. Sex-linked non criss-cross inheritance is holandric (if it passes directly from father to son) and hologynic (if it passes directly from mother to daughter).

14.3 Non-disjunction (Bridges, 1916)

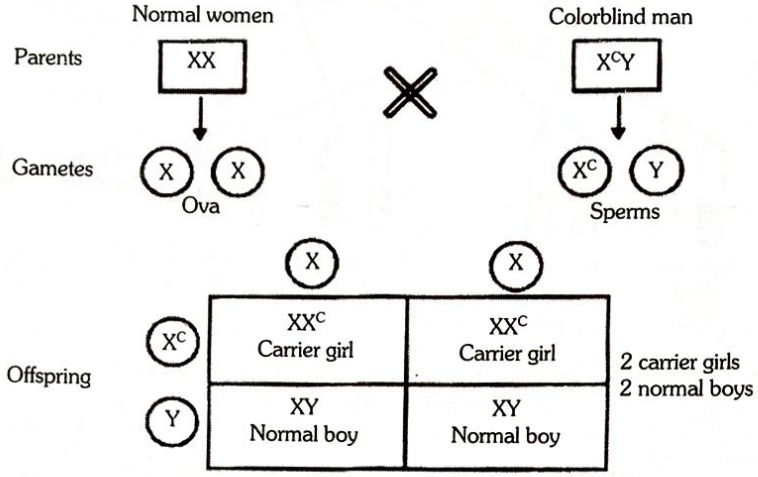
- Discovered by Bridges, when he found that an occasional white eyed female had a chromosome complement of XXY. Here XO is male. Occurrence of gene for red-white eye colour on X-chromosome was confirmed by Bridges.
- Like the cross III of Morgan, Bridges mated white eyed female Drosophila with red eyed male fly. All the males were white eyed and all the females red eyed (as found out by Morgan) except for an occasional while eyed female (hologynic inheritance).
- Study of karyotype of such white eyed female showed that it had XXY sex chromosome complement due to failure of X-X chromosome disjunction at the time of oogenesis so that instead of white eyed male it produced white eyed female.
- This also proved that X-chromosomes carries gene for red-white eye colour.Non-separation of homologous chromosomes during anaphase is called non-disjunction.
- It is of two types, meiotic non-disjunction (failure of synaptized homologous chromosomes to separate during anaphase I) and mitotic non-disjunction (failure of daughter chromosomes to separate during mitotic anaphase).
- Initial non-disjunction is called primary non-disjunction. It remains in the progeny.
- The presence of non-separated chromosomes in the progeny due to previous non-disjunction is called secondary non-disjunction. e.g., Bridges obtained a number of abnormal flies in the progeny of XXY like XXY, XXX, XYY, etc.

S.No.	Sex limited traits	Sex influenced traits	Holandric traits
1.	The genes of these traits are autosomal and found in both sexes but express in one sex only.	These are those autosomal genes which are influenced by the sex of the bearer. These traits appear more frequently in one sex than in the other.	These are Y-linked traits those inherit from male to male only.
2.	**Examples :** (i) Milk glands in female (ii) Beard in man (iii) Deep male voice (iv) Female or male musculature	**Examples :** (i) Pattern baldness (affected by male sex hormone /testo-sterone) (ii) Short finger in male	**Examples :** (i) Porcupine skin (ii) TDF (Testes determining factor) (iii) Hypertrichosis

14.4 Sex Linkage in Human Beings

Colour blindness and haemophilia (Bleeder's disease) are two common examples of sex-linked diseases in human beings.

(1) Colour blindness :– This is a human disease which causes the loss of ability to differentiate between red colour and green colour. The gene for this red-green colour blindness is present on X chromosome. Colour blindness is recessive to normal vision. If a colour blind man (X^cY) marries a girl with normal vision (XX), the daughters would have normal vision but would be carrier, while sons would also be normal (Shown in cross(a)).



Cross (a)

If the carrier girl (heterozygous for colour blindness, X^cX) now marries a colour blind man X^cY, the offspring would show 50% females and 50% males. Of the females, 50% would be carrier for colour blindness and the rest 50% would be colour blind. Of the males, 50% would have normal vision and the 50% would be colour blind (shown in cross (b)).

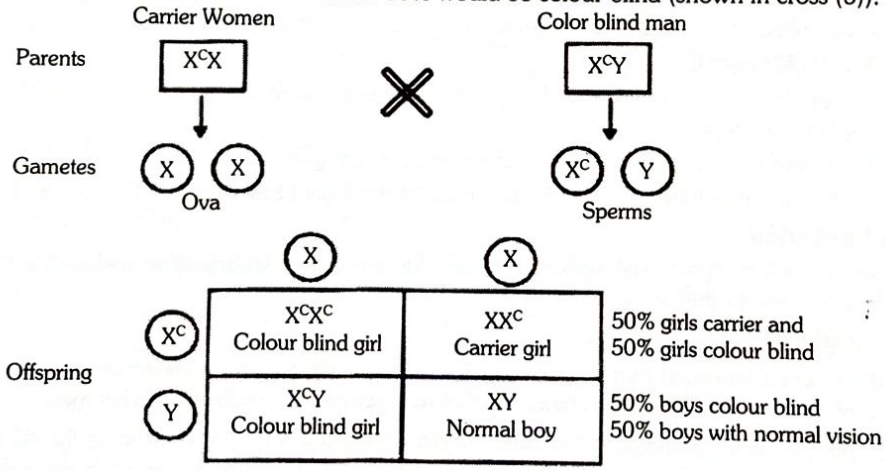


Figure :– Sex-linked inheritance of colour blindness – cross (a) and cross (b)

Cross (b)

(2) Haemophilia (Bleeder's disease) :– The person suffering from this disease cannot synthesize a normal blood protein called antihaemophilic globulin (AHG) required for normal blood clotting (Haemophilia A more severe). Therefore, even a very small cut may lead to continuous bleeding for a long time. This gene is located on X chromosome and is recessive. It remains latent in carrier females.

If a normal man marries a girl who is carrier for haemophilia, the progeny would consist of 50% females and 50% males. Of the females, 50% would be normal and the rest 50% would be haemophilia carrier. Of the males, 50% would be normal and rest would be haemophiliacs.

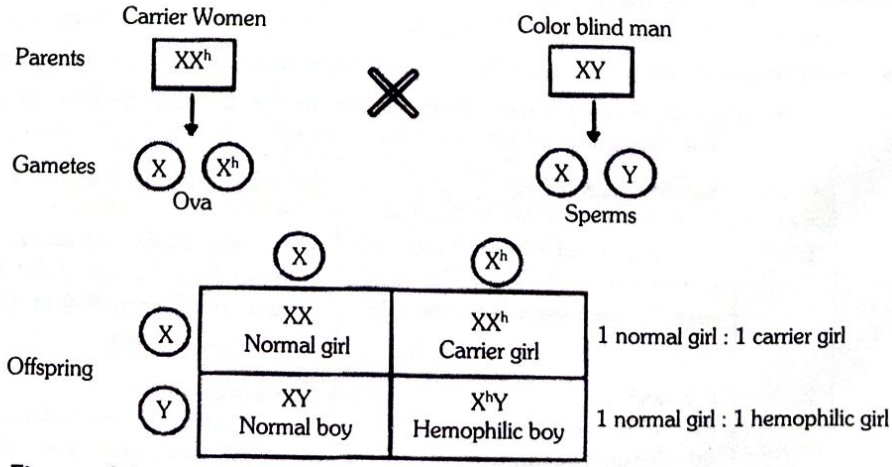


Figure :– Inheritance of hemophilia when the mother is carrier and the father is normal

15. Mutation

Mutations are sudden stable inheritable. Discontinuous variations, which appear in organisms due to permanent change in their genotypes (chromosomes and genes.). Mutation was termed by Hugo de Vries, sports by Darwin and saltations or discontinuous variations by Bateson, Shot gun by Moody. De vries proposed mutation theory of evolution. He observed 834 mutations in a population of 54343 giants of Oenothera lamarkiana (Evening Primrose).

- Genotype can change due to changes in chromosome number (Genomatic Mutations), chromosome structure (chromosomal aberration) and gene mutations (point mutation).
- Seth Wright (1971) is considered to be the first to record point or gene mutation. He noticed a lamb with unusually short legs. This short legged breed of sheep was known as ancon breed. Darwin called this variation as sports. Bateson (1894) termed them as discontinuous or salutatory variations.
- The credit for starting scientific study of mutations goes to Thomas Hunt Morgan (1910). He is known for his work on fruit fly, Drosophila melanogaster. He found white eyed mutant of Drosophila and since then about 500 mutations have been observed by geneticists from different parts of the world.

15.1 Types of Mutations

(1) On the basis of nature

- **Spontaneous and induced mutations :–** Spontaneous mutations are natural mutations. They have also been called background mutations. Such mutations occur at a frequency of 1×10^{-5} in nature. Induced mutations are caused in organisms due to specific factors such as radiations, ultra violet light or variety of chemicals. The agents which induce mutations on their application, are called mutagens or mutagenic agents.

(2) On the basis of type of cells in which mutations occur

- **Somatic mutations :–** These mutations occur in somatic cells, i.e., body cells or the cells other than germinal cells. The somatic mutations do not have any genetic or evolutionary importance. This is because only the derivatives or the daughter cells formed from the mutated cell will show mutation and not the whole organism.
- **Germinal mutations :–** These mutations occur in the gametes or germ cells and are also known as gametic mutations. Such mutations are heritable, and, therefore, are of great evolutionary significance. If the mutations are dominant, these are expressed in the next generation and if recessive, their phenotypic expressions remain suppressed.

(3) Forward and backward mutations :– Forward mutations is commonest type. Forward mutations is change from normal or wild type to new genotype (recessive or dominant). An organism which has undergone forward mutation, may again develop mutation which restores the original wild-type phenotype. Such reversions are known as backward mutations or reverse mutations. Mutations can occur at any stage during the life cycle of an organism.

(4) Genomatic Mutations :– Change in chromosome number. It is of two types.

- **Polyploidy/Euploidy :–**

It is presence of more than two genomes or sets of chromosomes, e.g., 3n (triploid), 4n (tetraploid), 5n (pentaploid), etc. Odd Polyploids are sexually sterile and propagated vegetatively. e.g. Banana, Pineapple. It can develop due to **(i)** Failure of spindle development. (ii) Failure of meiosis I. (iii) Fusion between unreduced gametes, unreduced and normal gametes, one egg with more than one sperm.

A monoploid has only one set of chromosomes Eg :- Made of bee's

A polyploidy state in which chromosome have dinided without division of nucleus is called endo polyploidy Eg. Human Muscle tissue.

Polyploidy is induced by (a) Decapitation, (b) High or low temperature, (c) Mechanical injury, (d) Infection, (e) Irradiation and (f) Chemicals like colchicine, granosan, sulphanilamide etc.

Polyploidy is of three types - autopolyploidy, allopolyploidy and autoallopolyploidy.

(a) Autopolyploidy :– It is the increase in number of the same genome, e.g., AAA, AAAA.

Banana is autotriploid. Seedless autotriploid variety of Water Melon has been evolved.

Triploid Sugar beet has more sugar content Colchicine induces autotetraploidy.

(b) Allopolyploidy :– It is the increase in number of chromosome sets due to coming together of genomes of two or more species-hence interspecific polyploidy, e.g., AABB AABBDD.

Allotetraploids (AABB) are also called amphidiploids when Allopolyploids produced due to hybridisation b/w two species followed by doubling of chromosomes e.g., Wheat (Durum AABB, Bread Wheat AABBDD), American Cotton, Nicotiana.

(c) Autoallopolyploidy :– Increase in chromosome sets due to coming together of different genomes and occurrence of one genome in more than diploid state, e.g., AAAABB (Helianthus tuberous}.There is a gigas effect with larger size and often higher yield.

Aneuploidy refer to numerical change in past of chromosome set & polyploidy refer to change in whole set of chromosome

- **Aneuploidy (Heteroploidy)**

It is the deletion or addition of one or a few chromosomes from the original genomes. It develops due to nondisjunction of homologous chromosomes in meiosis. It is of three type :

(a) **Hypoploidy** :– It is aneupoidy with loss of chromosomes. e.g., Monosomic, nullisomic.

(b) **Hyperploidy** :– It is aneuploidy with addition of chromosomes. e.g. Trisomic tetrasomic, pentasomic

(i) **Monosomic** :– It is an aneuploid in which a chromosome is deficient of its homologue causing genetic imbalance resulting in weaker than normal form, Monosomic is formed by the union of deficient gamete (n-1) and normal gamete. A sex chromosome monosomic in human beings is Turner's syndrome (44 + X). In double monosomic two different chromosomes are deficient of their homologues (2n - 1 - 1).

(ii) **Nullisomic** :– It is an aneuploid in which a pair of homologous chromosomes is deficient (2n -2). Nullisomy is lethal in diploid organisms but can survive in polyploidy. It is produced by the union of two gametes deficient in the same chromosome (n-1) x (n-1).

(iii) **Trisomic** :– Aneuploid which has one chromosome represented in triplicate (2n + 1).

It is formed by union of a normal gamete (n) with a gamete having an extra chromosome (n + 1).

Trisomics show gigas effect for certain genes and abnormality for others.

All the possible trisomics have been studied in Datura.

Double aneuploidy is change in number of two homologous chromosome pairs (e.g., 2n-1-1) while triple aneuploidy is changed chromosome number for three homologous chromosome pairs (e.g., 2n + 1 + 1 + 1).

(iv) **Tetrasomic** :– It is an aneuploid in which one chromosome is represented in quadruplicate. Tetrasomic is formed by union of two gametes each having the similar extra chromosome, (n + 1) x (n + 1). Two chromosomes are represented in quadruplicate in double tetrasomic (2n +2+2) and three in triple tetrasomic (2n+ 2 + 2 + 2).

(v) **Pentasomic** :– An aneuploid with one chromosome represented five limes (2n+ 3), e.g., a rare super female in human beings (44 + XXXXX).

(c) **Mixed Aneuploidy** :– It is an aneuploid with both hypoploidy (deficiency) and hyperploidy (addition), e.g., 2n+ 1A- 1B.

