DAY TWENTY NINE

Genetics

Learning & Revision for the Day

- Genetic Terms
- Mendel and his Experiments
- Deviation from Mendelism
- Interaction of Genes
- Chromosomal Theory of Inheritance
- Linkage
- Crossing Over
- Sex-Determination
- Sex-Linked Inheritance
- Mutation
- Genetic Disorders

The study of heredity and variations is called **genetics**.

Heredity (e.g. like begets like) is the transmission of characteristics from parent to offsprings. Such transmissible characters are called **hereditary characters**.

Variations are differences in morphological, physiological, cytological and behavioural characters shown by the individuals of the same species, race and family. The main sources of variations are crossing over, chance combination of chromosomes during meiosis, fertilisation and mutations. Mutation is the ultimate source of variations.

Archibald Garrod is considered as the Father of Experimental Genetics. The term 'Genetics' was coined by **W Bateson** in 1905.

Genetic Terms

Different genetic terminology are as follows

1. **Allele** is one of two or more alternative forms of a gene, in the gene pool that occupies same locus on homologous chromosomes.
2. **Dominant allele** is an allele which is expressed in metabolism or the appearance (phenotype) of heterozygotes. One allele is said to be dominant over another if a heterozygous individual with one copy of that allele has the same appearance as a homozygous individual with two copies of it.
3. **Recessive allele** is an allele whose expression is masked in heterozygotes by the expression of a dominant allele.
4. **Wild** or **mutant alleles** The allele, which was originally present in the population and is usually most common and dominant is called wild allele; whereas recessive allele is less common and forms from wild allele through mutation. It is also called mutant allele.
5. **Gene** is the basic unit of heredity; a sequence of DNA nucleotides on a chromosome that encodes a polypeptide or RNA molecule and so determines the nature of an individual's inherited traits.
6. **Locus** is the location of a gene on a chromosome.
7. **Genotype** is the total set of genes present in the cells of an organism. This term is often used to refer the set of alleles of a single gene.
8. **Phenotype** is the physical appearance or metabolic expression of the genotype; the observable manifestation of a trait (affecting an individual's structure, physiology or

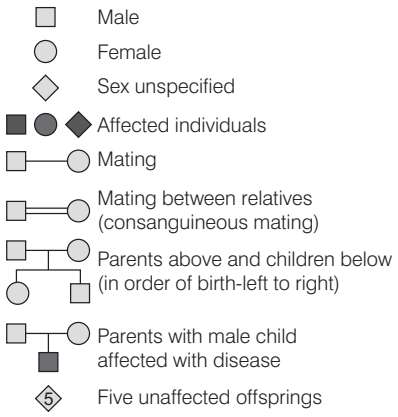
behaviour) that results from the biological activity of the DNA molecules.

9. **Haploid condition** is characterised by the presence of only one set of chromosomes. It is found in gametes, certain animals like protists and fungi and at certain stages in the life cycle of plants.
10. **Diploid condition** is characterised by the presence of two sets of chromosomes, which are referred to as homologous. Higher animals and plants are diploid in the dominant phase of their life cycles, so are some protists.
11. **Heterozygote** is a diploid individual carrying two different alleles of a gene on two homologous chromosomes. Most human beings are heterozygous for many genes.
12. **Homozygote** is a diploid individual carrying identical alleles of a gene on both homologous chromosomes.
13. **Hybrid** is an organism produced after crossing two genetically different individuals, such process of obtaining hybrids is known as hybridisation.
14. **Monohybrid cross** is made to study the inheritance of a single pair of allele.
15. **Dihybrid cross** is made to study the inheritance of two pairs of factors or alleles of two genes.
16. **Trihybrid ratio** is the ratio obtained in F_2-generation raised from a trihybrid cross followed by selfing or inbreeding of F_1 individuals. Eight phenotypes are formed. The phenotypic ratio is 27 : 9 : 9 : 9 : 3 : 3 : 3 : 1.
17. **Reciprocal cross** is a cross involving two types of individuals, where the male of one type is crossed with female of the second type and *vice-versa*.
18. **Back cross** is performed between F_1 progeny and any one of its parents.
19. **Test cross** is performed between F_1 progeny with its recessive parent. The test cross ratio in monohybrid cross is 1 : 1 and in dihybrid cross, ratio will be 1 : 1: 1 : 1.
20. **Pureline** is a strain of genetically pure, true breeding individuals, which have been derived from a single self-fertilised homozygous ancestor.

    It always produces offsprings which are true for its characters.
21. **Checker board Punnett square** is a square divided into smaller squares, which show the mathematical (probable) result of a cross, both phenotypic and genotypic. It is of three types, i.e. gametic, phenotypic and genotypic. Forked line or branching system is also used to know phenotypic and genotypic probabilities.

    It was devised by a British geneticist, Reginald Punnett in 1927.
22. **Genome** is the complete set of chromosome where every gene chromosome is represented singly as in gamete. A single genome is present in haploid cells, two in diploid cells and many in polyploid cells.
23. **Pedigree analysis** is the analysis of traits in several generations of a family. The inheritance of a particular trait is represented in the family tree over generations. It is a strong tool to trace the inheritance of a specific trait, abnormality or disease.

    Some specific symbols are used to show pedigree in a pedigree chart



Mendel and his Experiments

- **Gregor Johann Mendel**, an Austrian monk performed breeding experiments on the **garden pea** (*Pisum sativum*), for more than 8 years between 1856-1863. His experimental data was published in 1865.
- Mendel used garden pea for his experiments due to its well-defined characters, hermaphroditism, predominance of self-fertilisation, easy hybridisation and emasculation (i.e. removal of anthers before maturity).
- Mendel's success was mainly based on the fact that he considered a single character at one time.

Seven Pairs of Contrasting Characters Studied by Mendel

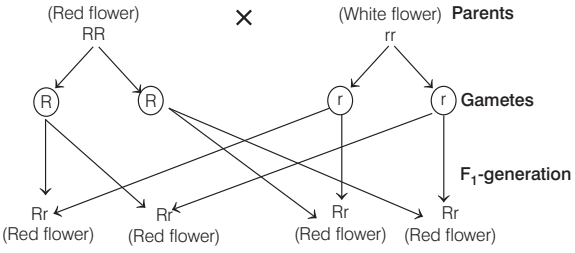
Characteristic	Chromosome Location (Known now)	Parental Appearance	
		Dominant	Recessive
Length of stem	4	Tall	Dwarf
Shape of seed	7	Round	Wrinkled
Colour of seed	1	Yellow	Green
Shape of pod	4	Inflated	Constricted
Colour of pod	5	Green	Yellow
Position of flower	4	Axial	Terminal
Colour of flower	1	Violet	White

- Genes controlling seven traits (characters) in pea studied by Mendel are now known to be located on only four chromosomes out of seven.
- Mendel's work was rediscovered by **Hugo de Vries** (Dutch biologist), **Carl Correns** (German botanist) and **Erich von Tschermak** (Austrian botanist) in 1900.