S. No.	Parent – 1	Parent – 2	Hybrid Product – 1* (Sterile)	Product –2 Produced by Chromosomal doubling	
1.	Radish *Raphanus sativus* *2n = 18*	Cabbage *Brassica oleracea* *2n = 18*	Diploid *Raphanobrassica* *2n = 18 (Sterile)*	Tetraploid *Raphanobrassica,2n = 36 (Fertile)* (I allopolyploide by Karpechencho)	Man Made
2.	Tetraploid wheat *Triticum durum* *2n = 28*	Rye *Secale cereal* *2n = 14*	Triploid *Triticale* *2n = 21 (Sterile)*	Hexaploid *Triticale* *2n = 42 (Fertile)* (by Muntzing)	
3.	Hexaploid wheat *Triticum aestivum* *2n = 42*	Rye *Secale cereale* *2n = 14*	Tetraploid *Triticale* *2n = 28 (Sterile)*	Octaploid *Triticale* *2n = 56 (Fertile)*	Natural product
4.	Old world cotton *Gossypium herbaceum* 2n = 26	American cotton *Gosspium raimondii* *2n = 26*	New world cotton *Gosspium hirsutum* *2n = 26 (Sterile)*	Tetraploid *Gossypium hirsutum* *2n = 52 (Fertile)*	

(5) Chromosome Aberrations

It is changes in chromosome morphology, which result in changes in number and sequence of genes without altering ploidy or gene structure. Chromosome aberrations involve breaking of chromosome segments, their loss or union with same (intra chromosomal aberration) or different chromosomes (inter chromosomal aberration).

The important types are as follows:

- **Deficiency** :– It is loss of chromosome segment. The lost segment may be terminal (deficiency or terminal deficiency) or intercalary (intercalary deficiency or deletion). Deficiency and deletion are interchangeable term. Terminal deficiency involves a single break near chromosome end. In intercalary/interstitial deficiency or deletion, chromosome breaks from two points (double break), separated segment is lost and the points of breaks get united. In heterozygous deficiency (deficiency in one out of two chromosomes of a homologous pair), synapsed chromosomes would show difference in length at one end (terminal deficiency) or loop (intercalary deficiency/deletion). Heterozygous deficiency may causes expression of recessive gene, is called pseudodominance.
- **Duplication** :– Presence of genes in excess of normal complement due to presence of extra chromosome segment.

(i) **Tandem/Repeat Duplication** :– Duplicating segment attached next to normal corresponding segment e.g. ABCDEFEF.

(ii) **Reverse Tandem Duplication** :– The duplicating segment is incorporated next to normal corresponding segment but in reverse order e.g., ABCDEFGHHG.

(iii) **Displaced Duplication** :– Duplicating segment is attached away from corresponding segment on same chromosome.

(iv) **Transposed Duplication :–** Duplicating segment is joined in a non homologous segment

Duplication increases the number of genes which increases genetic redundancy, allows development of variations and hence evolution. However, some duplications are harmful, e.g., development of bar eye in Drosophila.

- **Inversion :–** A chromosome segment having one or more genes gets separated and reinserted in its position but after inversion or rotation by 180° so that the order of genes is reversed. Inversion may include centromere (pericentric) or occur beyond centromere (paracentric).
- **Translocation :–** In it a chromosome segment separates and gets attached to a non homologous chromosome.
  - (i) **Reciprocal Translocation :–** Mutual exchange of segments b/w non homologous chromosomes.
  - (ii) **Simple Translocation :–** One sided translocation.

    If a segment inserted in interstitial position of a non homologous chromosome, called interstitial or shift translocation.

(6) Gene Mutations / Point mutations

They are sudden stable changes in the structure of gene or cistron due to change in nucleotide type and nucleotide sequence. Most of gene mutations involve a change in single nucleotide. Ancon Sheep was first recorded mutation (Seth Wright) followed by Hornless (Polled) Cattle.

Scientific study of mutations started with the noticing of white eye colour in Drosophila (Morgan).

A mutation from wild to a new type is called forward mutation. The reversal of mutated gene back to its original/wild form is called reverse/back mutation. Mutations affecting vegetative cells are called somatic mutations. They are not inheritable. In plants, somatic mutation can be passed to future generations through vegetative propagation & tissue culture. Germinal mutations occur in sex cells and are inheritable. These may be dominant or recessive. Mutations occurring early during development have better chances of spreading than which appearing in adult tissues. Mutations influencing morphological characters are more easily detected than the nutritious, biochemical, regulatory, behavioural and lethal mutations. A single mutation affecting more than one character is called pleiotropic mutation. Mutations have helped in development of new varieties of plants and animals, e.g., Sharbati Sonora from Sonora-64.

- **Molecular Mechanism of Mutation :–**

  Gene mutation can occur by three methods - inversion, substitution and frame-shift. In substitution there is replacement of one nitrogen base with another. It is of two types, transition and transversion. In frame shift mutation, the sequence of nucleotides is changed, so that new codons develop. It is of two types, deletion and insertion.

  (1) **Inversion :–** Base sequence of a segment is reversed,

  (2) **Transition Substitution :–** One nitrogen base is replaced by another of similar type, *e.g.*, A G, C T. It is induced by **tautomerisation :–** (alternate state due to rearrangement of hydrogen atoms, $-NH_2$ into $—NH$ or $—CO$ into $-COH$), deamination, base analogue and ionisation.

  (3) **Transversion Substitution :–** One nitrogen base is replaced by another of different type.

  a) **Insertion Frameshift :–** Addition of a nucleotide changes the reading of codons in down stream. Insertion can bring about expansion of genes. Certain genes have the tendency to expand and cause mutations.

  (b) **Deletion Frameshit :–** Deletion of a nucleotide changes the reading of codons in down stream.

  (4) **Tautomerization :–** The purines and pyrimidines in DNA and RNA may exist in several alternate forms or tautomers. Tautomerization occurs through rearrangement of electrons and protons in the molecule. Tautomers show changed base pairing so as to cause change in sequence like AT to CG.

- **Same Sense, Mis-Sense and Non-sense Mutations :–**
  - (i) **Non-sense Mutation :–** Such mutations arise when a normal codon, coding for an amino acid is changed into a chain terminating codon (UAG, UAA, UGA) resulting in the production of an incomplete polypeptide.

    Nonsense mutations rarely go unnoticed because the incomplete or shorter protein formed, is generally inactive.
  - (ii) **Mis-sense mutation :–** Change in base in a codon, producing a different amino acid at the specific site in a polypeptide. In mis-sense mutation, the change in one amino acid is frequently compatible with some biological activity, e.g., sickle cell anaemia.
  - (iii) **Same-sense mutation:** Due to wobble position, a change in one nucleotide of a codon does not change amino acid specificity. This is called same sense (or silent) mutation, *e.g.*, AGA = AGG = AGT = AGC.

- **Spontaneous Mutations :–**

  They are mutations which develop at random, naturally, automatically or spontaneously in an organism due to internal reasons without any relation to any external/environmental factor.

  The frequency of spontaneous mutations is different for different organisms and their different genes-1 in one million in Drosophila, 1 in 10 million cell generations in bacteria, 1 in 50,000 in human beings.

  The genes which mutate frequently are called mutable genes e.g., R-gene of colour in Maize (1 in 2000 gametes). Stable genes do not mutate even once in several million gametes.

  Mutator gene increase frequency of spontaneous mutations while antimutator genes prevent spontaneous mutations. Spontaneous mutations may be due to

  (i) Errors in replication. (ii) Failure of proof-reading machinery. (iii) Presence of tautomeric forms of nitrogen bases, e.g., imino tautomer instead of amino group (e.g., cytosine, adenine) or enol group instead of keto group (e.g., thymine, guanine). (iv) Slow spontaneous deamination of cytosine to uracil. (v) Back ground radiations.

- **Induced Mutations :–**

They are produced by specific external factors or chemicals, called mutagen.

Muller (1927) was the first scientist to produce induced mutations in Drosophila with the help of X-rays (upto 150 times the spontaneous sex-linked lethal mutations). So, muller is considered as father of Actinobiology. He got Nobel prize in 1946.

15.2 Mutagens

Physical and chemical factor, which causes new mutations and increase frequency of spontaneous mutations.

(1) Physical Mutagens :– Physical mutagens are of three types-temperature, ultra-violet radiations and ionising radiations.

- **Temperature:** Increase in temperature increases frequency of mutations Q10 =5. Low temperature is mutagenic in rice.
- **UV Radiations (Muller & Altenberg) :–** They induce hydrolysis of cytosine and formation of thymine dimers.

Thymine dimers cause bending and misreplication of DNA.

- **Ionising Radiations :–** They include X-rays (man-made; Roentgen, 1895), cosmic rays, α-particle, β-particle, γ rays.

P-32 gets incorporated in DNA but starts decaying and changed to sulphur. DNA develops breaks in these regions.

Ionising radiations produce charged particles which ionise biomolecules and bring about several changes in DNA.

There is deamination & dehydration of N_2 , formation of peroxides & oxidation of deoxyribose.

(2) Chemical Mutagens :– They are chemicals, which induce mutations. Most of the mutagens are also carcinogens.

- **Nitrous Acid :–** It delaminates cytosine to uracil, guanine to xanthin and adenine to hypoxanthin.

This results in replacement of A-T with H-C, C-G with U-A and C-X.

- **Alkylating Agents :–** Causes methylation and ethylation of nitrogen bases, e.g., methyl guanine, ethyl guanine.

The alkylated base may slip out of DNA duplex or cause change of base pair, e.g. C-G to A-T.

Alkylating agents include ethyl ethane sulphonate (EES), ethyl methane sulphonate (EMS), diethyl sulphate (DES), dimethyl nitrosamine (DMN), nitrogen mustard, ethyl dibromid", griseofulvin, endrin, etc.

- **Base Analogues :–** 5-bromouracil (Bu), 5-iodouracil (Iu), 5-fluorouracil (Fu) replace thymine and pair with guanine.

2-aminopurine is replaces adenine with its tautomer pairing with cytosine. Enol state of thymine pairs with guanine,

- **Acridines :–** Proflavin, acriflavin, euflavin and acridine orange are intercalated in between base pairs leading to their insertion and deletion so as to cause frame-shift or gibberish mutations.

Main source of spontaneous mutations are natural radiations coming from cosmic rays of sun.

16. Multiple alleles in animals

Several examples of multiple alleles in animals are as follows

(i) Among animals, coat colour in rabbit is considered as the best example of multiple alleles. e.g.

(a) Fully coloured or agouti (wild type) = C
(b) Chinchila pattern (uniform light grey pattern) = C^{ch}
(c) Himalayan pattern (body white with extremities) = c^{h}
(d) Albino (pure white) = c

The gene C is dominant over all other alleles. Chinchila is recessive to full colour but dominant to others. Similarly, Himalayan pattern is dominant only over albino pattern and albino pattern is seen only in homozygous recessive state. Thus, different phenotypes and their possible genotypes are as follows

(a) Agouti : CC, CC^{ch}, Cc^{h}, Cc
(b) Chinchila : $c^{ch}C^{ch}$, $C^{ch}c^{ch}$, $C^{ch}c$
(c) Himalayan : $c^{h}c^{h}$, $c^{h}c$
(d) Albino : cc

(ii) Eye colour in Drosophila also appears as a good example. Here, normal eye colour is red and white, which shows simple dominant recessive relationship. Afterwards, different eye colour shades are also discovered, which are controlled by different allelomorphs occupying the same locus at X-chromosome. These are as follows

(a) Apricot colour : w^{a}
(b) Ivory colour : w^{I}
(c) Blood colour : w^{bl}
(d) Pearl colour : w^{p}
(e) Cherry colour : w^{ch}
(f) Tinged colour : w^{t}

With wild type as red = W and recessive = w, these all patterns show intermediate inheritance when brought together.

(iii) ABO blood group in humans also serves as very good example of multiple alleles. This is described in detail in the chapter human genetics.

17. Pedigree Analysis

A record of inheritance of certain genetic traits for two or more generations presented in the form of a diagram or family tree is called pedigree.

- Parents are shown by horizontal line while their offsprings are connected to it by a vertical line. The offsprings are also shown in the form of a horizontal line below the parents and numbered with arabic numerals.
- Pedigree analysis is study of pedigree for the transmission of particular trait and finding the possibility of absence or presence of that trait in homozygous or heterozygous state in a particular individual.
- It is useful for the genetic counselors to advise intending couples about the possibility of having children with genetic defects like haemophilia, colour blindness, alkaptonuria, phenylketonuria, thalassaemia, sickle cell anemia (recessive traits), brachydactyly, myotonic dystrophy and polydactyly (dominant traits).
- Pedigree analysis indicates that Mendel's principles are also applicable to human genetics with some modifications found out later, like quantitative inheritance, sex linked characters and other linkages.

Symbols used in Pedigree analysis :

Square – Male (Normal)

Circle – Female (Normal)

– Affected individuals

– Symbol with a dot – carrier for recessive trait

– Line between square and circle – mating / marriage line

– Heterozygotes for autosomal recessive

– Death

– Abortion / Still birth

– Propositus

– Proposita

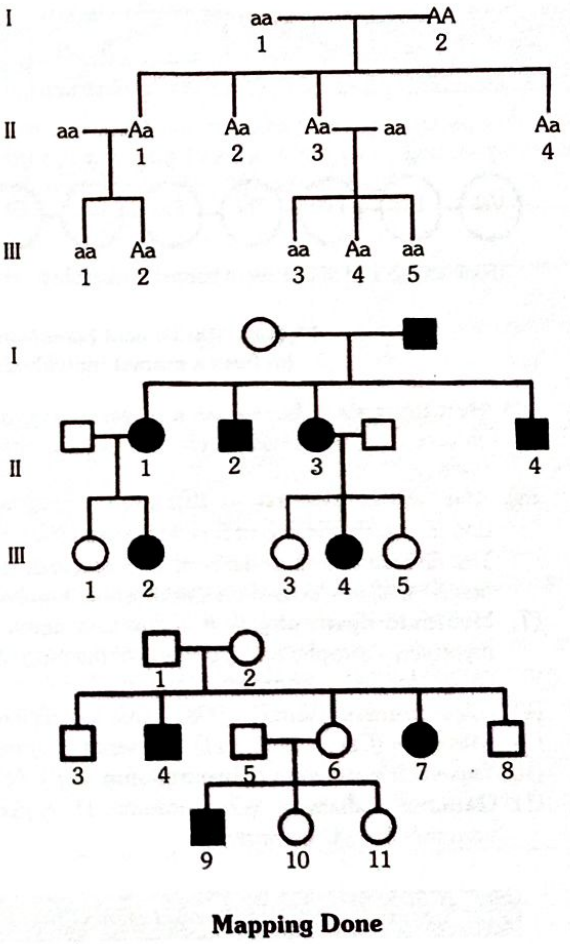
– Consanguineous marriage

– Monozygotic twin

– Dizygotic twin

– Sex unspecified

– Five unaffected offsprings



Mapping Done

Proband is person from which case history starts. If it is male, it is called propositus, if it is female it is called proposita.

All the members of II generation will, therefore, be heterozygous (Aa). This is further confirmed by marriage of II – 1 with homozygous recessive (Aa × aa = 2Aa, 2aa) and bearing children of both the parental types. Marriage of II – 3 with the homozygous recessive can produce both recessive and heterozygotes as are present here.

18. Genetic Disorders

18.1 Mendelian Disorders

(1) Sickle-Cell Anaemia :– It is autosomal recessive disorder. In this disorder, the RBCs become sickle shaped under low O_2 concentration. The affected persons die young. Other heterozygous for this trait are having normal phenotype and long lived.

The disease is due to base substitution of sixth codon in the gene coding for β chain of haemoglobin. The middle base of a DNA triplet coding for the amino acid glutamic acid is mutated so that the triplet now codes for valine instead.

The mutant haemoglobin molecule undergoes polymerization under low O_2 tension causing the change in the shape of RBC from biconcave disc to elongated sickle like structure.

(2) Thalassaemia :– It is recessive autosomal disease caused due to reduced synthesis of α or β polypeptide of haemoglobin.

β thalassaemia is a major problem, individuals suffering from major thalassaemia often die before ten years of age.

(3) **Phenylketonuria :–** Recessive autosomal disorder (Chromosome 12) related to phenylalanine metabolism to tyrosine. This disorder is due to absence of a liver enzyme called phenylalanine hydroxylase. Due to lack of this enzyme, phenylalanine follows another pathway and gets converted into phenyl pyruvic acid. The phenyl pyruvic acid upon accumulation in joints causes arthritis and if it hits the brain, then it causes mental retardation known as phenyl pyruvic idiocy. These are also excreted through urine because of poor absorption by kidney.

(4) **Cystic Fibrosis :–** It is an autosomal recessive disorder common among Caucasian Northern Europeans. Persons suffering from this disease are having extremely salty sweat. It is due to decreased Na^+ and Cl^- reabsorption in the ducts. Disease is due to a gene present on chromosome 7. Due to a defective glycoprotein, thick mucus develops in pancreas and lungs and formation of fibrous cyst occurs in pancreas.

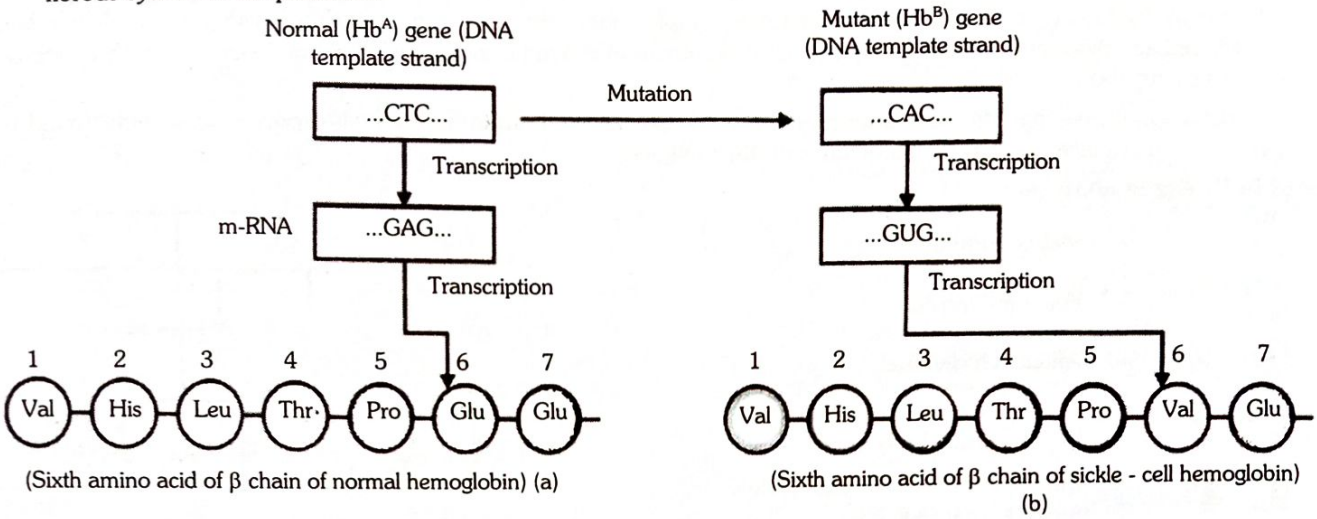


Figure :– Amino acid composition of the relevant portion of β-chain haemoglobin: (a) from a normal individual; (b) from an individual with sickle-cell anaemia.

(5) **Huntington's Chorea :–** It is an autosomal dominant disorder. The gene responsible for this disorder is present on chromosome 4. Disease is characterized by gradual degradation of brain tissue in the middle age and consequent shrinkage of brain.

(6) **Alzheimer's Disease :–** This autosomal recessive disease results in mental deterioration (loss of memory, confusion, anxiety) and ultimately the loss of functional capacities.
The disease is due to deposits of β-amyloid, a short protein in brain which results in degradation of neurons. Its involves two defective alleles located on chromosome number 19 and 21. This disease is common in Down syndrome.

(7) **Myotonic dystrophy :–** It is due to a dominant autosomal mutant gene located on the long arm of chromosome 19. Mild myotonia – atrophy and weakness of the musculature of the face and extremities, is most common.
Other Mendelian disorders:

(8) **Alkaptonuria** (Garrod, 1908) – Due to deficiency of oxidase enzyme

(9) **Albinism** (Chromosome 11) – Absence of tyrosinase

(10) **Tay-Sach's disease (Chromosome 15) –** Absence of hexosaminidase B.

(11) **Gaucher's disease** (Chromosome 1) – Due to the inhibition of glucocerebrosidase enzyme action which leads to accumulation of cerebroside.

Chart of Formula

1.	Formula of gametes	2^n	
2.	Number of zygotes/offsprings	$(\text{Gametes})^n$	n = Number of traits / hybrid (n)
3.	Number of phenotype	2^n	
4.	Number of genotype	3^n	
5.	Number of genotypes in multiple allelism	$\frac{n}{2}(n+1)$, n = Number of alleles	
6.	Number of phenotype for polygenes	$2n + 1$	n = Pair of polygenes
7.	Number of genotype for polygenes	3^n	
8.	Recombination frequency or cross over value	$\frac{\text{Number of recombinants}}{\text{Total number of offsprings}} \times 100$	
9.	Number of barr body	Number of X-chromosomes – 1	
10.	Number of Y spots	Number of Y-chromosomes	
11.	Frequency of dominant trait in population	$P^2 + 2pq$	
12.	Frequency of recessive trait in population	q^2	

18.2 Other abnormalities due to autosomal dominant gene mutation

(1) Polydactyly – Presence of extra fingers and toes
(2) Brachydactyly – Abnormal short fingers and toes

18.3 Abnormalities due to sex linked (X-linked) recessive gene mutation

(1) Haemophilia A – Due to lack of antihaemophilic-globulin
Haemophilia B – Due to lack plasma thromboplastin
(2) Red-green colour blindness – Daltonism
Protanopia – Red colour blindness
Tritanopia – Blue colour blindness
Deuteranopia – Green colour blindness
(3) Lesch Nyhan syndrome – Deterioration of muscles at an early stage
(4) Lesch Nyhan syndrome – Deterioration of nervous system
Due to HGPRT deficiency (Hypoxanthin guanine phosphoribosyl transferase)

18.4 Chromosomal Disorders

A.	Autosomal abnormalities	(Due to mutation in body chromosome)
(i)	Down's Syndrome	It occurs due to trisomy of 21st Chromosome. The affected individual is short statured with small round head, furrowed tongue and partially open mouth. Palm is broad with characteristic palm crease and mental retardation. Physical and psychomotor development is retarded.
(ii)	Edward's syndrome	Trisomy of 18th chromosome
(iii)	Patau's syndrome	Trisomy of 13th chromosome
(iv)	Cri du chat syndrome	Due to deletion in short arm of 5th chromosome.

B.	Allosomal or Sex Chromosomal Disorder	
(i)	Klinefelter's syndrome	It occurs due to the trisomy of X-chromosome in male, resulting into a karyotype of 47, (44 + XXY). Individuals have long legs, sparse body hair, small prostrate gland, small testes, reduced mental intelligence and enlarged breasts (Gynaecomastia). Such individuals are sterile.
(ii)	Turner's Syndrome	It is caused due to absence of one of the X- chromosomes in female i.e., 45 with chromosome
(iii)	Superfemale	AA + XXX, AA + XXXX
(iv)	Jacob's syndrome of Super male	AA + XYY, also called as criminal syndrome.

27. Principles of Inheritance and Variation – Multiple Choice Questions

1. Mendelism

1. Term '*genetics*' was given by
(a) Mendel (b) Morgen
(c) Bateson (d) Boveri

2. Mendel was the native of
(a) France (b) Sweden
(c) India (d) Austria

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

4. How many pairs of contrasting characters in pea pod were choosen by Mendel
(a) 2 (b) 3
(c) 4 (d) 7

5. Mendel was lucky, because
(a) He was born in Austria which is a nice country
(b) He used pea plant for his experiment which is rich in protein
(c) The genes for different characters are located on different chromosomes *in Pisum sativum*
(d) The *Pisum sativum* is short-lived plant

6. The alleles are
(a) A pair of genes governing a specific character such as tallness or dwarfness
(b) Multiple forms of genes
(c) Genes governing eye characters
(d) Genes present in allosomes

7. An organism with two identical alleles for a given trait is
(a) Homozygous (b) Segregating
(c) Dominant (d) A hermaphrodite

8. Test cross is used to
(a) Check heterozygosity in F_1 generation
(b) Check heterozygosity in F_2 generation
(c) Check independent assortment
(d) Check segregation

9. An exception to Mendel's law is
(a) Law of independent assortment
(b) Law of segregation
(c) Law of dominance
(d) Law of linkage

10. Mendel enunciated
(a) Two principles of inheritance
(b) Three principles of inheritance
(c) Four principles of inheritance
(d) Five principles of inheritance

11. A cross between plants having *RRYY* and *rryy* composition will yield plants with
(a) Round and yellow seeds
(b) Round and green seeds
(c) Wrinkled and yellow seeds
(d) Wrinkled and green seeds

12. 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
(a) 2, 2, 2 (b) 2, 2, 1
(c) 1, 2, 2 (d) 1, 1, 2

13. Some of the dominant traits studied by Mendel were
(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

14. The genotypes of offspring in a genetic cross is called graphical representation to calculate the probability of all possible
(a) Pedigree analysis (b) Karyotype
(c) Punnett square (d) Chromosome map
(e) Genotype ratio

15. The colour based contrasting traits in seven contrasting pairs, studied by Mendel in pea plant were
(a) 1 (b) 2
(c) 3 (d) 4

16. Which of the following depicts the Mendel's dihybrid ratio
(a) 3 : 1 (b) 9 : 3 : 3 : 1
(c) 9 : 7 (d) 15 : 1

17. If a plant heterozygous for tallness is selfed, the F_2 generation has both tall and dwarf plants. This proves the principle of
(a) Dominance (b) Segregation
(c) Independent assortment (d) Incomplete dominance

18. 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
(a) One (b) Three
(c) Four (d) Eight

19. 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
(a) TT × tt (b) Tt × tt
(c) TT × Tt (d) Tt × Tt

20. A cross between two tall plants resulted in offspring having few dwarf plants. What would be the genotypes of both the parents
(a) TT and Tt (b) Tt and Tt
(c) TT and TT (d) Tt and tt

21. In a dihybrid cross, if you get 9:3:3:1 ratio it denotes that
(a) The alleles of two genes are interacting with each other
(b) It is a multigenic inheritance
(c) It is a case of multiple allelism
(d) The alleles of two genes are segregation independently

22. Mendel's Law of independent assortment holds good for genes situated on the
(a) Non-homologous chromosomes
(b) Homologous chromosomes
(c) Extra nuclear genetic element
(d) Same chromosome

23. It is said that Mendel proposed that the factor controlling any character is discrete and independent. His proposition was based on the
(a) Results of F_3 generation of a cross
(b) Observations that the offspring of a cross made between the plants having two contrasting characters shows only one character without any blending
(c) Self pollination of F_1 offsprings
(d) Cross pollination of F_1 generation with recessive parent

24. Two genes 'A' and 'B' are linked. In a dihybrid cross the number of phenotypes and genotypes are
(a) 1 : 1 : 1 : 1 (b) 9 : 3 : 3 : 1
(c) 3 : 1 (d) 1 : 1

25. Recessive characters are expressed
(a) Only when they are present on X chromosomes of male
(b) Only when they are present on X chromosomes of female
(c) On any autosome
(d) On both the chromosomes of female

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

27. 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
(a) Rrr_0 (b) $RrRr_0$
(c) Rrr_0r_0 (d) rrR_0r_0

28. 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
(a) $\frac{1}{4}$ (b) $\frac{1}{8}$
(c) $\frac{1}{16}$ (d) $\frac{3}{16}$

29. 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
(a) 2 (b) 4
(c) 6 (d) 8

30. 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
(a) Black eye (b) Brown eye
(c) Green eye (d) Blue eye

31. 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
(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

32. 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
(a) TTRR (b) TtRR
(c) TtRr (d) TTRr

33. 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

	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

34. In the F_2 generation of a Mendelian dihybrid cross the number of phenotypes and genotypes are
(a) Phenotypes – 4; genotypes - 16
(b) Phenotypes – 9; genotypes - 4
(c) Phenotypes – 4; genotypes - 8
(d) Phenotypes – 4; genotypes - 9

35. A tobacco plant which is heterozygous for albinism (a recessive character) is self pollinated if 1200 seeds are subsequently germinated, how many of the seedlings would have the parental genotype
(a) 300 (b) 600
(c) 900 (d) 1200

2. Interaction of Gene and Cytoplasmic Inheritance

1. 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
(a) Complementary genes
(b) Epistasis
(c) Codominance
(d) Incomplete dominance

2. The gene interaction when one gene masks the effect

Or

When a gene pair hides the effect of another, the phenomenon is called
(a) Complementary gene action
(b) Supplementary gene action
(c) Duplicate gene action
(d) Epistasis

3. Complete dominance is absent in
(a) Pisum sativum (b) Mirabilis jalapa
(c) Lathyrus odoratus (d) Oenothera lamarckiana

4. The F_2 generation offspring in a plant showing incomplete dominance, exhibit
(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