Three Laws of Inheritance

Mendel proposed three laws of heredity which are as follows

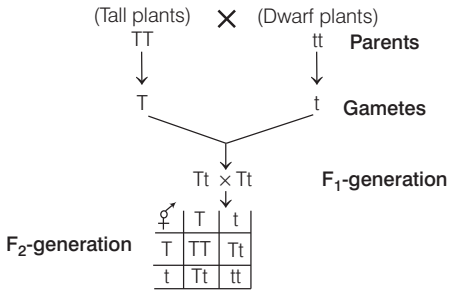
1. **Law of dominance** It states that only one of the forms of parental traits is expressed in the F_1, i.e. the F_1-generation always displays only one of the parental traits. He described that the trait which were always seen in F_1-generation are **dominant** traits and the trait that disappeared are **recessive.**



Monohybrid cross

2. **Law of segregation** It states that two alleles of each gene pair separate when gametes are produced during meiosis.

   Mendel's law of segregation is universal and without any exception. It is also called **law of purity of gametes** or **law of splitting of hybrids** because gametes always remain pure and may carry either the dominant or the recessive factor of a single gene but never both.



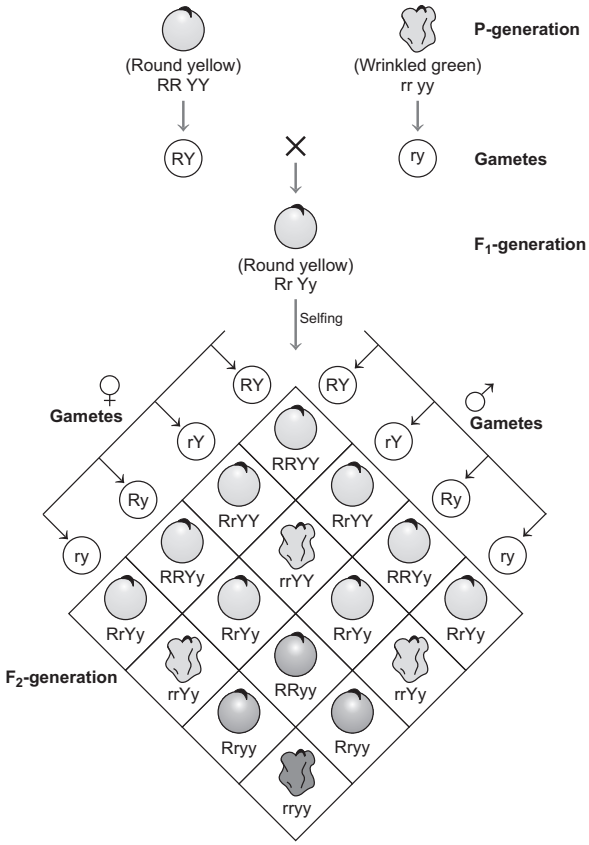
Phenotype ratio 3(Tall) : 1 (Dwarf)

Genotype ratio 1(TT) : 2 (Tt) : 1(tt)

Monohybrid cross

3. **Law of independent assortment** It states that pairs of alleles separate independently of each other during gamete formation. This law was deduced from dihybrid cross.

When a dihybrid cross is made and the offsprings of F_1-generation are self-bred, dihybrid ratio 9 : 3 : 3 : 1 (phenotypic ratio) is obtained, where 9/16 individuals carry both the dominant traits, 3/16 carry first dominant and second recessive trait, 3/16 carry first recessive and second dominant trait, where as, 1/16 carry both the recessive traits.



Phenotypic ratio Round yellow : Round green
9 3
: Wrinkled yellow : Wrinkled green
3 1

Genotypic ratio 1 : 2 : 2 : 4 : 1 : 2 : 1 : 2 : 1

Results of a dihybrid cross where the two parents differed in two pairs of contrasting traits : seed colour and seed shape

Deviation from Mendelism

There are many exceptions and deviations of Mendel's laws such as

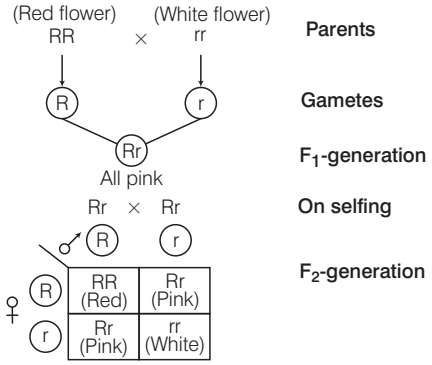
- Mendel observed that one allele dominates over the other but **incomplete dominance** in 4 O'clock plant *(Mirabilis jalapa)* and snapdragon are some exceptions.

- Mendel observed only two alleles for a character but body colour of rabbit shows **multiple allelism** (i.e. the presence of more than two alleles for a gene).
- As per law of independent assortment, any two or more than two pairs of characters assort independently but **linkage** is an exception for law of independent assortment.

1. Incomplete Dominance

- It is an exception to the law of dominance, where none of the two contrasting alleles is dominant.
- The expression of character in F_1-generation is intermediate or mixture of expression of two characters.
- This phenomenon can be observed in *Mirabilis jalapa* (4 O'clock plant) in which, as a result of hybridisation between white (rr) and red (RR) flowered plants, pink flowers are formed in F_1-generation and this event is known as incomplete dominance or blending inheritance.

Incomplete Dominance in Snapdragon



Phenotypic Ratio : Red : Pink : White
(RR) : (Rr) : (rr)
1 : 2 : 1

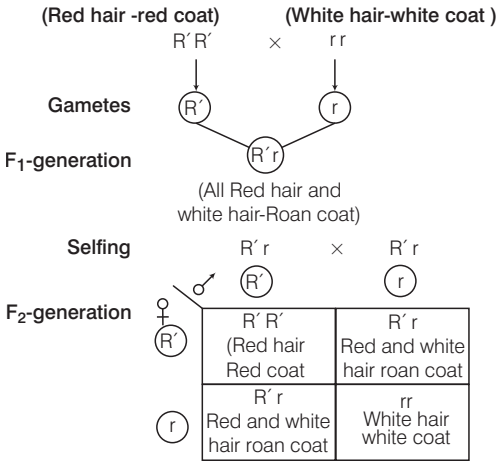
Monohybrid cross in snapdragon, where one allele is incompletely dominant over the other allele

First case of incomplete dominance or blending inheritance was reported in *Mirabilis jalapa* (4 O'clock plant) by **Carl Correns** (1903).

2. Codominance

- It is the phenomenon in which both alleles are expressed at the same time.
- This results in a different phenotype in the heterozygous individual, e.g. the roan coat of horses (i.e. patches of two different colours on the skin) illustrate this.

Genotype and Phenotype of Codominance in Horse



Phenotypic Ratio/ Genotypic Ratio – Red coat : Roan coat : White coat

(R´R´) : (R´r) : (rr)
1 : 2 : 1

3. Multiple Allelism (Inheritance of Human Blood Groups)

- It is the phenomenon of having genes with more than two alleles. These are called multiple alleles.
- The human blood group gene provides an example of multiple allele as well as an interesting dominance relationship.
- There are three A, B, O blood group alleles, usually given the symbols I^A, I^B and i. I^A and I^B are codominant to each other but both are dominant to i.
- Blood groups A, B and O were discovered by **Karl Landsteiner** whereas blood group AB was discovered by **von Decastello** and **Sturli.**
- A set of three multiple alleles present on the chromosome is responsible for four types of blood groups.
- These alleles are I^a, I^b and i. Here, letter I stands for isohemagglutinins.
- The allele I^a produces A antigen, I^b produces B antigen and i (or I^o) does not produce any antigen.