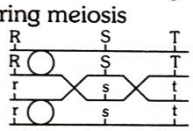
5. The phenotypic ratio obtained in quantitative inheritance of a dihybrid cross is

Or

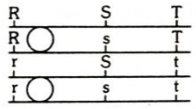
In a cross between red kernelled and white kernelled varieties of wheat showing polygenic inheritance the phenotypic ratio in F_2 generation will be
(a) 1 : 2 : 1
(b) 1 : 4 : 6 : 4 : 1
(c) 1 : 6 : 15 20 : 15 : 6 : 1
(d) 9 : 3 : 3 : 1

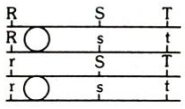
3. Linkage and Crossing Over

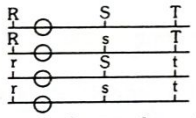
1. Coupling and repulsion are the two faces of
   (a) Crossing over (b) Linkage
   (c) Chiasmata (d) Mutation
2. Crossing–over occurs in the
   (a) Leptotene stage (b) Pachytene stage
   (c) Anaphase stage (d) Diakinesis stage
3. Linkage was first observed in
   (a) Field pea (b) Sweet pea
   (c) Pea (d) Grass pea
4. Chiasma shows the sites of
   (a) Spindle formation (b) Synapsis
   (c) Crossing over (d) None of these
5. Distance between the genes and percentage of recombination shows
   (a) A direct relationship (b) An inverse relationship
   (c) A parallel relationship (d) No relationship
6. Depending upon the distance between any two genes which is inversely proportional to the strength of linkage, cross overs will vary from
   (a) 50–100% (b) 0–50%
   (c) 75–100% (d) 100–150%
7. For the preparation of genetic maps, the recombination frequencies between genes are additive over short distances but not over long distances due to
   (a) Multiple cross overs (b) Lethal mutation
   (c) Epistasis (d) Synaptonemal complex
8. The figure shows a homologous (bivalent) pair of chromosomes during meiosis

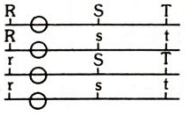


Which one of the following option correctly illustrates the final products of the second meiotic division

(a) 

(b) 

(c) 

(d) 

9. All genes located on the same chromosome
   (a) Form different groups depending upon their relative distance
   (b) Form one linkage group
   (c) Will not form any linkage groups
   (d) Form interactive groups that affect the phenotype

4. Chromosomes and Genes

1. The terminal end of a chromosome is called
   (a) Centromere (b) Chromomere
   (c) Telomere (d) Metamere
2. The name chromatin was coined by
   (a) Flemming (b) Robert Brown
   (c) George Palade (d) Camillo Golgi
   (e) Rudolf Virchow
3. Who used the word "*chromosome*"
   (a) Huxley (b) Flemming 1888
   (c) Kollikar 1888 (d) Waldeyer 1888
4. Gene can be defined as
   (a) Unit of segregation
   (b) Unit of physiological activity
   (c) Unit of recombination
   (d) Unit of function
5. Chromosomal theory of inheritance was based on
   (a) Segregation of genes
   (b) Diploidy and haploidy
   (c) Sex linkage
   (d) Presence of sex chromosomes
6. A male child would be born to parents if
   (a) Father is healthier than mother
   (b) Genetic chromosomal constitution of child is XX
   (c) Mother feeds well during pregnancy
   (d) Genetic chromosomal constitution of child is XY
7. What is the chromosome number of plasmodium
   (a) 18 (b) 14
   (c) 10 (d) 9
8. A normal metaphase chromosome with a middle centromere is

   Or

   Chromosomes whose arms are equal are called
   (a) Metacentric (b) Sub-metacentric
   (c) Acrocentric (d) Telocentric
9. In humans, most number of genes are located on chromosome
   (a) 1 (b) 6
   (c) X (d) 21
10. In sex linkage, the speciality is
    (a) Atavism (b) Criss-cross inheritance
    (c) Reversion (d) Gene flow
11. In man, sperms contain autosomes and
    (a) Only Y chromosome
    (b) Only X chromosome
    (c) Both X and Y chromosomes
    (d) Either X or Y chromosomes
12. In humans chromosomal condition of male is
    (a) 44 AA + XO (b) 44 AA + XX
    (c) 44 AA + XY (d) 44 AA + XXY
13. Whereas the number of chromosomes is reduced to half in first reduction division of meiosis, then what is the need for second mitotic division
    (a) For the segregation of replicated chromosomes
    (b) For equal distribution of haploid chromosomes
    (c) For the formation of four gametes
    (d) For equal distribution of genes on chromosomes
14. The function of chromosomes of carrying the genetic information from one cell generation to another is performed by
    (a) RNA (b) DNA
    (c) Histones (d) Calcium
15. In man sexlinked characters are mainly transmitted through
    (a) Y-chromosome
    (b) Autosomes
    (c) X-chromosome
    (d) X-chromosome, Y-chromosome and Autosomes
16. In order to calculate map distance of genes on a chromosome, one must know the
    (a) Number of mutant genes
    (b) Cross over percentage
    (c) Recombination frequency of each gene locus
    (d) Non-cross over percentage
17. Genes carried on chromosomes was first proved by
    (a) Mendel (b) Watson
    (c) Crick (d) Bridges
18. The core of nucleosome is made up of
    (a) H_1, H_2A, H_2B, H_3 (b) H_1, H_2A, H_2B, H_4
    (c) $H_1, H_2A, H_2B, H_3, H_4$ (d) H_2A, H_2B, H_3, H_4
19. Structural element of chromatin is
    (a) Histone (b) Acid protein and DNA
    (c) Nuclear matrix (d) Nucleosome

20. Nucleosomes are
(a) Units of DNA (b) Units of RNA
(c) Units of proteins (d) Units of chromosomes

21. What are allosomes
(a) Granular structures on chromosomes
(b) Node like structures on chromosomes
(c) Sex chromosomes
(d) None of the above

22. One functional unit of gene which specifies synthesis of one polypeptide is known as
(a) Recon (b) Clone
(c) Codon (d) Cistron

23. The terms *cistron*, *recon* and *muton* were proposed by
(a) W. Ingram (b) Bateson
(c) J. Lederberg (d) S. Benzer

24. The theory of jumping genes was propounded by or Noble prize for the concept of jumping gene was given to
(a) Mendel (b) Morgan
(c) Barbara Mc Clintock (d) Sanger

25. Genetically active area of chromosome is called
(a) Euchromatin (b) Heterochroatin
(c) Heptan (d) Cistron

26. Nucleosomes are bounded by
(a) RNA (b) Histone H_4
(c) Histone H_3 (d) DNA

27. Which one of the following true
(a) One gene one protein
(b) One gene one polypeptide
(c) One gene many polypeptide
(d) All of the above

28. In sickle cell anaemia glutamic acid is replaced by valine. Which one of the following triplets codes for valine
(a) G G G (b) A A G
(c) G A A (d) G U G

29. Which of the following will not result in variations among siblings
(a) Independent assortment of genes
(b) Crossing over
(c) Linkage
(d) Mutation

30. In a certain taxon of insects some have 17 chromosomes and the other have 18 chromosomes. The 17 and 18 chromosome-bearing organisms are
(a) Males, and females, respectively
(b) Females and males, respectively
(c) All males
(d) All females

31. The distance between two genes in a chromosome is measured in cross-over units which represent
(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

32. In polytene chromosomes dark bands are visible. These bands are formed by the apposition of
(a) Protein particles
(b) Chromomeres on chromonemata
(c) Nucleosomes
(d) None

33. Polytene or giant chromosomes are found in
(a) Salivary glands of man
(b) Salivary glands of woman
(c) Salivary glands of all animals
(d) Salivary glands of *Drosophila*

34. *Drosophila melanogastor* has 8 chromosomes in somatic cell. How many linkage groups will be there
(a) 4 (b) 8
(c) 2 (d) 5

35. Balbiani rings are present in
(a) Polysomes (b) Autosomes
(c) Polytene chromosomes (d) None of the above

36. In *Pisum sativum* there are 14 chromosomes. How many pairs with different chromosomal composition can be prepared
(a) 14 (b) 7
(c) 2^{14} (d) 2^7

37. Experimental verification of the chromosomal theory of inheritance was given by
(a) Gregor Johann Mendel (b) Hugo de Vries
(c) Langdon Down (d) Henking
(e) Thomas Hunt Morgan

38. 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
(a) V, W, T, U (b) T, V, W, U
(c) T, W, U, V (d) V, T, W, U

5. Multiple Allelism

1. In humans, height shows a lot of variation. It is an example of
(a) Multiple alleles
(b) Pleiotropic inheritance
(c) Polygenic / Quantitative inheritance
(d) Pseudoalleles

2. The offspring produced from a marriage have only O or A blood groups. Of the genotypes given below, the possible genotypes of the parents would be
(a) I^AI^A and I^AI^O (b) I^OI^O and I^OI^O
(c) I^AI^A and I^OI^O (d) I^AI^O and I^OI^O

3. A person with blood group 'A' can be given blood of which group
(a) *A* and *B* (b) *B* and *O*
(c) *A* and *O* (d) *A*, *B*, *AB* and *O*

4. A person having blood group *O* can receive blood of
(a) Group *O*, *B* and *AB* (b) Group *A*, *B* and *AB*
(c) Group *B* and *AB* (d) Group 'O' only

5. When red blood corpuscles containing both A and B antigens are mixed with your blood serum, they agglutinate. Hence your blood group is ..... type
(a) AB (b) O
(c) A (d) B

6. Universal donors have no antigens in RBC and have both *a* and *b* antibodies. They belong to blood group
(a) *A* (b) *B*
(c) *AB* (d) *O*

7. Persons of blood group *A* contain
(a) Antigen *A* and antibodies *b*
(b) Antigen *A* and antibodies *a*
(c) Antigen *A* and *B* and no antibodies
(d) No antigens and both *a* and *b* antibodies

8. Blood group agglutinogen is
(a) Glycoprotein (b) Phosphoprotein
(c) Haemoprotein (d) Phospholipid

9. Donors and recipients in a blood transfusion process can be
(a) Only father and son
(b) Only brother and sister
(c) Only maternal uncle and niece
(d) All the above

10. Rh factor is present in
(a) All vertebrates
(b) All mammals
(c) All reptiles
(d) Man and rhesus monkey only

11. Which blood group can be given to patients of any blood group
(a) O (b) A
(c) B (d) AB

12. Which one of the following is hereditary character of blood
(a) Blood group (b) Haem
(c) Nucleus (d) None of the above

13. Person having genotype I^A I^B would show the blood group as AB. This is because of
(a) Pleiotropy (b) Co-dominance
(c) Segregation (d) Incomplete dominance

14. A woman with blood group 'O' has a child with blood group 'O'. She claims that a man with blood group 'A' as the father of her child. What would be the genotype of the father, if her claim is right
(a) I^OI^O (b) I^AI^B
(c) I^AI^O (d) I^BI^O

15. Mother and father of a person with 'O' blood group have 'A' and 'B' blood group, respectively. What would be the genotype of both mother and father
(a) Mother is homozygous for 'A' blood group and father is heterozygous for 'B'
(b) Mother is heterozygous for 'A' blood group and father is homozygous for 'B'
(c) Both mother and father are heterozygous for 'A' and 'B' blood group, respectively
(d) Both mother and father are homozygous for 'A' and 'B' blood group, respectively

16. A man with blood group 'AB' marries a woman with 'O' blood group. In this situation
(a) The blood groups of their children will be the same as that of the mother
(b) The blood group of the children differs from both the parents
(c) While 50% of children will have father's blood group, the remaining will have mother's blood group
(d) None of the above

17. Which of the following statements is the most appropriate for sickle cell anaemia
(a) It cannot be treated with iron supplements
(b) It is a molecular disease
(c) It confers resistance to acquiring malaria
(d) All of the above

6. Genetic Variation

1. Number of sex chromosomes is normal in
(a) Super female (b) Turner's syndrome
(c) Klinefelter's syndrome (d) Down's syndrome

2. Mating between two individuals differing in genotype to produce genetic variation is called
(a) Domestication (b) Introduction
(c) Hybridisation (d) Mutation

3. Occurrence of cell containing multiples of 2n genomes in diploid organisms is known as
(a) Aneuploidy (b) Alloploidy
(c) Amphiploidy (d) Endopolyploidy

4. The hereditary disease in which the urine of a person turns black on exposure to air due to the presence of homogentisic acid is known as
(a) Ketonuria (b) Phenylketonuria
(c) Haematuria (d) Alkaptonuria

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

6. Mutation is
(a) Sudden change in morphology
(b) Change in characters
(c) Change in heritable characters
(d) None of these

7. Genomic mutation is
(a) Change in number genes
(b) Change in number of chromosomes
(c) Change in shape of chromosomes
(d) All of these

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

9. The functional unit of mutation is
(a) Gene (b) Muton
(c) Recon (d) Cistron

10. Edward syndrome is on account of .....
(a) 45 chromosomes instead of 46
(b) Presence of three chromosomes on 18th pair of autosome
(c) Presence of three chromosomes on 21st pair of autosome
(d) Presence of three pair of sex chromosomes

11. The gene for diabetes mellitus is
(a) Autosomal dominant (b) Autosomal recessive
(c) Sex-linked dominant (d) Sex linked recessive

12. Mutation caused by a mutagen is
(a) Natural (b) Chemical
(c) Spontaneous (d) Induced

13. UV radiations cause
(a) Formation of thymine dimers
(b) Deletion of base pairs
(c) Methylation of bases
(d) Addition of base pairs

14. The number of chromosomes in Klinefelter's syndrome is
(a) 47 (44 + XXY)
(b) 47 (44 + XXX)
(c) 47 (46 + 1 chromosome 21)
(d) None of these

15. Which of the following chromosomal constitution refers to Jacob's syndrome in human
(a) $44+XO$ (b) $44+XXY$
(c) $44+XYY$ (d) $45+XYY$

16. Addition or deletion of a single nucleotide results in which type of mutation
(a) Deficiency (b) Duplication
(c) Frameshift mutation (d) None of these

17. In man, which of the following genotypes and phenotypes may be the correct result of aneuploidy in sex chromosomes
(a) 22 pairs + XXY males
(b) 22 pairs + XX females
(c) 22 pairs + XXXY females
(d) 22 pairs + Y females

18. Addition of one or more haploid set of its own genome in an organism results in
(a) Autopolyploidy (b) Allopolyploidy
(c) Aneuploidy (d) Diploid

19. Which is correct for Turner's syndrome
(a) It is a case of monosomy
(b) It causes sterility in females
(c) Absence of Barr body
(d) All of the above