Possible Blood Groups of Children for Known Blood Groups of Parents

Blood groups of parents (Known)	Genotype of parents (known)	Blood groups of Children	
		Possible	Not Possible
O and O	$I^o\ I^o \times I^o\ I^o$	O	A, B, AB
O and A	$I^o\ I^o \times I^a\ I^o$	O, A	B, AB
A and A	$I^a\ I^o \times I^a\ I^o$	O, A	B, AB
O and B	$I^o\ I^o \times I^b\ I^o$	O, B	A, AB
B and B	$I^b\ I^o \times I^b\ I^o$	O, B	A, AB

Blood groups of parents (Known)	Genotype of parents (known)	Blood groups of Children	
		Possible	Not Possible
A and B	$I^a\ I^a \times I^b\ I^b$ $I^a\ I^a \times I^b\ I^o$ $I^a\ I^o \times I^b\ I^o$	O, A, B, AB	None
O and AB	$I^o\ I^o \times I^a\ I^b$	A, B	O, AB
A and AB	$I^a\ I^o \times I^a\ I^b$	A, B, AB	O
B and AB	$I^b\ I^o \times I^a\ I^b$	A, B, AB	O
AB and AB	$I^a\ I^b \times I^a\ I^b$	A, B, AB	O

NOTE Inheritance of blood groups in humans represents dominance (I^A or I^B over i), codominance (I^A and I^B) and multiple allelism (I^A, I^B and i).

Some other examples of multiple allelism are

(a) Colour loci in corn. (b) Skin colour in rodents.

(c) Eye colour in *Drosophila.*

(d) Self-incompatibility genes in some plants.

4. Pleiotropy

- It is the phenomenon in which a gene called pleiotropic gene that affects many aspects of a phenotype or controls several phenotypic characters.
- Pleiotropic genes along with their main effect also affect or modify the different genes and thus, have more than one phenotypic effect. Some examples of pleiotropy are
  - 'Vestigial wing' mutation of *Drosophila* not only controls the size and shape of the wings but also affects several other features including reduced fecundity.
  - Gene for phenylketonuria in human beings also interferes with synthesis of melanin pigment.
  - Sickle-cell anaemia and starch synthesis in pea.

5. Polygenic Inheritance

- It is the phenomenon in which genes have a small effect when acting individually but collectively produce a significant phenotypic expression. Such genes are called **polygenes**, e.g. genes for height or weight.
- Polygenes are present on different chromosomes and they segregate independently during meiosis. That's why polygenic or quantitative inheritance becomes a genetic problem for dihybrid cross (i.e. for two genes) and possibly much more, if more than two genes are involved for a particular trait.

Interaction of Genes

Mendel considered that each character is controlled by single gene but we know today that a character may be controlled by two or more than two genes. In such a situation, two or more than two genes may interact to give rise to a particular phenotype.

The gene interaction is of two types

1. Intragenic Interaction

In this type of interaction two alleles of a gene, which are present on same gene locus on the two homologous chromosomes interfere to produce modified phenotype, e.g. incomplete dominance, codominance, multiple allele and lethal genes, etc.

2. Intergenic Interaction

- It is one where two or more independent genes belonging to same or different chromosomes interact to form different expression, e.g. epistasis, duplicate genes, complementary genes, supplimentary genes, inhibitory genes, etc.
- If A and B are two genes, both dominant over their respective recessive alleles a and b, then the interaction will depend upon the presence of both dominant alleles A and B, the absence of A, the absence of B and the absence of both A and B.
- This interaction of genes will result into abbreviated phenotypic ratio, i.e. the phenotypic ratio other than expected 9 : 3 : 3 : 1.

(i) **In epistasis**, a gene at one locus 'masks' or inhibits the expression of a gene at a distinct locus.

- In epistasis, out of two independent genes one expressing itself is called **epistatic,** while other is masked called **hypostatic.**
- Such interactions give modified F_2 ratio as 12 : 3 : 1. Epistasis is of two types

(a) **Dominant epistasis**, e.g. fruit colour in squash.

(b) **Recessive epistasis**, e.g. coat colour of Labrador retrievers.

(ii) **Complementary genes** were observed by **W Bateson** and **RC Punnett** in sweet pea (*Lathyrus odoratus*).

In such type of interaction, two separate pairs of genes interact to produce the phenotype in such a way that neither of the dominant genes is expressive unless the other one is present. Complementary gene interaction shows 9 : 7 ratio in F_2-generation.

(iii) **In supplementary genes** interaction, two non-allelic gene pairs control the phenotypic expression of a trait but the dominant gene of one pair expresses itself only when the dominant gene of the other pair is present, whereas the dominant gene of the other pair is expressed even in the absence of the dominant gene of the first pair.

Interaction of supplementary genes is found in flower colour of **snapdragon** (*Antirrhinum majus*), which shows 9 : 3 : 4 ratio in F_2-generation.

(iv) **Duplicate genes** are two pairs of genes which are able to produce the same effect individually as well as in combination. **GS Shull** reported interaction of duplicate genes in *Capsella bursa pastoris* (Shepherd's purse),

Which shows triangular and top-shaped capsule in 15 : 1 ratio in F_2-generation. The possible genotype can give large number of phenotypes, such event is known as continuous variation.

Chromosomal Theory of Inheritance

It was proposed by **Walter Sutton** and **Theodor Boveri** in 1902. They made a correlation between Mendel's conclusions about genes (inherited traits) and the behaviour of chromosomes during mitosis and meiosis.

Chromosomal theory of inheritance described that

- Chromosomes are in pairs and genes or their alleles are located on chromosomes.
- Homologous chromosomes separate during meiosis so that alleles are segregated.
- Meiotic products have one of each homologous chromosome but not both.
- Fertilisation restores the pairs of chromosomes.

Linkage

- It is the tendency of alleles of different genes to be passed together from one generation to next.
- If the chromosomes in a gamete producing cell are not rearranged (or lack recombination) during meiosis, it would be expected that all the genes on a particular chromosome would be transferred together into whichever gamete gets that particular chromosome.
- That is, the genes are linked together on their chromosomes.
- Linkage was discovered by **W Bateson, ER Saunders** and **RC Punnett** (1905), when they were working with sweet pea *(Lathyrus odoratus)*.
- They found test cross ratio 7 : 1 : 1 : 7 instead of 1 : 1 : 1 : 1 and described it as coupling and repulsion.
- Linkage is an exception to the law of independent assortment.
- It is inversely proportional to the distance between the genes present on chromosomes.
- **TH Morgan** (1910) reported linkage in *Drosophila* and gave the term 'linkage' to this phenomenon.
  He also reported that coupling and repulsion are two aspects of the linkage.
- A **linkage group** is a linearly arranged group of linked genes, i.e., all the linked genes of a chromosome form a linkage group.
- The number of linkage group corresponds to the haploid number of chromosomes.

Linkage can be of two types

(i) **Complete linkage** is seen when only the parental combinations are produced. Crossing over remains absent in this case, e.g. male *Drosophila*, female silkworm (*Bombyx mori*).

(ii) **Incomplete linkage** is seen when some recombinations are also produced along with parental combinations in the offsprings.

Linkage Map

- **Linkage maps** or **genetic maps** are the graphical representation of relative distances between linked genes of a chromosome or a linkage group.
- In linkage map, one map unit is equal to 1% recombination.
- The map unit is also called **Centi Morgan** after the name of **Thomas H Morgan.**
- The first chromosome maps were prepared by **Sturtevent** in 1911.

Crossing Over

- The term' crossing over' was introduced by **Morgan** and **Cattell** for separation of linked genes. It is a process occurring in the prophase-I of meiosis, in which exchange of non-sister chromatids called **genetic recombination** takes place.
- Recombination involves the reshuffling of parental genes at their linkages so as to produce new genotypes. It develops due to crossing over, random fusion of gametes and independent assortment of chromosomes during meiosis.
- The frequency of Crossing Over Value (COV) is calculated using formula

$$\frac{\text{Total number of recombination}}{\text{Total number of progeny}} \times 100$$

- The significance of crossing over are
  - It provides proof for linear arrangement of genes on chromosomes.
  - It is the basis for linkage maps or genetic maps construction.
  - Recombinations or new gene combinations are produced due to the crossing over, which is a source of variations.

Sex-Determination

It is the method by which the distiction between male and female is established in a species. The chromosomes involved in sex-determination are called **sex chromosomes**. All other chromosomes are called **autosomal** chromosomes or **autosomes.** Although sex chromosomes provide the most common means of sex-determination. However, it is not the only mechanism to determine sex, such as

- In bees, males are haploid (n), while females are diploid ($2n$).
- Sex may be determined by a single allele or multiple alleles as in some wasps.
- The environmental factors also determine sex in some turtles, as these have indeterminate genetic sex-determining mechanisms.

There are many types of chromosomal mechanisms as follows

(i) **In humans, XX-XY system** is seen in which females are homomorphic XX and males are heteromorphic XY.

This is found in mammals including humans and some insects including *Drosophila*.

(ii) **In birds, ZW-ZZ system** is seen where females are heteromorphic ZW and males are homomorphic ZZ.

It occurs in birds, some fishes and moths. It is essentially the opposite of XY in mammals.

(iii) **In insects, XX-XO system** is seen, in which the females have two X-chromosomes.

Males have only one X and no additional sex chromosome, e.g. true bugs, grasshopper, etc.

(iv) **ZO-ZZ type** of sex-determination occurs in some butterflies and moths.

It is exactly opposite to the XX-XO type of sex-determination. Here, the females have odd sex chromosome (AA + Z), while males have two homomorphic sex chromosomes (AA + ZZ).

(v) **Compound chromosome system** is very complex due to multiple numbers of X and Y-chromosomes, e.g. in *Ascaris incurva,* a nematode, there are 26 autosomes, eight X-chromosomes and one Y-chromosome.

Males have 26A + 8X + Y for 35 chromosomes. Females have 26A + 16X for 42 chromosomes. This type of system is also common in spiders.

(vi) **Haplo-diploidy mechanism** or **male haploidy** or **arrhenotoky** is common in hymenopteran insects, e.g. honeybees, wasps, ants, etc.

In these insects, three types of individuals are seen

(a) **Diploid queen** are fully functional females developed from fertilised eggs.

(b) **Diploid workers** are sterile females developed from fertilised eggs.

(c) **Haploid males or drones** are functional males, developed parthenogenetically from haploid eggs.

(vii) **Genic balance mechanism** helps to determine the sex of organism by calculating the ratio between X-chromosomes and autosomes, e.g. *Drosophila*.

Different Phenotypes, Chromosomal Complement and X/A Ratio in *Drosophila*

Phenotype	Chromosomal Complement	X/Autosomal Set
Normal female	XX + 2*n* autosomes	1.00
Normal male	XY + 2*n* autosomes	0.50
Metafemale	XXX + 2*n* autosomes	1.50
Metamale	X + 3*n* autosomes	0.33
Intersex	XX + 3*n* autosomes	0.67

NOTE **Gynandromoprhs** or **sex mosaic** are flies with one half having male characteristics and another half having female characteristics.

Sex-Linked Inheritance

The characters or traits carried on sex chromosomes are called sex-linked traits and their inheritance is called sex-linked inheritance.

- First of all **TH Morgan** (1910) observed sex-linked inheritance in *Drosophila* (fruitfly).
- In humans, colour blindness (red-green blindness) and haemophilia (bleeder's disease) are important sex-linked (X-linked) diseases.
- The genes located exclusively on X-chromosomes are called **X-linked** and the genes that occur only on Y-chromosome are called **holandric genes**.
- The genes present on homologous regions in sex chromosomes are called **XY-linked genes**.

The various types of sex-linked inheritance are given below

1. **X-linked inheritance** is carried out by X-linked genes which are represented twice in females.
   - The recessive X-linked genes show *criss–cross* inheritance (**digynic** or **diandric**).
   - Males are more affected than females because there is no homologue of this gene on Y-chromosome.
   - Females are affected only in heterozygous condition for a dominant trait, while for a recessive trait they are affected in homozygous condition.
2. **In X-Y linked inheritance**, genes occur on homologous sections of X and Y chromosomes so that they are inherited like autosomal genes, e.g. nephritis, xeroderma pigmentosum.
3. **In holandric or Y-linked inheritance**, genes are located on non-homologous segment of Y-chromosome, whose alleles do not occur on X-chromosome. Such characters are transferred from father to son, e.g. hypertrichosis of ears in man.
4. **Sex limited traits** develop only in one sex.
   - They are produced and controlled by genes which may be located on autosomes in only one sex.
   - These are inherited according to Mendel's law, e.g. moustaches in males, breast in females, brilliant plumage in peacock, etc.
5. **Sex influenced traits** are the autosomal traits in which the dominant expression depends on the sex hormones of the individual, e.g. baldness in males, length of index finger, etc.

Mutation

It is a phenomenon, which results in alteration of DNA sequences and consequently results in changes in the genotype and the phenotype of an organism. This phenomenon also leads to variation in DNA.

Depending upon the cause, mutations are of two types

1. **Gene mutations** or **Point mutations** are changes in gene structure due to alteration either in nucleotide number, its type or the sequence. They are of following types
   - **Tautomerism** is the changed pairing of purine with purine and pyrimidine with pyrimidine. Tautomers are alternate forms of bases and are produced due to the rearrangement.
   - **Substitution** involves the change in one or more nitrogenous base pair with other.

     It is further of two types
     - **Transition** in which a purine or a pyrimidine in a triplet code of DNA is replaced by its type, e.g. GC $\rightarrow$ AT or AT $\rightarrow$ GC
     - **Transversions** are mutation in which a purine is replaced by pyrimidine or *vice-versa*, e.g.

       GC $\rightarrow$ CG or TA, AT $\rightarrow$ TA or CG
   - **Frameshift mutation** involves addition or deletion of single nitrogenous base which results in shift of reading frame in forward or backward direction.
2. **Chromosomal mutations** are caused due to the change in arrangement of genes or number of chromosomes (genomatic mutations).

   (i) Morphological aberrations in chromosomes include
   - **Deletion** or **deficiency**, here a segment of chromosome gets lost. In deletion, intercalary segment is lost whereas in deficiency, a terminal segment is lost. e.g. deletion of short arm of chromosome 4 causes Wolf-Hirschhorn's syndrome.
   - **Duplication,** here the deleted chromosomal segment gets attached/inserted to its normal homologous chromosome.
   - **Inversion,** here which a piece of chromosome is removed and rejoined in reverse order.
   - **Translocation**, here the mutual exchange of chromosome segments between non-homologous chromosome occurs. It helps a gene to move from one linkage group to another.