20. Thalassemia and sickle cell anemia are caused due to a problem in globin molecule synthesis. Select the correct statement
(a) Both are due to a qualitative defect in globin chain synthesis
(b) Both are due to a quantitative defect in globin chain synthesis
(c) Thalassemia is due to less synthesis of globin molecules
(d) Sickle cell anemia is due to a quantitative problem of globin molecules

21. Conditions of a karyotype $2n+1, 2n-1$ and $2n+2, 2n-2$ are called
(a) Aneuploidy (b) Polyploidy
(c) Allopolyploidy (d) Monosomy

22. Occasionally, a single gene may express more than one effect. The phenomenon is called
(a) Multiple allelism (b) Mosaicism
(c) Pleitropy (d) Polygeny

23. The inheritance pattern of a gene over generations among humans is studied by the pedigree analysis. Character studied in the pedigree analysis is equivalent to
(a) Quantitative trait (b) Mendelian trait
(c) Polygenic trait (d) Maternal trait

24. Transition type of gene mutation is caused when
(a) GC is replaced by TA (b) CG is replaced by GC
(c) AT is replaced by CG (d) AT is replaced by GC

25. Chromosome complement with 2n-1 is called as
(a) Monosomy (b) Nullosomy
(c) Trisomy (d) Tetrasomy

26. The point mutations A to G, C to T, C to G and T to A in DNA are
(a) Transition, transition, transversion and transversion respectively
(b) Transition, transversion, transition and transversion respectively
(c) Transversion, transversion, transition and transition respectively
(d) All four are transition

27. A man homozygous for brown colour marries a lady heterozygous for brown colour is dominant. What will be the fate of their children
(a) All brown (b) Three brown
(c) Two brown (d) None of the above

28. Match the following

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

The correct match is

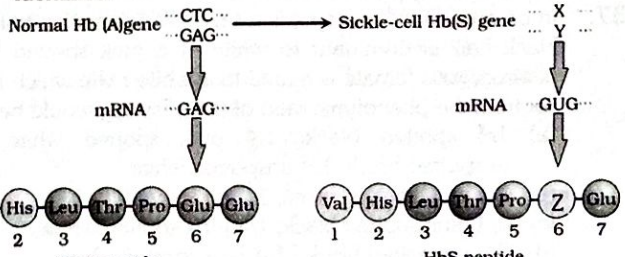
	A	B	C	D
(a)	I	IV	III	II
(b)	III	IV	II	I
(c)	IV	I	II	III
(d)	I	IV	II	III

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

30. Normally DNA molecule has A-T, G-C pairing. However, these bases can exist in alternative valency status, owing to rearrangements called
(a) Point mutation
(b) Analogue substitution
(c) Frame-shift mutation
(d) Tautomerisational mutation

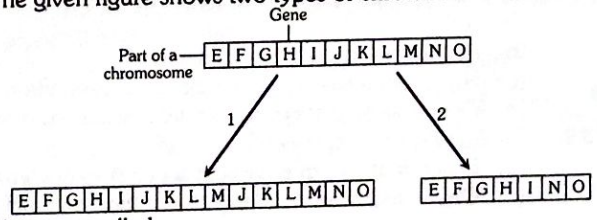
31. Which of the following corresponds to mutagens
(a) Chemicals and radiations which cause changes in the genetic material of a cell
(b) Various archaebacteria that produce methane
(c) Chemicals which react with ozone molecules and destroy them
(d) RNA molecules that infect plant cells and cause diseases

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



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

33. The given figure shows two types of chromosome mutation



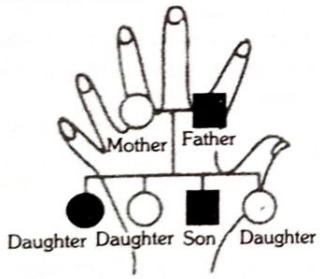
These are called
(a) 1 - Inversion, 2 - Substitution
(b) 1 - Inversion, 2 - Deletion
(c) 1 - Duplication, 2 - Substitution
(d) 1 - Duplication, 2 – Deletion

34. Sickle cell anaemia is caused by the substitution of
(a) Valine by glutamic acid at sixth position of alpha chain of haemoglobin
(b) Valine by glutamic acid at sixth position of beta chain of haemoglobin
(c) Glutamic acid by valine at sixth position of alpha chain of haemoglobin
(d) Glutamic acid by valine at sixth position of beta chain of haemoglobin
(e) Glutamic acid by methionine at sixth position of alpha chain of haemoglobin

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

(a) 22 + XX, 22 + YY, 22, 22
(b) 22 + X, 22 + Y, 22 + XY, 22
(c) 22 + XY, 22 + XY, 22, 22
(d) 22 + X, 22 + X, 22 + Y, 22 + Y

36. In the given human hand pedigree which character is represented and what is the probability of disease occurrence in fifth child



(a) Polydactyly (X- linked recessive disorder), 50%
(b) Polydactyly (X- linked dominant disorder), 50%
(c) Polydactyly(autosomal recessive disorder),50%
(d) Polydactyly (autosomal dominant disorder),50%

37. In an organism, pink spot is a sex-linked recessive trait and black hair is dominant to white. If a pink spotted black heterozygous female is mated to a white male which is not spotted, the phenotypic ratio of the offspring would be
(a) 1/4 spotted black; 1/4 pink spotted white; 1/4 unspotted black; 1/4 unspotted white
(b) 1/2 pink spotted black; 1/2 pink spotted white
(c) 3/4 pink spotted black; 1/4 pink spotted white
(d) 3/4 unspotted black ; 1/4 pink spotted white

38. When two genes are situated very close to each other in a chromosome
(a) The percentage of crossing over between them is very high
(b) Hardly any cross over are detected
(c) No crossing over can take place between them
(d) Only double cross overs can take place between them

39. Euploidy is best explained by
(a) Exact multiples of a haploid set of chromosomes
(b) One chromosome less than the haploid set of chromosomes
(c) One chromosome more than the haploid set of chromosomes
(d) One chromosome more than the diploid set of chromosomes

40. Match list I with List II and select the correct answer using code given below

List I (syndrome)
(1) Patau's syndrome (2) Kline-Felter's syndrome
(3) Down's syndrome (4) Turner's syndrome

List II (Chromosomal abnormality)
(A) 44 + XXY = 47
(B) 44 + X = 45
(C) 46 + 1 = 47, Chromosome 13th
(D) 46 + 1 = 47, Chromosome 21st

Code

	1	2	3	4
(a)	A	B	C	D
(b)	D	C	B	A
(c)	C	B	D	A
(d)	C	A	D	B

41. A mutation which substitutes one purine base with another purine base is called
(a) Transversion (b) Transduction
(c) Transition (d) Transfection

7. Sex Determination

1. Sex chromosomes for the first time was discovered in which plant
(a) Sphaerocarpus (b) Pisum sativum
(c) Neurospora (d) Lathyrus odoratus

2. When released from ovary human egg contain
(a) One *Y* chromosome (b) Two *X* chromosome
(c) One *X* chromosome (d) *XY* chromosome

3. The chromosomes responsible for the determination of sex are called
(a) Autosomes (b) Allosomes
(c) Multiple alleles (d) Heterosis

4. The first plant in which chromosomal basis of sex determination was discovered is
(a) *Melandrium* (Lychnis) (b) *Rumex*
(c) *Sphaerocarpus* (d) *Coccinia*

5. A family has five girls and no son. Probability of son as the 6th child will be
(a) 50% (b) 75%
(c) Full (d) No chance

6. If somatic cells of a human male contain single barrbody, the genetic composition of the person would be

Or

The genotype of a boy having sexual characters of a girl is
(a) XYY (b) XXY
(c) XO (d) XXXY

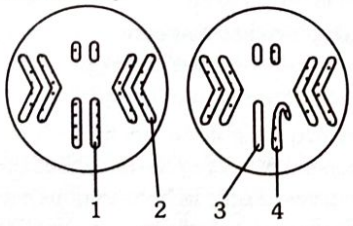
7. Which type of gene regulate sex-determination in Spinach plant
(a) Homozygous genes (b) Heterozygous genes
(c) Single gene (d) Multiple genes

8. ZZ / ZW type of sex determination is see in
(a) Platypus (b) Snails
(c) Cockroach (d) Peacock

9. Barr bodies and drumsticks are of what significance to genetists and biologists
(a) They indicate the presence of abnormal sex cells
(b) They indicate the presence of more than one X chromosome in the cells
(c) They indicate male calls
(d) They signify the presence of sex linked traits

10. Based on Lyon's hypothesis, what will be the number of Barr bodies found in a human female suffering from Down's syndrome
(a) 0 (b) 1
(c) 2 (d) 3

11. The following figure refer to the chromosome complement of each sex of fruit fly



By which number is a Y chromosome labelled
(a) 4 (b) 3
(c) 2 (d) 1

8. Sex Linked Inheritance

1. Which one of the following is a genetically transmitted character
(a) Colour blindness (b) Hydrocephalus
(c) Hemophilia (d) Muscular dystrophy
(e) All of these

2. The most common type of haemophilia results from the congenital absence of
(a) Factor II (b) Factor V
(c) Factor VIII (d) Factor XI

3. Which of the following diseases belongs to the same category as colourblindness in man
(a) Nightblindness (b) Presbyopia
(c) Diabetes incipidus (d) Haemophilia

4. Sex linked disease is
(a) Haemophilia (b) Colourblindness
(c) Sickle-cell anaemia (d) Both (a) and (b)

5. Sex influenced characters are due to
(a) Y-linked genes
(b) X-linked genes
(c) Autosomal genes
(d) Y-linked gene modification

6. A colour blind son will born when
(a) Mother is normal and father normal
(b) Mother is colour blind and father normal
(c) Mother is normal and father is colour blind
(d) All the cases are correct

7. The frequency of a character is found to be increasing when
(a) It is dominant (b) It is recessive
(c) It is adaptable (d) It is inheritable

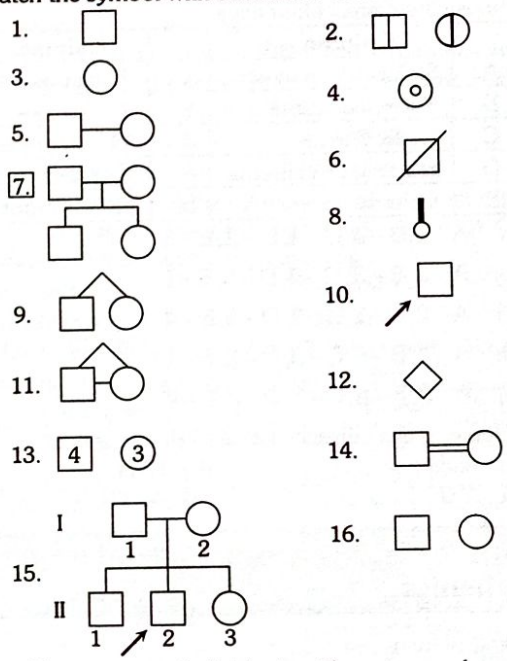
8. A marriage between normal visioned man and colourblind woman will produce which of the following types of offsprings
(a) Normal sons and carrier daughters
(b) Colourblind sons and carrier daughters
(c) Colourblind sons and 50% carrier daughters
(d) 50% colourblind sons and 50% carrier daughters

9. If a character is always transmitted directly from a father to all his sons and from their sons to all their sons, then which chromosome carries the gene for the character
(a) Autosomes (b) X chromosome
(c) Y chromosome (d) None of the above

10. If a genetic disease is transferred from a phenotypically normal but carrier female to only some of the male progeny, the disease is
(a) Autosomal dominant (b) Autosomal recessive
(c) Sex-linked dominant (d) Sex-linked recessive

11. More men suffer from colour blindness than women because
(a) Women are more resistant to disease than men
(b) The male sex hormone testosterone causes the disease
(c) The colour blind gene is carried on the 'Y' chromosome
(d) Men are hemizygous and one defective gene is enough to make them colour blind

12. Match the symbol with associated statement



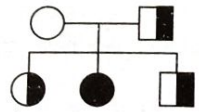
A. Heterozygous individuals with autosomal recessive
B. Diseased (or death)
C. Female carrier of an X - linked recessive gene
D. Individuals with normal trait
E. Consanguineous mating (marriage of blood relatives)
F. Unknown sex
G. Mating
H. Male
I. Female
J. Affected individual
K. Abortion or still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

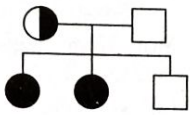
	B	C	E	F
(a)	6	14	14	12
(b)	16	13	2	11
(c)	3	1	2	7
(d)	16	1	2	7

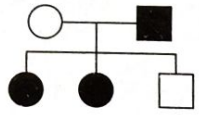
13. The following is a pedigree chart of a family with five children. It shows the inheritance of attached, ear - lobes as opposed to the free ones. The squares represent the male and circles the female individuals

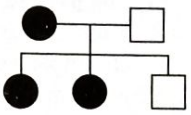
Which one of the following conclusions drawn is correct
(a) The parents are homozygous dominant
(b) The parents are homozygous recessive
(c) The parents are heterozygous
(d) The trait is Y – linked

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

(a) 

(b) 

(c) 

(d) 

15. Match the items in Column – I with Column – II and choose the correct alternative

	Column – I		Column – II
A.	Sickle-cell anaemia	1.	7^{th} chromosome
B.	Phenylketonuria	2.	4^{th} chromosome
C.	Cystic fibrosis	3.	11^{th} chromosome
D.	Huntington's disease	4.	X-chromosome
E.	Colur blindness	5.	12^{th} chromosome

(a) A–1, B–3, C–4, D–2, E–5

(b) A–2, B–3, C–4, D–5, E–1

(c) A–2, B–1, C–3, D–5, E–4

(d) A–4, B–5, C–3, D–2, E–1

(e) A–3, B–5, C–1, D–2, E–4

16. 'Haemophilia' disease is caused due to lack of

(a) ADH (b) STH

(c) AHF (d) ACTH

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

1. Identical twins are

(a) Heterozygous (b) Homozygous

(c) Monozygotic (d) Dizygotic

2. Study of human race is called

(a) Eugenics (b) Entomology

(c) Ecology (d) Pathology

3. Genetically identical progeny is produced when an individual

(a) Practices self–fertilization

(b) Produces identical gametes

(c) Practices reproduction

(d) Practices in breeding without meiosis

4. Two offspring developed in the same uterus but from fertilization of two different ova are

(a) Dizygotic twins (b) Monozygotic twin

(c) Fraternal twins (d) Both (a) and (c)