   (ii) Genomatic mutations include
   - **Euploidy**, here an additional set of chromosomes is present. It can be monoploidy or haploidy and polyploidy. Polyploidy is further of three types
     - **Autopolyploidy** leads to numerical increase of some genome. It induces gigas effect, e.g. maize, rice.
     - **Allopolyploidy** develops through hybridisation between two species followed by doubling of chromosomes. It produces new species, e.g. *Triticale*.
     - **Autoallopolyploidy**, here the genome is in more than one diploid state, e.g. *Helianthus*.
   - **Aneuploidy** involves loss (hypoploidy) or addition (hyperploidy) of one or more chromosomes, e.g. monosomy ($2n-1$), nullisomy ($2n-2$), trisomy ($2n+1$), etc.

Genetic Disorders

These can be divided into two main categories

1. Mendelian Disorders

These occur mainly due to the alteration or mutation in a single gene.

- These disorders may be dominant or recessive.
- These are transmitted from one generation to the next and follows Mendel's principles of heredity.
- Examples of Mendelian disorders are haemophilia, colour blindness, sickle-cell anaemia, thalassemia, phenylketonuria, etc.

(i) **Haemophilia** (Bleeder's disease) It is **X-linked recessive** disease, which was first studied by **John Cotto** in 1803. Haemophilia was common in royal families of Europe.
- Haemophilia-A is characterised by lack of antihaemophilic globulin (factor-VIII) protein.
- About 4/5 cases of haemophilia belong to haemophilia-A.
- Haemophilia-B or Christmas disease results from a defect in Plasma Thromboplastic Component (PTC or factor-IX).

(ii) **Colour blindness** It is a condition, in which certain colours (green and red) cannot be distinguished due to the lack of one or more colour absorbing pigments in the cone cells of retina.

In humans, the most common colour blindness is red-green colour blindness, which is a sex-linked (i.e. **X-linked recessive**) defect caused by a recessive gene and is thus more common in males than females.

(iii) **Sickle-cell anaemia** It is an **autosome-linked** recessive trait. It can be transmitted from parents to the offspring when both the partners are carrier or affected for the gene.
- This disease is characterised by sickle-shaped RBCs instead of normal ones.
- Due to the abnormal shape, haemoglobin carries very less content of O_2.
- This disease is controlled by a single pair of allele, Hb^A and Hb^s, out of the possible genotypes only homozygous individuals for Hb^s (Hb^sHb^s) show the diseased phenotype.
- Heterozygous (Hb^AHb^s) individuals appear apparently unaffected but they are carrier of the disease as there is 50% probability of transmission of the mutant gene to the progeny.

(iv) **Phenylketonuria** It is also an **autosomal recessive trait**.

- The affected individual lacks an enzyme that converts the amino acid phenylalanine into tyrosine.
- This phenylalanine gets accumulated and converted into phenyl pyruvic acid and other derivatives inside brain.
- These are excreted through urine because of its poor absorption by kidney.

(v) **Thalassemia** It is an inherited **autosomal recessive** blood disease. In thalassemia, the genetic defect results in reduced rate of synthesis of one of the globin chains that make up haemoglobin.

- Reduced synthesis of one of the globin chains can cause the formation of abnormal haemoglobin molecules, thus causing anaemia.
- There are two forms of thalassemia
  - **α-thalassemia** which is governed by genes on chromosomes 16, α-globin chain production is defected.
  - **β-thalassemia** which is governed by genes on chromosome 11, β- globin chain production is defected.

NOTE Polydactyly and Huntington's chorea diseases show autosomal dominant inheritance and hence, they appear in every generation.

2. Chromosomal Disorders

These are caused due to the absence or excess or abnormal arrangement of one or more chromosomes.

(i) **Down's syndrome** It is characterised by the presence of an additional copy of the chromosome number 21 (trisomy of 21 occurs). It is caused due to the **non-disjunction** (failure of chromosomes to separate) of chromosomes.

- The affected individual is short-statured with small round head, furrowed tongue and partially open mouth.
- Palm is broad with characteristic palmcrease. Physical, psychomotor and mental development is retarted.

(ii) **Klinefelter's Syndrome** It is also caused due to the presence of an additional copy of X-chromosome resulting into a karyotype of 47 (XXY).

Such individuals have overall masculine development, however, the feminine development (development of breast, i.e. gynaecomastia) is also expressed. Such individuals are sterile.

(iii) **Turner's Syndrome** It is caused due to the absence of one of the X-chromosomes, i.e. 45 with XO. These females are sterile as ovaries are rudimentary besides other features including lack of other secondary sexual characters.

(iv) **Cri-du-chat Syndrome** It is caused due to a deletion in the short arm of chromosome number 5.

The affected newborn cries in a high pitched sound like mewing of a cat.

NOTE Patau's syndrome occurs due to trisomy of 13 chromosome. Edward syndrome occurs due to trisomy of 18 chromosome.

DAY PRACTICE SESSION 1

FOUNDATION QUESTIONS EXERCISE

1 The allele which is unable to express its effect in the presence of another is called

(a) codominant (b) supplementary
(c) complementary (d) recessive

2 An allele is dominant, if it is expressed in

(a) both homozygous and heterozygous states
(b) second generation
(c) heterozygous combination
(d) homozygous combination

3 Phenotype of an organism is the result of

(a) mutations and linkages
(b) cytoplasmic effects and nutrition
(c) environmental changes and sexual dimorphism
(d) genotype and environment interactions

4 Mating of an organism to a double recessive in order to determine whether it is homozygous or heterozygous for a character under considerat

(a) reciprocal cross
(b) test cross
(c) dihybrid cross
(d) back cross

5 Select the correct statement. → NEET 2018

(a) Spliceosomes take part in translation
(b) Punnett square was developed by a British scientist
(c) Franklin Stahl coined the term 'linkage'
(d) Transduction was discovered by S Altman

6 Which one of the following symbols and its representation, used in human pedigree analysis is correct? → CBSE-AIPMT 2010

(a) □═○ = Mating between relatives
(b) ◯ = Unaffected male
(c) □ = Unaffected female
(d) ◆ = Male affected

7 Which one from those given below is the period of Mendel's hybridisation experiments? → NEET 2017

(a) 1856-1863 (b) 1840-1850
(c) 1857-1869 (d) 1870-1877

8 In his class experiments on pea plants, Mendel did not use → CBSE-AIPMT 2015

(a) seed colour (b) pod length
(c) seed shape (d) flower position

9 Among the following characters, which one was not considered by Mendel in his experiments on pea? → NEET 2017

(a) Stem–Tall or Dwarf
(b) Trichomes–Glandular or Non-glandular
(c) Seed–Green or Yellow
(d) Pod–Inflated or Constricted

10 A true breeding plant is → NEET-II 2016

(a) one that is able to breed on its own
(b) produced due to cross-pollination among unrelated plants
(c) near homozygous and produces offspring of its own kind
(d) always homozygous recessive in its genetic constitution

11 Mendel developed his basic principles of heredity by

(a) microscopic study of chromosomes and genes
(b) mathematical analysis of the offsprings of pea plants
(c) breeding experiments with *Drosophila*
(d) ultracentrifugation studies of cell organelles

12 Which one of the following cannot be explained on the basis of Mendel's law of dominance? → CBSE-AIPMT 2010

(a) The discrete unit controlling a particular character is called a factor
(b) Out of one pair of factors one is dominant and the other recessive
(c) Alleles do not show any blending and both the characters recover as such in F_2-generation
(d) Factors occur in pairs

13 When two heterozygous tall plants are crossed, some short plants appear in the offsprings. The appearance of these short plants illustrates

(a) segregation (b) intermediate inheritance
(c) crossing-over (d) codominant inheritance

14 A tall true breeding garden pea plant is crossed with a dwarf true breeding garden pea plant. When the F_1 plants were selfed, the resulting genotypes were in the ratio of → NEET-I 2016

(a) 1 : 2 : 1 : : Tall heterozygous : Tall homozygous : Dwarf
(b) 3 : 1 : : Tall : Dwarf
(c) 3 : 1 : : Dwarf : Tall
(d) 1 : 2 : 1 :: Tall homozygous : Tall heterozygous : Dwarf

15 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

(a) 25% will be tall with red fruit
(b) 50% will be tall with red fruit
(c) 75% will be tall with red fruit
(d) all of the offsprings will be tall with red fruit

16 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?

I. Only round seeds with yellow cotyledons
II. Only round seeds with green cotyledons
III. Only wrinkled seeds with yellow cotyledons
IV. Only wrinkled seeds with green cotyledons

(a) All except II (b) All except IV
(c) All except III (d) All of these

17 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?

(a) 50 : 50 (b) 9 : 1
(c) 1 : 3 (d) 3 : 1

18 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 this person?

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

19 If Mohan has 6 girls, the percentage of probability of 7th child to be girl is

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

20 How many different kinds of gametes will be produced by a plant having the genotype AABbCC?

(a) Three (b) Four
(c) Nine (d) Two

21 In guinea pigs, black fur (B) is dominant over white fur (b) and rough fur (R) is dominant over smooth fur (r). A cross between two guinea pigs hybrid for both traits (BbRr × BbRr) produces some offsprings that have rough, black fur and some that have smooth, black fur. The genotypes of these offsprings illustrate the genetic concept of

(a) intermediate inheritance
(b) multiple alleles
(c) independent assortment
(d) codominance

22 In racoons, a dark face mask is dominant over a bleached face mask. Several crosses were made between racoons that were heterozygous for dark face mask and racoons that were homozygous for bleached face mask. What percentage of the offsprings would be expected to have a dark face mask?
(a) 0% (b) 50% (c) 75% (d) 100%

23 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 → CBSE-AIPMT 2012
(a) codominance
(b) dihybrid cross
(c) monohybrid cross with complete dominance
(d) monohybrid cross with incomplete dominance

24 RR (red) *Antirrhinum* is crossed with WW (white) one. Offsprings RW are pink. This is an example of
(a) dominant–recessive (b) incomplete dominance
(c) hybrid (d) supplementary genes

25 A gene showing codominance has → CBSE-AIPMT 2015
(a) one allele dominant on the other
(b) alleles tightly linked on the same chromosome
(c) alleles that are recessive to each other
(d) both alleles independently expressed in the heterozygote

26 Which Mendelian idea is depicted by a cross in which the F_1-generation resembles both the parents? → NEET 2013
(a) Incomplete dominance (b) Law of dominance
(b) Inheritance of one gene (d) Codominance

27 Inheritance of ABO blood group shows
(a) polyploidy (b) incomplete dominance
(c) multiple allelism (d) polygeny

28 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 the presence of both 'A' and 'B' type proteins in 'AB' blood group individuals. This is an example of → NEET 2013
(a) codominance (b) incomplete dominance
(c) partial dominance (d) complete dominance

29 Which of the following characteristics represents 'inheritance of blood groups' in humans? → NEET 2018
1. Dominance 2. Codominance
3. Multiple allele 4. Incomplete dominance
5. Polygenic inheritance
(a) 2, 4 and 5 (b) 1, 2 and 3
(c) 2, 3 and 5 (d) 1, 3 and 5

30 In the ABO system of blood groups, if both antigens are present but no antibody, the blood group of the individual would be
(a) B (b) O (c) AB (d) A

31 A person with blood group A has
(a) antigen A and antibody b (b) antigen B and antibody a
(c) Both antibodies (d) no antibody and no antigen

32 The genotypes of a husband and wife are I^A $I^B$and I^Ai. Among the blood types of their children, how many different genotypes and pehnotypes are possible? → NEET 2017
(a) 3 genotypes, 2 phenotypes
(b) 3 genotypes, 4 phenotypes
(c) 4 genotypes, 3 phenotypes
(d) 4 genotypes, 4 phenotypes

33 A pleiotropic gene → CBSE-AIPMT 2015
(a) is expressed only in primitive plants
(b) is a gene evolved during Pliocene
(c) controls a trait only in combination with another gene
(d) controls multiple traits in an individual

34 Inheritance of skin colour in humans is an example of
(a) chromosomal aberration (b) point mutation
(c) polygenic inheritance (d) codominance

35 Which one of the following pairs of features is a good example of polygenic inheritance?
(a) Human height and skin colour
(b) ABO blood group in humans and flower colour of *Mirabilis jalapa*
(c) Hair pigment of mouse and tongue rolling in humans
(d) Human eye colour and sickle-cell anaemia

36 Match the following columns.

Column I	Column II
A. Dominance	1. Many genes govern a single character
B. Codominance	2. In a heterozygous organism only one allele expresses itself
C. Pleiotropy	3. In a heterozygous organism both alleles express themselves fully
D. Polygenic inheritance	4. A single gene influences many characters

→ NEET-I 2016

Codes

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

37 Which one of the following pairs is incorrectly matched? → NEET 2018
(a) XO type sex-determination – Grasshopper
(b) ABO blood grouping – Codominance
(c) Starch synthesis in pea– Multiple alleles
(d) TH Morgan – Linkage

38 Interaction of epistatic genes shows the ratio
(a) 9 : 6 : 1 (b) 13 : 3
(c) 12 : 3 : 1 (d) 15 : 1

39 Fruit colour in squash is an example of → CBSE-AIPMT 2014
(a) recessive epistasis
(b) dominant epistasis
(c) complementary genes
(d) inhibitory genes

40 The term 'linkage' was coined by → CBSE-AIPMT 2015
(a) TH Morgan (b) T Boveri
(c) G Mendel (d) W Sutton

41 In a test cross involving F_1 dihybrid flies, more parental-type offsprings were produced than the recombinant type offsprings. This indicates → NEET-I 2016
(a) chromosomes failed to separate during meiosis
(b) the two genes are linked and present on the same chromosome
(c) both of the characters are controlled by more than one gene
(d) the two genes are located on two different chromosomes

42 Which one of the following statements is not true for two genes that show 50% recombination frequency? → NEET 2013
(a) The genes may be on different chromosomes
(b) The genes are tightly linked
(c) The genes show independent assortment
(d) If the genes are present on the same chromosome, they undergo more than one crossovers in every meiosis

43 Two genes situated very close on the chromosome show
(a) hardly any crossing over
(b) high crossing over
(c) no crossing over
(d) only double crossing over

44 The number of linkage groups in *Drosophila, Pisum,* corn and mice are
(a) 4, 7, 10 and 19, respectively
(b) 4, 7, 19 and 20, respectively
(c) 4, 7, 10 and 20, respectively
(d) 10, 12, 17 and 20, respectively

45 In a dihybrid cross, two recessive genes showed 10% recombinants. The distance between two genes is
(a) 20 map units
(b) 10 map units
(c) 30 map units
(d) 40 map units

46 Crossing over occurs between
(a) sister chromatids
(b) non-sister chromatids
(c) non-homologous chromosomes
(d) All of the above