10. NEET

1. The first great "geneticist" was **[1991]**

Or

Who is considered as father of genetics

(a) Engler (b) Mendel

(c) Schwann (d) Miller

2. Which one of the following cannot be explained on the basis of Mendel's Law of Dominance **[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

3. Test cross in plants or in *Drosophila* involves crossing **[2006, 2010, 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

4. An allele is said to be dominant if **[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

5. When a gene exists in more than one form, the different forms are called **[1994, 2002, 2015]**

(a) Heterozygous (b) Complementary genes

(c) Genotypes (d) Alleles

6. A cross in which an organism showing a dominant phenotype is crossed with the recessive parent in order to know its genotype is called **[1995, 1995, 2010, 2010, 12]**

(a) Monohybrid cross (b) Back cross

(c) Test cross (d) Dihybrid cross

7. The cross used to ascertain whether the plant is homozygous or heterozygous is **[1994, 1994, 2002, 2001, 2006, 2008]**

Or

A cross between a homozygous recessive and a heterozygous plant is called **[1995, 2003]**

(a) Linkage cross (b) Reciprocal cross

(c) Test cross (d) Monohybrid cross

8. Mendel's law of heredity can be explained with the help of **[1999]**

(a) Mitosis (b) Meiosis

(c) Both mitosis and meiosis (d) None of the above

9. 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 **[2012]**

(a) Co–dominance

(b) Dihybrid cross

(c) Monohybrid cross with complete dominance

(d) Monohybrid cross with incomplete dominance

10. Which genotype represents a true dihybrid condition **[1991]**

(a) *Tt Rr* (b) *tt rr*

(c) *Tt rr* (d) *Tt RR*

11. 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 **[2000, 2002, 2003]**

(a) 1 : 2 : 1 (b) 9 : 3 : 3 : 1

(c) 3 : 1 (d) 1 : 3

12. 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*. **[2004, 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

13. When two genetic loci produce identical phenotypes in *cis* as well as in *trans* position, they are considered to be **[1995, 1999]**
(a) Pseudo alleles
(b) The parts of the same gene
(c) Multiple alleles
(d) Different genes

14. A collection of plants and seeds having diverse alleles of all the genes of a crop is called **[2011]**
(a) Genome (b) Herbarium
(c) Germplasm (d) Gene library

15. In his classic experiments on pea plants, Mendel did not use **[2015]**
(a) Pod length (b) Seed shape
(c) Flower position (d) Seed colour

16. 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 **[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

17. Among the following charactes, which one was not considered by Mendel in his experiments on pea? **[2017]**
(a) Stem – Tall of Dwarf
(b) Trichomes – Glandular or non-glandular
(c) Seed – Green or Yellow
(d) Pod – Inflated or Constricted

18. The segregation of paired hereditary factors that Mendel postulated occurs during **[1993]**
(a) Anaphase of first meiotic division
(b) Metaphase of second meiotic division
(c) During interphase between two meiotic divisions
(d) Prophase of first meiotic division

19. 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 **[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

20. 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 **[2007]**
(a) 50 : 50 (b) 9 : 1
(c) 1 : 3 (d) 3 : 1

21. From a cross *Aa BB* × *aa BB*, following genotypic ratio will be obtained in F_1 generation **[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*

22. A self-fertilizing trihybrid plant forms **[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

23. 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 **[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

24. 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 **[2014, 2014]**
(a) 0.6 (b) 0.7
(c) 0.4 (d) 0.5

25. A normal woman whose father was albino marries a man who is albino. What proportion of normal and albino can be expected among their off springs **[1994]**
(a) 1 normal : 1 albino (b) All albino
(c) 2 normal : 1 albino (d) All normal

26. In order to find out the different types of gametes produced by a pea plant having the genotype AaBb, it should be crossed to a plant with the genotype **[2005]**
(a) aaBB (b) AaBb
(c) AABB (d) aabb

27. Match the terms in column-I with their description in column-II and choose the correct option

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

[2016]

	(A)	(B)	(C)	(D)
(a)	(ii)	(i)	(iv)	(iii)
(b)	(ii)	(iii)	(iv)	(i)
(c)	(iv)	(i)	(ii)	(iii)
(d)	(iv)	(iii)	(i)	(ii)

28. In which mode of inheritance do you expect more maternal influence among the off spring **[2006]**
(a) Y-linked (b) X-linked
(c) Autosomal (d) Cytoplasmic

29. The most likely reason for the development of resistance against pesticides in insects damaging a crop is **[2004]**
(a) Directed mutation
(b) Acquired heritable changes
(c) Random mutations
(d) Genetic recombination

30. Genes for cytoplasmic male sterility in plants are generally located in **[2005]**
(a) Mitochondrial genome (b) Cytosol
(c) Chloroplast genome (d) Nuclear genome

31. Phenotype of an organism is the result of **[2006]**
(a) Environmental changes and sexual dimorphism
(b) Genotype and environment interactions
(c) Mutations and linkages
(d) Cytoplasmic effects and nutrition

32. When both alleles express their effect on being present together, the phenomenon is called **[2009, 2015]**

Or

Which Mendelism idea is depicted by a cross in which the F_1 generation resembles both the parents **[2013]**

(a) Dominance (b) Codominance
(c) Pseudodominance (d) Amphidominance

33. 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 **[2007]**

(a) AaBb (b) AaBB
(c) AABb (d) AABB

34. 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 **[2010]**

(a) *rrrr* (b) *RR*
(c) Rr (d) rr

35. When an albino female plant of maize is crossed with normal green male plant, all plants in the progeny are albino because **[1989, 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

36. 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 **[1997, 2000]**

(a) Nucleus (b) Chloroplast
(c) Mitochondria (d) Cytoplasm

37. Fruit colour in squash is an example of **[2014]**

(a) Complementary genes (b) Inhibitory genes
(c) Recessive epistasis (d) Dominant epistasis

38. Exchange of genetic material between chromatids of homologous chromosomes during meiosis is called **[1996, 2007, 2012, 2013]**

(a) Synapsis (b) Chiasmata
(c) Transformation (d) Crossing over

39. Which one of the following is the most suitable medium for culture of *Drosophila melanogaster* **[2006]**

(a) Ripe banana (b) Cow dung
(c) Moist bread (d) Agar agar

40. Crossing over in diploid organism is responsible for **[1991, 1998, 2010]**

(a) Dominance of genes
(b) Linkage between genes
(c) Segregation of alleles (genes)
(d) Recombination of linked allele (genes)

41. Crossing over that results in genetic recombination in higher organisms occurs between **[2004, 2004, 2004, 2006, 2010]**

(a) Two daughter nuclei
(b) Two different bivalents
(c) Sister chromatids of a bivalent
(d) Non-sister chromatids of a bivalent

42. Number of linkage group in *Pisum sativum* is **[2004]**

Or

How many pairs of contrasting characters in pea plants were studied by Mendel in his experiments **[2015]**

(a) 2 (b) 5
(c) 7 (d) 9

43. The linkage map of X-chromosome of fruit fly has 66 units with yellow body gene (y) at one end and bobbed hair (b) gene at the other end. The recombination frequency between these two genes (y and b) should be **[2003]**

(a) 100 % (b) 66 %
(c) 50 % (d) 5.50 %

44. Select the correct statement from the ones given below with respect to dihybrid cross **[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

45. 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 **[1990, 2005]**

(a) Law of segregation
(b) Law of multiple factor hypothesis
(c) Law of independent assortment
(d) Law of dominance

46. In a testcross involving F_1 dihybrid files, more parental-type offspring were produced than the recombinant type offspring. This indicates **[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

47. Which one of the following pairs is correctly matched **[1993, 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

48. 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 **[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

49. Which of the following statements is not true of two genes that show 50% recombination frequency **[2013]**

(a) If the genes are present on the same chromosome, they undergo more than one cross overs in every meiosis
(b) The genes may be on different chromosomes
(c) The genes are tightly linked
(d) The genes show independent assortment

50. When a cluster of genes shows linkage behaviour they **[2003]**

(a) Induce cell division
(b) Do not show a chromosome map
(c) Show recombination during meiosis
(d) Do not show independent assortment

51. The exchange of one part of a chromosome to the other part of same or another chromosome is called **[2002]**

Or

The movement of gene from one linkage group to another is called **[2015, 2015]**

(a) Inversion (b) Mutation
(c) Translocation (d) Linkage

52. Select the correct match **[2018]**

(a) G. Mendel – Transformation
(b) T.H. Morgan – Transduction
(c) F_2 × Recessive parent – Dihybrid cross
(d) Ribozyme – Nucleic acid

53. Centromere is a part of chromosome which helps in the **[1995, 1998, 2001]**

(a) Division of centrosomes
(b) Formation of spindle fibres
(c) Movement of chromosomes
(d) Formation of nuclear spindle

54. The structure present over chromosome is **[1987, 2002, 1995, 2003, 1997]**

(a) Nucleolus (b) Centromere
(c) Centrochrome (d) Golgi bodies

55. An unfertilized human egg contains **[1991, 1992, 1993, 1999, 2000, 1995]**

(a) Two *X* chromosomes
(b) One *X* and *Y* chromosome
(c) One *Y* chromosome only
(d) One *X* chromosome only

56. A chromosome, in which the centromere is situated close to its end so that one arm is very short and other very long is **[1997, 1998, 2002,2009, 2015]**

(a) Acrocentric (b) Metacentric
(c) Sub-metacentric (d) Telocentric

57. Tizo and Levan's contribution is very significant because they **[1993]**

(a) Gave the number of human chromosomes
(b) Pointed out mutational changes
(c) Identified Barr bodies
(d) Detected sex linkage

58. A child receives **[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

59. DNA elements, which can switch their position, are called **[1998, 2004, 2005, 2006, 2009, 2010, 2012]**

(a) Exons
(b) Introns
(c) Cistrons
(d) Transposons/Jumping genes

60. The point at which the polytene chromosomes appear to be attached together is known as **[1995, 2006]**

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

61. Telomerase is an enzyme which is a **[2005]**

(a) Repetitive DNA (b) RNA
(c) Simple protein (d) Ribonucleoprotein

62. Genes located at the same locus of chromosomes are called **[1997, 2015]**

(a) Polygenes (b) Oncogenes
(c) Multiple alleles (d) None of these

63. A gene is said to be dominant, if **[1992, 2002]**

(a) It is never expressed in any condition
(b) It is expressed only in heterozygous condition
(c) It expresses its effect only in homozygous stage
(d) It is expressed both in homozygous and heterozygous conditions

64. Which organism was used by Beadle and Tatum to proposed one gene-one enzyme hypothesis **[2004, 2004, 2007]**

(a) *E.coli* (b) Nostoc
(c) Drosophila (d) Neurospora

65. Which one of the following conditions of the zygotic cell would lead to the birth of a normal human female child **[1993, 2011]**

(a) Only one *X* chromosome
(b) One *X* and one *Y* chromosome
(c) Two *X* chromosomes
(d) Only one *Y* chromosome

66. "One gene one enzyme" theory was proposed by **[1994, 1995, 2000, 2001, 2004, 2003,2006, 2008]**

(a) G.W. Beadle and E.L. Tatum
(b) O.T. Avery and M. McCarthy
(c) J.H. Tijo and A. Levan
(d) C.E. Ford and J.H. Tijo

67. In split genes, the coding sequences are called **[1995]**

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

68. What would be the number of chromosomes of the aleurone cells of a plant with 42 chromosomes in its roots tip cells **[2011]**

(a) 21 (b) 42
(c) 63 (d) 84

69. What are those structures that appear as 'beads-on-string' in the chromosomes when viewed under electron microscope **[2011]**

(a) Base pairs (b) Genes
(c) Nucleotides (d) Nucleosomes

70. The fruit fly *Drosophila melanogaster* was found to be very suitable for experimental verification of chromosomal theory of inheritance by Morgan and his colleagues because **[2010]**

(a) It reproduces parthenogenetically
(b) A single mating produces two young flies
(c) Smaller female is easily recognisable from larger male
(d) It completes life cycle in about two weeks

71. Each chromosome at the anaphase stage of a bone marrow cell in our body has **[1995]**

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

72. Identify the correct order of organization of genetic material from largest to smallest **[2015]**

(a) Genome, chromosome, nucleotide, gene
(b) Genome, chromosome, gene, nucleotide
(c) Chromosome, genome, nucleotide, gene
(d) Chromosome, gene, genome, nucleotide

73. Depending upon size and centromere position, the 46 chromosomes have been divided into a number of groups **[1993, 2002]**

(a) 6 (b) 5
(c) 7 (d) 10

74. The grouping of human chromosomes is based on **[1993]**

(a) Secondary constrictions alone
(b) Dot–like satellites alone
(c) Banding patterns alone
(d) All the above

75. The polytene chromosomes were discovered for the first time in **[1995, 2012]**

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

76. Telomere repetitive DNA sequences control the function of eukaryote chromosomes because they **[2007]**
(a) Act as replicons
(b) Are RNA transcription initiator
(c) Help chromosome pairing
(d) Prevent chromosome loss

77. Lampbrush chromosomes are visible **[1996, 2006]**
(a) In diplotene of meiosis (b) In prophase of meiosis
(c) In interphase (d) In metaphase of meiosis

78. A male human is heterozygous for autosomal genes *A* and *B* and is also hemizygous for hemophilic gene *h*. What proportion of his sperms will be *abh* **[2004]**
(a) $\frac{1}{16}$ (b) $\frac{1}{4}$
(c) $\frac{1}{8}$ (d) $\frac{1}{32}$

79. The recessive genes located on *X*-chromosome in humans are always **[2004]**
(a) Expressed in males (b) Expressed in females
(c) Lethal (d) Sub-lethal

80. At a particular locus, frequency of '*A*' allele is 0.6 and that of '*a*' is 0.4. What would be the frequency of heterozygotes in a random mating population of equilibrium **[2005]**
(a) 0.16 (b) 0.48
(c) 0.36 (d) 0.24

81. Wild type *E.coli* cells are growing in normal medium with glucose. They are transferred to a medium containing only lactose as the sugar. Which one of the following changes take place **[1995]**
(a) The lac–Operon is repressed
(b) All Operons are induced
(c) *E. coli* cells stop dividing
(d) The lac–Operon is induced

82. A woman has an X-linked condition on one of her *X* chromosomes. This chromosome can be inherited by **[2018]**
(a) Both sons and daughters
(b) Only grandchildren
(c) Only sons
(d) Only daughters