47 Crossing over is the exchange of parts of chromosomes between two non-sister chromatids. Crossing over in diploid organism is responsible for
(a) linkage between genes (b) recombination of genes
(c) segregation of alleles (d) dominance of genes

48 ZZ/ZW type of sex-determination is seen in
(a) platypus (b) snails (c) cockroach (d) peacock

49 What type of sex-determination is seen in butterflies?
(a) XX-XY type (b) ZO-ZZ type
(c) XX-XO type (d) ZW-ZZ type

50 Which one of the following conditions correctly describes the manner of determining the sex in the given example? → CBSE-AIPMT 2014
(a) XO type of sex chromosomes determine male sex in grasshopper
(b) XO condition in humans as found in Turner's syndrome, determines female sex
(c) Homozygous sex chromosomes (XX) produce male in *Drosophila*
(d) Homozygous sex chromosomes (ZZ) determine female sex in birds

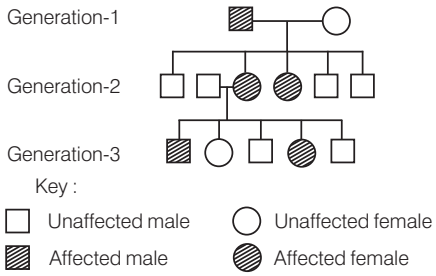
51 Man is hemizygous for
(a) X-chromosome (b) A-chromosome
(c) B-chromosome (d) E-chromosome

52 Individuals with patches of other sex are called
(a) gynandromorph (b) androgynous
(d) andromorphs (d) gynomorph

53 Sex is determined in human beings
(a) by ovum
(b) at the time of fertilisation
(c) 40 days after fertilisation
(d) 7th-8th week when genitals differentiate in foetus

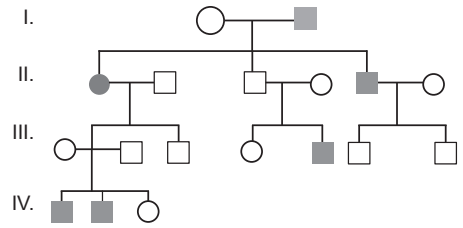
54 The recessive genes located on X-chromosome in humans are always
(a) lethal (b) sublethal
(c) expressed in males (d) expressed in females

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



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

56 In the following human pedigree, the filled symbols represent the affected individuals. Identify the type of given pedigree. ➞CBSE-AIPMT 2015



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

57 Genes located on Y-chromosomes are called
(a) holandric genes (b) autosomal genes
(c) sex-linked genes (d) mutant genes

58 A woman has an X-linked condition on one of her X-chromosomes. This chromosome can be inherited by ➞NEET 2018
(a) only grandchildren (b) only sons
(c) only daughter (d) Both (b) and (c)

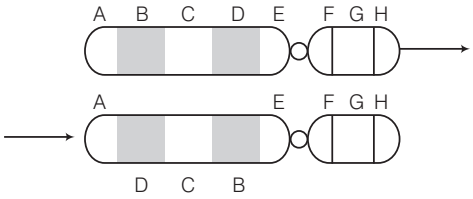
59 Point mutation involves
(a) insertion (b) change in single base pair
(c) duplication (d) deletion

60 If the sequence of genes on a chromosome is changed from ABCDEFG to ABCDFEG, then it is
(a) addition (b) deletion (c) substitution (d) inversion

61 In a mutational event, when adenine is replaced by guanine, it is the case of
(a) frameshift mutation (b) transcription
(c) transition (d) transversion

62 The mechanism that causes a gene to move from one linkage group to another is called ➞NEET-II 2016
(a) inversion (b) duplication
(c) translocation (d) crossing over

63 Given below is a representation of a kind of chromosomal mutation. What is the kind of mutation represented?



(a) Deletion (b) Duplication
(c) Inversion (d) Reciprocal translocation

64 A cell at telophase stage is observed by a student in a plant brought from the field. He tells his teacher that this cell is not like other cells at telophase stage. There is no formation of cell plate and thus the cell is containing more number of chromosomes as compared to other dividing cells. This would result in ➞NEET-I 2016
(a) polyploidy (b) somaclonal variation
(c) polyteny (d) aneuploidy

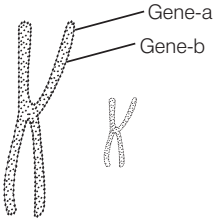
65 The cri-du-chat syndrome is caused by change in chromosome structure involving
(a) deletion (b) duplication
(c) inversion (d) translocation

66 The incorrect statement with regard to haemophilia is ➞NEET 2013
(a) it is a sex-linked disease
(b) it is a recessive disease
(c) it is a dominant disease
(d) a single protein involved in the clotting of blood is affected

67 Haemophilia is more commonly seen in human males than in human females because
(a) this disease is due to an X-linked dominant mutation
(b) a greater proportion of girls die in infancy
(c) this disease is due to an X-linked recessive mutation
(d) this disease is due to a Y-linked recessive mutation

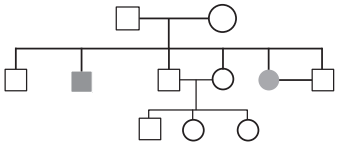
68 Which of the following most appropriately describes haemophilia? ➞NEET-I 2016
(a) X-linked recessive gene disorder
(b) Chromosomal disorder
(c) Dominant gene disorder
(d) Recessive gene disorder

69 Given below is a highly simplified representation of the human sex chromosomes from a karyotype. The genes '*a*' and '*b*' could be of



(a) colour blindness and body height
(b) attached earlobe and Rhesus blood group
(c) haemophilia and red-green colour blindness
(d) phenylketonuria and haemophilia

70 Study the pedigree chart given below.



What does it show?
(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

71 A colourblind 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 colourblind? → CBSE-AIPMT 2015

(a) 0.5 (b) 1
(c) Nil (d) 0.25

72 If a colourblind man marries a woman who is homozygous for normal colour vision, the probability of their son being colourblind is → NEET-II 2016

(a) 0 (b) 0.5
(c) 0.75 (d) 1

73 A man whose father was colourblind marries a woman, who had a colourblind mother and normal father. What percentage of male children of this couple will be colourblind? → CBSE-AIPMT 2014

(a) 25% (b) 0%
(c) 50% (d) 75%

74 If a colourblind woman marries a normal visioned man, their sons will be

(a) all normal visioned
(b) one-half colourblind and one-half normal
(c) three-fourth colourblind and one-fourth normal
(d) all colourblind

75 Number of chromosomes in Down's syndrome is

(a) 46 (b) 47 (c) 48 (d) 49

76 A disease caused by an autosomal primary non-disjunction is → NEET 2017

(a) Down's syndrome (b) Klinefelter's syndrome
(c) Turner's syndrome (d) Sickle-cell anaemia

77 A woman with 47 chromosomes due to three copies of chromosome 21 is characterised by

(a) Down's syndrome (b) Triploidy
(c) Turner's syndrome (d) Super femaleness

78 Number of X-chromosomes in Turner's syndrome is

(a) 3 (b) 2
(c) 1 (d) zero

79 A human female with Turner's syndrome → CBSE-AIPMT 2014

(a) has 45 chromosomes with XO
(b) has one additional X-chromosome
(c) exhibits male characters
(d) is able to produce children with normal husband

80. If both parents are carriers for thalassemia, which is an autosomal recessive disorder, what are the chances of pregnancy resulting in an affected child? → NEET 2013

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

81. Thalassemia and sickle-cell anaemia are caused due to a problem in globin molecule synthesis. Select the correct statement. → NEET 2017

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

82 Pick out the correct statement. → NEET-I 2016

I. Haemophilia is a sex-linked recessive disease.
II. Down's syndrome is due to aneuploidy.
III. Phenylketonuria is an autosomal recessive gene disorder.
IV. Sickle-cell anaemia is an X-linked recessive gene disorder.