83. ABO blood grouping is controlled by gene *I* which has three alleles and show co-dominance. There are six genotypes. How many phenotypes in all are possible **[2007, 2010, 2012]**
(a) Six (b) Three
(c) Four (d) Five

84. Inheritance of blood group is a condition of
(A) Co-dominance
(B) Incomplete dominance
(C) Multiple allelism
(D) Multiple gene **[1990, 2006, 2008, 2009]**
(a) A, B (b) B, D
(c) B, C (d) A, D
(e) A, C

85. Who was the scientist to introduce ABO blood groups **[1988, 1989, 1993, 1994, 2005]**
(a) Wiener (b) Levine
(c) Fisher (d) Landsteiner

86. Which one of the following is an example of polygenic inheritance **[2006, 2007, 2008, 2013]**
(a) Pod shape in garder pea
(b) Skin colour in humans
(c) Flower colour in *Mirabilis jalapa*
(d) Production of male honey bee

87. Which of the following are most abundant types of antibodies **[1999, 2008, 2011, 2012]**
(a) *IgA* (b) *IgE*
(c) *IgG* (d) *IgM*

88. The problem due to Rh^- factor arises when the blood two (Rh^+ and Rh^-) mix up **[1999]**
(a) In a test tube (b) Through transfusion
(c) During pregnancy (d) Both (a) and (c)

89. When dominant and recessive alleles express themselves together, it is called **[2001]**
(a) Dominance (b) Co-dominance
(c) Amphidominance (d) Pseudodominance

90. Biological marriage of one of the following should be avoided **[1990, 1995, 2001]**

Or

After examining the blood groups of a couple, the doctor advised them not to have more than one child. The blood group of the couple are likely to be **[1990, 1990, 1995, 2000, 2002, 2003, 2005, 2006, 2008]**

Or

In which of the following situations, is there a risk factor for children of incurring erythroblastosis foetalis **[2010]**
(a) Rh^+ male and Rh^- female
(b) Rh^+ male and Rh^+ female
(c) Rh^- male and Rh^+ female
(d) Rh^- male and Rh^- female

91. A child's blood group is '*O*'. The parents blood groups cannot be **[1994, 2005]**
(a) AB and O (b) B and O
(c) A and B (d) A and A

92. The most popularly known blood grouping is the ABO grouping. It is named ABO and not ABC, because "O" in it refers to having **[2009]**
(a) Other antigens besides A and B on RBCs
(b) Overdominance of this type of the genes for A and B types
(c) One antibody only-either anti-A or anti-B on the RBCs
(d) No antigens A and B on RBCs

93. Person with blood group AB is considered as universal recipient because he has **[1992, 1995, 1996, 1999, 2003, 2006, 2014]**
(a) No antigen on RBC and no antibody in the plasma
(b) Both A and B antigens in the plasma but no antibodies
(c) Both A and B antigens on RBC but no antibodies in the plasma
(d) Both A and B antibodies in the plasma

94. A person with unknown blood group under ABO system, has suffered much blood loss in an accident and needs immediate blood transfusion. His one friend who has a valid certificate of his own blood type, offers for blood donation without delay. What would have been the type of blood group of the donor friend **[2011,2012]**
(a) Type A (b) Type B
(c) Type AB (d) Type O

95. You are required to draw blood from a patient and to keep it in a test tube for analysis of blood corpuscles and plasma. You are also provided with the following four types of test tubes. Which of them will you ***not*** use for the purpose. **[2004]**
(a) Test tube containing heparin
(b) Test tube containing sodium oxalate
(c) Test tube containing calcium bicarbonate
(d) Chilled test tube

96. If two persons with 'AB' blood group marry and have sufficiently large number of children, these children could be classified as 'A' blood group : 'AB' blood group 'B' blood group in 1 : 2 : 1 ratio. Modern technique of protein electrophoresis reveals presence of both 'A' and 'B' type proteins in 'AB' blood group individuals. This is an example of **[2013]**
(a) Complete dominance (b) Co-dominance
(c) Incomplete dominance (d) Partial dominance

97. Which of the following substances, if introduced into the blood stream, would cause coagulation of blood at the site of its introduction **[2005]**
(a) Fibrinogen (b) Prothrombin
(c) Heparin (d) Thromboplastin

98. In a mutational event, when adenine is replaced by guanine, it is a case of **[2004]**
(a) Transition (b) Transversion
(c) Frameshift mutation (d) Transcription

99. Which of the following characteristics represent 'Inheritance of blood groups' in humans
(i) Dominance
(ii) Co – dominance
(iii) Multiple allele
(iv) Incomplete dominance
(v) Polygenic inheritance **[2018]**
(a) (i), (iii) and (v) (b) (ii), (iv) and (v)
(c) (i), (ii) and (iii) (d) (ii), (iii) and (v)

100. Which of the following pairs is wrongly matched **[2018]**
(a) T.H. Morgan : Linkage
(b) XO type sex determination : Grasshopper
(c) ABO blood grouping : Co-dominance
(d) Starch synthesis in pea : Multiple alleles

101. Mongoloid condition is related to **or** In mongolism a patient shows **[1995, 2001]**
(a) Monosomy (b) Trisomy
(c) Nullisomy (d) None of the above

102. Which of the following is a genetic disease **[1990, 1993]**
(a) Phenylketonuria (b) Blindness
(c) Cataract (d) Leprosy

103. Point (Gene mulation) mutation involves **[1995, 2001, 2009]**
(a) Insertion (b) Change in single base pair
(c) Duplication (d) Deletion

104. A person who is trisomic for twenty first pair of chromosomes is **[1990, 1993, 2000, 2008, 2012, 2013]**
(a) Klinefelter's syndrome (b) Down's syndrome
(c) Turner's syndrome (d) None of these

105. Disorders of amino acid metabolism results in **[1993, 2004]**
(a) Alkaptonuria (b) Phenylketonuria
(c) Albinism (d) All the above

106. Condition of sex chromosomes in a male child of Down's syndrome will be **[1991]**
(a) *XY* (b) *XXY*
(c) *XX* (d) *XO*

107. The most striking example of point mutation is found in a disease called **[1995]**
(a) Night blindness (b) Thalassemia
(c) Down's syndrome (d) Sickle-cell anaemia

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

109. Mutation rates are affected by **[1990, 1997, 2006, 2011]**
(a) Temperature
(b) X–rays
(c) Gamma and beta radiation
(d) All of the above

110. Which of the following is the main category of mutation **[1999]**
(a) Genetic mutation (b) Zygotic mutaion
(c) Somatic mutaion (d) All of these

111. Mental retardation in man, associated with sex chromosomal abnormality is usually due to **[1998]**
(a) Reduction in *X* complement
(b) Increase in *X* complement
(c) Moderate increase in *Y* complement
(d) Large increase in *Y* complement

112. The formation of multivalents at meiosis in diploid organism is due to **[1998]**
(a) Monosomy
(b) Inversion
(c) Deletion
(d) Reciprocal translocation

113. Both sickle cell anaemia and Huntington's chorea are **[2006]**
(a) Pollutant-induced disorders
(b) Virus-related diseases
(c) Bacteria-related diseases
(d) Congenital disorders

114. The chromosomal condition in Turner' syndrome is **[2011, 2012]**

Or

A human female with Turner's syndrome **[2014]**
(a) 21 Trisomy with XY (b) 44 Autosomes + XXY
(c) 44 Autosomes + XYY (d) 44 Autosomes + XO
(e) 18 Trisomy with XY

115. An abnormal human baby with 'XXX' sex chromosomes was born due to **[2015]**
(a) Formation of abnormal ova in the mother
(b) Fusion of two ova and one sperm
(c) Fusion of two sperms and one ovum
(d) Formation of abnormal sperms in the father

116. The genetic defect-adenosine deaminase (ADA) deficiency may be cured permanently by **[2009, 2012]**
(a) Periodic infusion of genetically engineered lymphocytes having functional ADA cDNA
(b) Administering adenosine deaminase activators
(c) Introducing bone marrow cells producing ADA into cells at early embryonic stages
(d) Enzyme replacement therapy

117. Alzhimer disease in humans is associated with the deficiency of **[2009]**
(a) Dopamine
(b) Glutamic acid
(c) Acetylcholine
(d) Gamma aminobutyric acid (GABA)

118. Sickle cell anaemia is **[2009]**
(a) An autosomal linked dominant trait
(b) Caused by substitution of valine by glutamic acid in the beta globin chain of haemoglobin
(c) Caused by a change in a single base pair of DNA
(d) Characterized by elongated sickle like RBCs with a nucleus

119. Which one of the following conditions in humans is correctly matched with its chromosomal abnormality/ linkage **[1994, 1998, 2000, 2002, 2003, 2008, 2012]**

Or

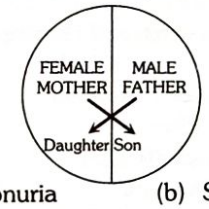
An abnormal human male phenotype involving an extra X-chromosome is a case of **[1995 1996, 1997, 2003, 2004]**

(a) Erythroblastosis foetalis - X-linked
(b) Down syndrome – 44 autosomes + XO
(c) Klinefelter's syndrome – 44 autosomes +XXY
(d) Colouor blindness – Y-linked

120. Haploids are more suitable for mutation studies than the diploids. This is because **[2008]**

(a) Haploids are more abundant in nature than diploids
(b) All mutations, whether dominant or recessive are expressed in haploids
(c) Haploids are reproductively more stable than diploids
(d) Mutagens penetrate in haploids more effectively than is diploids

121. Represented below is the inheritance pattern of a certain type of traits in humans. Which one of the following conditions could be an example of this pattern **[2012]**



(a) Phenylketonuria (b) Sickle cell anaemia
(c) Haemophilia (d) Thalassemia

122. Albinism is a congenital disorder (non synthesis of melanin) resulting from the lack of the enzyme **[1994, 2003, 2012]**

(a) Catalase (b) Fructokinase
(c) Tyrosinase (d) Xenthine oxidase

123. Which one of the following is a wrong statement regarding mutations **[2012]**

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

124. If a diploid cell is treated with colchicine, then it becomes **[2002]**

(a) Tetraploid (b) Diploid
(c) Triploid (d) Monoploid

125. Down's syndrome is caused by an extra copy of chromosome number 21. What percentage of offspring produced by an affected mother and a normal father would be affected by this disorder **[2003, 2007]**

(a) 25% (b) 100%
(c) 75% (d) 50%

126. Albinism is known to be due to an autosomal recessive mutation. The first child of a couple with normal skin pigmentation was an albino. What is the probability that their second child will also be an albino **[1998]**

(a) 100% (b) 25%
(c) 50% (d) 75%

127. If both parents are carriers for thalessemia, which is an autosomal recessive disorder, what are the chances of pregnancy resulting in an affected child **[2013]**

(a) 100% (b) No chance
(c) 50% (d) 25%

128. Genetic variation in a population arises due to **[2013]**

(a) Recombination only
(b) Mutations as well as recombination
(c) Reproductive isolation and selection
(d) Mutations only

129. The **incorrect** statement with regard to haemophilia is **[2013]**

(a) A single protein involved in the clotting of blood is affected
(b) It is a sex-linked disease
(c) It is a recessive disease
(d) It is a dominant disease

130. How many genome types are present in a typical green plants cell **[1998]**

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

131. A cell at telophase stage is observed by a student in a plant brought from the field. He tells his teacher that this cell is not like other cells at telophase stage. There is no formation of cell plate and thus the cell is containing more number of chromosomes as compared to other dividing cells. This would result in **[2016]**

(a) Aneuploidy (b) Polyploidy
(c) Somaclonal variation (d) Polyteny

132. Pleitropy is a condition in which a single gene **[2005, 2006, 2013, 2015]**

(a) Controls only one phenotype
(b) Controls more than one phenotype
(c) Does not control any phenotype
(d) None of these

133. Barr bodies (seen in saliva test in Olympic games) are found in human and are associated with **[1992, 1997, 1998, 1999, 2002, 2005]**

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

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

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

135. Barr bodies are **[1993]**

(a) Chromatin negative (b) Not influenced by stains
(c) Chromatin positive (d) Poorly staining

136. Foetal sex can be determined by examining cells from amniotic fluid looking for **[1991]**

(a) Barr bodies (b) Chiasmata
(c) Sex chromosomes (d) Kinetochores

137. *Drosophila* flies with one half of the body male and other half female is referred to as **[1994]**

(a) Gynandromorph (b) Hermaphrodite
(c) Super female (d) Intersex

138. In human female, barr bodies are formed by **[1996]**

(a) Inactivation of mother's *X* chromosome
(b) Inactivation of father's *X* chromosome
(c) Inactivation of both mother's and father's *X* chromosomes
(d) Inactivation of either mother's or father's *X* chromosome

140. Genetic identity of a human male is determined by **[1997, 1999, 2000]**

(a) Autosome (b) Nucleolus
(c) Sex chromosome (d) Cell organelles

141. In *Drosophila*, the sex is determined by **[1994, 1995, 2003, 2004, 2011]**

(a) Whether the egg is fertilized or develops parthenogenetically

(b) The ratio of number of X-chromosomes to the sets of autosomes

(c) X and Y chromosomes

(d) The ratio of pairs of X-chromosomes to the pairs of autosomes

142. In our society women are blamed for producing female children. Choose the correct answer for the sex-determination in humans **[1990, 2013]**

(a) Due to some defect like aspermia in man

(b) Due to the genetic make up of the particular sperm which fertilizes the egg

(c) Due to the genetic make up of the egg

(d) Due to some defect in the women

143. Loss of a X chromosome in a particular cell during its development, results into **[1998]**

(a) Diploid individual (b) Triploid individual

(c) Gynandromorphs (d) (a) and (b) both

144. Which one of the following conditions correctly describes the manner of determining the sex in the given example **[2011, 2015]**

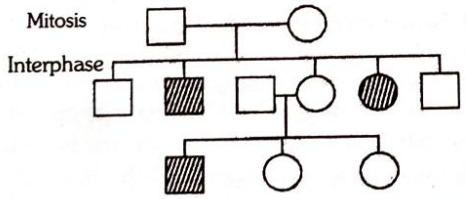
(a) Homozygous sex chromosomes (XX) produce male in *Drosophila*

(b) Homozygous sex chromosomes (ZZ) determine female sex in birds

(c) XO type of sex chromosomes determine male sex in grasshopper

(d) XO condition in humans as found in Turner Syndrome, determines female sex

145. Study the pedigree chart given below



What does its show **[2009]**

(a) Inheritance of a sex-linked inborn error of metabolism like phenylketonuria

(b) Inheritance of a condition like phenylketonuria as an autosomal recessive trait

(c) The pedigree chart is wrong as this is not possible

(d) Inheritance of a recessive sex-linked disease like haemophilia

146. Which one of the following symbols and its representation, used in human pedigree analysis is correct **[2010]**

(a) ◆ = male affected

(b) □═○ = mating between relatives

(c) ○ = unaffected male

(d) □ = unaffected female

147. Which of the following is not a hereditary disease **[2005]**

(a) Cretinism (b) Cystic fibrosis

(c) Thalassaemia (d) Haemophilia

148. Sickle cell anaemia is due to **[1990]**

(a) Hormones (b) Viruses

(c) Genes (d) Bacteria

149. All the sons are haemophilic and daughter are normal of a haemophilic father and normal mother. This character is **[1996]**

(a) X–linked recessive (b) Y–linked recessive

(c) X–linked dominant (d) Y–linked dominant

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

(a) Recessive (b) Dominant

(c) Codominant (d) Complementary

151. A fruit fly is heterozygous for sex-linked genes when meted with normal female fruit fly, the males specific chromosome will enter egg cell in the proportion **[1997]**

(a) 1 : 1 (b) 2 : 1

(c) 3 : 1 (d) 7 : 1

152. A diseased man marries a normal woman. They get three daughters and five sons. All the daughters were diseased and sons were normal. The gene of this disease is **[2002]**

(a) Autosomal dominant (b) Sex linked recessive

(c) Sex limited character (d) Sex linked dominant

153. Pattern baldness, moustaches and beard in human males are examples of **[2003]**

Or

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

(a) Sex-determining traits (b) Sex linked traits

(c) Sex limited traits (d) Sex differentiating traits

154. Which one is the **incorrect** statement with regards to the importance of pedigree analysis **[2013]**

(a) It confirms that DNA is the carrier of genetic information

(b) It helps to understand whether the trait in question is dominant or recessive

(c) It confirms that the trait is linked to one of the autosome

(d) It helps to trace the inheritance of a specific trait

155. A colour blind man marries the daughter of a colour blind person. Then in their progeny **[1988, 1992, 1994, 2002, 2008]**

(a) None of their daughters are colour blind

(b) All the sons are colour blind

(c) All the daughters are colour blind

(d) Half of their sons are colour blind

156. Select the incorrect statement from the following **[2009]**

(a) Linkage is an exception to the principle of independent assortment in heredity

(b) Galactosemia is an inborn error of metabolism

(c) Small population size results in random genetic drift in a population

(d) Baldness is a sex-limited trait

157. A normal-visioned man whose father was colour-blind, marries a woman whose father was also colour-blind. They have their first child as a daughter. What are the chances that this child would be colour-blind **[1990, 2009, 2012]**

(a) 50% (b) 100%

(c) 0% (d) 25%

158. If a colourblind woman marries and a normal visioned man, their sons will be **[1994, 1996, 1999, 2000, 2005, 2006]**

(a) Three-fourths colourblind and on-fourth normal

(b) All colourblind

(c) All normal visioned

(d) One-half colourblind and one-half normal

159. A woman with normal vision, but whose father was colour blind, marries a colour blind man. Suppose that the fourth child of this couple was a boy. Thus boy **[2005]**

(a) Will be partially colour blind since he is heterozygoustor for the colour blind mutant alble
(b) Must have normal colour vision
(c) Must be colour blind
(d) May be colour blind or may be of normal vision

160. A man and a woman, who do not show any apparent signs of a certain inherited disease, have seven children (2 daughters and 5 sons). Three of the sons suffer from the given disease but none of the daughters are affected which of the following mode of inheritance do you suggest for this disease **[2005]**

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

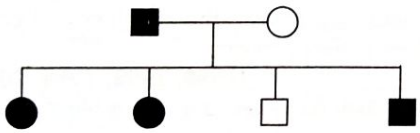
161. Haemophilia is more commonly seen in human males than in human females because **[2005, 2008]**

(a) This disease is due to an X-linked dominant mutation
(b) A grater proportion of girls die in infancy
(c) This disease is due to an X-linked recessive mutation
(d) This disease is due to a Y-linked recessive mutation

162. A woman with two genes for haemophillia and one gene for colourblindness on one of the *X* chromosomes marries a normal man. How will the progeny be **[1998, 2012]**

(a) All sons and daughters haemophillic and colourblind
(b) Haemophillic and colourblind daughters
(c) 50% haemophillic colourblind sons and 50% normal sons
(d) 50% haemophillic daughters and 50% colourblind daughters

163. Study the pedigree chart of a certain family given below and select the correct conclusion which can be drawn for the character **[2010]**



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

164. A colour blind man marries a woman with normal sight who has no history of colour blindness in her family. What is the probability of their grandson being colour blind **[2015]**

(a) 1 (b) Nil
(c) 0.25 (d) 0.5

165. Pick out the correct statements

(A) Haemophilia is a sex-linked recessive disease
(B) Down's syndrome is due to aneuploidy
(C) Phenylketonuria is an autosomal recessive gene disorder
(D) Sickle cell anaemia is a X-linked recessive gene disorder **[2016]**

(a) (A) and (D) are correct
(b) (B) and (D) are correct
(c) (A), (C) and (D) are correct
(d) (A), (B) and (C) are correct

166. If a colour-blind man marries a woman who is homozygous for normal colour vision, the probability of their son being colour-blind is **[2016]**

(a) 1 (b) 0
(c) 0.5 (d) 0.75

167. An organism which receives identical alleles of a particular gene from both parents is **[1993]**

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

168. Study of improvement of human race by providing ideal nature is **[1990, 1991, 1998]**

Or

Improvement of genetic characters and present day generation on the basis of best nutrition and training is called **[1995]**

(a) Eugenics (b) Euphenics
(c) Euthenics (d) None of these

169. Conjoint twins are also known as **[1988]**

(a) Fraternal twins (b) Siamese twins
(c) Zygotic twins (d) None of these

11. AIIMS

1. Who is known as father of physiological genetics or father of biochemical genetics **[2009]**

(a) Slatyer (b) Charles Elton
(c) Taylors (d) Archibald Garrod

2. Pure homozygous offsprings in a dihybrid cross in the F_2 generation will be **[1993]**

(a) 1/2 (b) 1/4
(c) 1/8 (d) 1/16

3. The percentage of heterozygous individuals obtained in F_2 generation from selfing the plants with genotype *Rr* would be **[1994]**

(a) 24 (b) 50
(c) 75 (d) 100

4. 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 **[1993]**

(a) *Rr Pp* × *rr pp* (b) *RR PP* × *rr pp*
(c) *RR pp* × *rr pp* (d) *RR Pp* × *rr pp*

5. 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 **[1993]**

(a) 1 smooth : 3 sugary (b) 3 smooth : 1 sugary
(c) 1 smooth : 1 sugary (d) All sugary

6. How many different types of gametes can be formed by F_1 progeny, resulting from the following cross

AA BB CC × aa bb cc **[2004]**

(a) 3 (b) 8
(c) 27 (d) 64

7. Knowing that albinism is determined by a recessive gene in man; presence of albinism in children born to a couple proves that **[1992]**

(a) Both the father and the mother are heterozygous for albinism
(b) The father is homozygous normal but the mother is heterozygous or vice versa
(c) The father is homozygous for albinism but the mother is heterozygous or vice versa
(d) (a) and (c) are correct

8. Genes present in the cytoplasm of eukaryotic cells, are found in **[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

9. *Mirabilis jalapa* is a good example of **[2001]**
(a) Complete dominance (b) Plastid inheritance
(c) Both (a) and (b) (d) None of the above

10. 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 **[2005, 07, 08]**
(a) None (b) Less than 5 per cent
(c) One third (d) Half

11. Which of the following is associated with multiple phenotypes **[2000]**
(a) Epistasis (b) Pleiotropy
(c) Polygenic inheritance (d) Mutation

12. When synapsis is complete all along the chromosome, the cell is said to have entered a stage called **[2005]**
(a) Zygotene (b) Pachytene
(c) Diplotene (d) Diakinesi

13. Distance between two linked genes upon a chromosome is measured in cross over units, is **[1998]**
(a) Ratio of crossing over between them
(b) Cross-over value
(c) Number of other genes between them
(d) None of these

14. The genes, which are confined to differential region of *Y* chromosome only, are called **[1998]**
(a) Mutant (b) Autosomal
(c) Holandric (d) Completely sex-linked

15. The total number of nitrogenous bases in human genome is estimated to be about **[2004, 08]**
(a) 3.5 million (b) 35 thousand
(c) 35 million (d) 3.1 billion

16. Holandric genes are **[2010]**
(a) Carried by 'X' chromosomes
(b) Carried by different parts of 'Y' chromosomes
(c) Carried by 'X' and 'Y' chromosomes
(d) Carried by autosomes

17. The bacterial genome refers to the total number of genes located upon a **or** The term 'genome' refers to the total number of genes combined in a **[1994]**
(a) Haploid set of chromosomes
(b) Diploid set of chromosomes
(c) Tetraploid set of chromosomes
(d) Hexaploid set of chromosomes

18. Primary source of allelic variation is **[2005]**
(a) Independent assortment (b) Recombination
(c) Mutation (d) Polyploidy

19. Usually the recessive character is expressed only when present in a double recessive condition. However, a single recessive gene can express itself in human beings when the gene is present on **[1992]**
(a) Any autosome
(b) *X* chromosome of female
(c) *X* chromosome of male
(d) Either on autosome or *X* chromosome

20. During serological test in which anti–human serum is mixed with blood of another animal, blood of which animal gives the thickest precipitate **[1993]**
(a) Gibbon (b) Chimpanzee
(c) Dog (d) Mule

21. A woman is married for the second time. Her first husband was *ABO* blood type *A*, and her child by that marriage was type *O*. Her new husband is type *B* and their child is type *AB*. What is the woman's *ABO* genotype and blood type **[2009]**
(a) I^AI^O; Blood type *A* (b) I^AI^B; Blood type *AB*
(c) I^BI^O; Blood type *B* (d) I^OI^O; Blood type *O*

22. The monosomic condition in human beings depicted as XO is referred to as **[1999]**
(a) Criminal syndrome (b) Down's syndrome
(c) Klinefelter's syndrome (d) Turner's syndrome

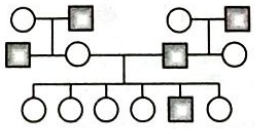
23. Genotype of a Down's syndrome is **[1993]**
(a) 45 + *XX* (b) 44 + *XY*
(c) 44 + *XXY* (d) 22 + *XY*

24. Discontinuous variations are **[2001]**
(a) Mutations (b) Acquired characters
(c) Essential features (d) Nonessential features

25. The "cri-du-chat" syndrome is caused by change in chromosome structure involving **[2005]**
(a) Deletion (b) Duplication
(c) Inversion (d) Translocation

26. The sex determination pattern in honeybee is called **[1993]**
(a) Female haploidy (b) Haplodiploidy
(c) Gametic diploidy (d) Gametogony

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



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

28. The expression of genes for the production of milk in only females is a **[1993]**
(a) X linked character (b) Y linked character
(c) Sex limited genes (d) Sex influenced genes

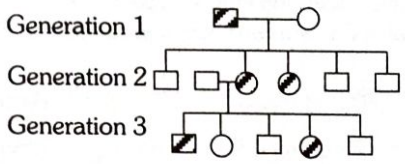
29. Haemophilia is caused due to lack of **[1992]**
(a) ADH (b) AHF
(c) STH (d) ACTH

30. The daughter born to haemophilic father and normal mother could be **[1992]**
(a) Normal (b) Carrier
(c) Haemophilic (d) None

31. One of the genes present exclusively on the X-chromosome in humans is concerned with
(a) Baldness
(b) Red-green colour blindness
(c) Facial hair/moustaches in males
(d) Night blindness

32. Person whose father is colourblind marries a lady whose mother is daughter of a colourblind man. Their children will be **[2013]**
(a) All normal
(b) All colour blind
(c) All sons colour blind
(d) Some sons normal and some colour blind

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



Key : **[2005]**

- ☐ Unaffected male
- ◪ Affected male
- ○ Unaffected female
- ◒ Affected female

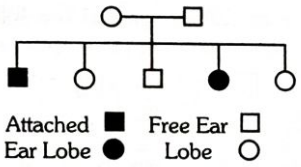
The trait traced in the above pedigree chart is

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

34. If a boy's father has haemophilia and his mother has one gene for haemophilia; what is the chance that the boy will inherit the disease **[1999]**

(a) 25% (b) 50%
(c) 75% (d) 100%

35. Given below is a pedigree chart of a family with five children. It shows the inheritance of attached earlobes as opposed to the free ones. The squares represent the male individuals and circles the female individuals



Which one of the following conclusions drawn is correct **[2004]**

(a) The parents are homozygous recessive
(b) The trait is Y-linked
(c) The parents are homozygous dominant
(d) The parents are heterozygous

36. A normal woman whose father was colourblind marries a normal man. What kinds of children would be expected and in what proportion **[2008]**

(a) Daughters normal, 50% of sons colourblind
(b) Daughters normal, all sons colourblind
(c) 50% of daughters colourblind, all sons normal
(d) All daughters colourblind, sons normal

12. Assertion & Reason

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

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

1. Assertion : In humans, the gamete contributed by the male determines whether the child produced will be male or female.
Reason : Sex in humans is a polygenic trait depending upon a cumulative effect of some genes on X-chromosome and some on Y-chromosome.

2. Assertion : Among the primates, chimpanzee is the closest relative of the present day humans.
Reason : The banding pattern in the autosome numbers 3 and 6 of man and chimpanzee is remarkably similar.

3. Assertion : Clones are produced by sexual reproduction and same sexual process.
Reason : These are prepared by group of cells descended from many cells or by inbreeding of a heterozygous line.

4. Assertion : The genetic complement of an organism is called genotype.
Reason : Genotype is the type of hereditary properties of an organism.

5. Assertion : Haemophilia never occurs in women.
Reason : Gene for haemophilia is located on X chromosome.

6. Assertion : Kinetochore helps in the movement of chromosomes.
Reason : It has points for attachment of microtubules.

7. Assertion : The lampbrush chromosomes are called diplotene chromosomes bivalents.
Reason : The number of loops is maximum during diplotene.

8. Assertion : Restriction endonuclease recognize short palindromic sequence and cut at specific sites.
Reason : When a restriction endonuclease acts on palindrome, it cleaves both the strands of DNA molecules.

9. Assertion : Somaclonal variations may be present in plants produced from callus.
Reason : Somaclonal variations are caused due to recombination during meiosis.