(a) II and IV
(b) I, III and IV
(c) I, II and III
(d) I and IV

Directions (Q. Nos. 83 and 84) *In each of the following questions a statement of Assertion is given followed by a corresponding statement of Reason just below it. Of the statements, mark the correct answer as*

(a) If both Assertion and Reason are true and Reason is the correct explanation of Assertion
(b) If both Assertion and Reason are true, but Reason is not the correct explanation of Assertion
(c) If Assertion is true, but Reason is false
(d) If both Assertion and Reason are false

83. **Assertion** The duplicate genes are also called pseudoalleles.
Reason Duplicate genes although present on different locus but have the ability to produce same or almost same trait of a character.

84. **Assertion** In humans, female sex is determined by XX-chromosomes.
Reason Male sex is determined by XY-chromosomes.

DAY PRACTICE SESSION 2

PROGRESSIVE QUESTIONS EXERCISE

1 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
(a) aaBB (b) AaBb
(c) AABB (d) aabb

2 Lack of independent assortment of two genes *A* and *B* in fruitfly (*Drosophila)* is due to the
(a) repulsion (b) recombination
(c) linkage (d) crossing over

3 A normal woman whose father was colourblind is married to a normal man. The sons would be
(a) 75% colourblind (b) 50% colourblind
(c) all normal (d) all colourblind

4 Which cross would best illustrate Mendel's law of segregation?
(a) TT × tt (b) Hh × hh
(c) Bb × Bb (d) rr × rr

5 The contrasting pairs of factors in Mendelian crosses are called
(a) multiple alleles (b) allelomorphs
(c) alloloci (d) paramorphs

6 A man has blood group A. His wife has blood group B. Their children can have blood group (s)
(a) AB (b) A or B
(c) A or B or AB (d) A or B or AB or O

7 How many different types of gametes can be formed by F_1 progeny, resulting from the following cross?
AA BB CC × aa bb cc
(a) 3 (b) 8
(c) 27 (d) 64

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

9 Which of the following gametes are produced by individual with genotype AaBb?
(a) Aa, Bb
(b) AB,ab
(c) AB, Ab, aB, ab
(d) AB, Aa, Bb, abg

10 Which one of the following statements is correct?
(a) There will be no independent assortment in the presence of linkage
(b) There will be no segregation in the presence of linkage
(c) There will be no dominance in the presence of linkage
(d) There will be no crossing over in the presence of dominance

11 A common test to find the genotype of a hybrid is by
(a) crossing of one F_2 progeny with male parent
(b) crossing of one F_2 progeny with female parent
(c) studying the sexual behaviour of F_1 progenies
(d) crossing of one F_1 progeny with recessive parent

12 In a chromosome map, *B* and *C* give crossing over 3% and *A* and *B* 8%. What will be the percentage cross over between *A* and *C*?

$A \xrightarrow{8\%} B$, $B \xrightarrow{3\%} C$

$A \xrightarrow{?} C$

(a) 12% (b) 24%
(c) 7% (d) 11%

13 *Drosophila* flies with XXY genotype are females but human beings with such genotype are abnormal males. It shows that
(a) Y-chromosome is essential for sex-determination in *Drosophila*
(b) Y-chromosome is female determining in *Drosophila*
(c) Y-chromosome is male determining in human beings
(d) Y-chromosome has no role in sex-determination either in *Drosophila* or in human beings

14 Select the incorrect statement from the following
(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

15 Two genes R and Y are located very close on the chromosomal linkage map of maize plant. When RRYY and rryy genotypes are hybridised, then F_2-segregation will show
(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

16 ABO blood groups in humans are controlled by the gene I. It has three alleles I^A, I^B and i. Since, there are three different alleles, six different genotypes are possible. How many phenotypes can occur?

(a) Three (b) One
(c) Four (d) Two

17 In pea plants, the long-stem trait (L) is dominant and the short-stem trait is recessive. Two pea plants were crossed, producing seeds that yielded 165 long-stem plants and 54 short-stem plants. The genotypes of the parent plants were most likely

(a) Ll and LL (b) Ll and Ll
(c) ll and ll (d) LL and ll

18 Down's syndrome is caused by an extra copy of chromosome number 21. What percentage of offsprings produced by an affected mother and a normal father would be affected by this disorder?

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

19 In a given plant, red colour (R) of fruit is dominant over white fruit (r) and tallness (T) is dominant over dwarfness (t). If a plant with genotype RRTt, is crossed with plant of genotype rrtt, what will be the percentage of tall plants with red fruits in the next generation?

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

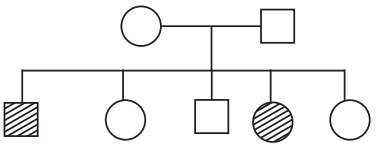
20 A normal visioned man whose father was colourblind, marries a woman whose father was also colourblind. They have their first child as a daughter. What are the chances that this child would be colourblind? → CBSE-AIPMT 2012

(a) 100% (b) 0% (c) 25% (d) 50%

21 The mating of two curly-haired brown guinea pigs results in some offsprings with brown curly hair, some with brown straight hair, some with white curly hair and even some with white straight hair. This mating illustrates which of Mendel's laws?

(a) Dominance
(b) Segregation
(c) Independent assortment
(d) Sex-linkage

22 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?



(a) The parents are homozygous recessive
(b) The trait is Y-linked
(c) The parents are homozygous dominant
(d) The parents are heterozygous

23 Select the correct statement from the ones given below with respect to dihybrid cross.

(a) Tightly linked genes on the same chromosome show higher recombinations
(b) Genes far apart on the same chromosome shown very few recombinations
(c) Genes loosely linked on the same chromosome show similar recombinations as the tightly linked ones
(d) Tightly linked genes on the same chromosome show very few recombinations

24 Match the following columns.

	Column I		Column II
A.	Multiple alleles	1.	Interaction between different genes
B.	Epistasis	2.	X-linked disease
C.	Crossing over	3.	Autosomal disease
D.	Colour blindness	4.	The presence of more than two alleles for a gene
		5.	Pachytene stage

Codes

	A	B	C	D		A	B	C	D
(a)	1	2	3	4	(b)	4	1	5	2
(c)	2	1	4	5	(d)	5	4	1	2

25 **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.

(a) If both Assertion and Reason are true and Reason is the correct explanation of Assertion
(b) If both Assertion and Reason are true, but Reason is not the correct explanation of Assertion
(c) If Assertion is true, but Reason is false
(d) If both Assertion and Reason are false

ANSWERS

SESSION 1

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

SESSION 2

1 (d)	**2** (c)	**3** (b)	**4** (c)	**5** (b)	**6** (d)	**7** (b)	**8** (a)	**9** (c)	**10** (a)
11 (d)	**12** (d)	**13** (c)	**14** (d)	**15** (d)	**16** (c)	**17** (b)	**18** (a)	**19** (c)	**20** (b)
21 (c)	**22** (d)	**23** (d)	**24** (b)	**25** (c)					