Chapter 5
PRINCIPLES OF INHERITANCE AND VARIATION

One mark questions:

1. What is genetics ?
   Study of heredity and variation OR study of gene.

2. What is an allele ?
   An alternate form of a gene for a character present on identical loci of two homologous chromosomes.

3. What is phenotype ?
   A physically expressed external character in an individual.

4. What is genotype ?
   Genetic make up of an individual for a phenotype.

5. What is dominance?
   A gene or a character expressed in heterozygous condition of an organism.

6. What is recessiveness ?
   A gene or a character which fails to express in heterozygous condition.

7. What is linkage ?
   Genes present on a chromosome remain together and inherited in a group.

Two mark questions

1. Differentiate homozygous individual from heterozygous individual.
   Homozygous – Individual having similar genes in allelic pair.
   Heterozygous- Individual having dissimilar genes in allelic pair.

2. What is test cross ? Mention its significance.

Cross between F1 hybrid and its recessive parent.
To know the genotype of F1.

3. What is pleotrophic gene ? Give an example.
   One gene influencing multiple characters in an individual.
   Eg., In human an autosomal recessive gene for phenylketoneuria, influences the characters like mental retardation, reduction in hair and skin colour.

4. Write any four abnormalities of Down syndrome
   - Short stature with round head
   - Furrowed tongue and partially open mouth
   - Palm is broad with characteristic palm crease
   - Physical, psychomotor and mental developments are retarded

5. Write the chromosomal complement and two abnormalities of Klinefelter's syndrome.

   Chromosomal complement - *44AA+ XXY*
   Abnormalities -
   - *Gynoecomastia* - Feminine characters like absence of facial hair and presence of enlarged breast.
   - Underdeveloped testis.
   - Absence of spermatogenesis.
   - Sterility.

6. Write the chromosomal complement and two abnormalities of Turner's syndrome.

   Chromosomal complement – 44AA + XO
   Abnormalities -
   - Short stature.
   - Breast and ovaries are poorly developed.
   - Delay or absence o puberty.
   - Incomplete or absence of oogenesis.
   - Loose skin or webbed neck.
   - Sterility.

7. Drosophila melanogaster is a model organism in genetic studies. Justify?
   - They could be grown on simple synthetic medium in the laboratory

- They complete their life cycle in about two weeks
- Single mating could produce a large number of progeny flies
- There is clear differentiation of sexes
- Hereditary variations can be seen in low power microscopes

8. What is polygenic inheritance? Give an example

An expression of a trait is controlled by three or more number of genes is called polygenic inheritance.

Eg: skin colour of humans

Three mark questions

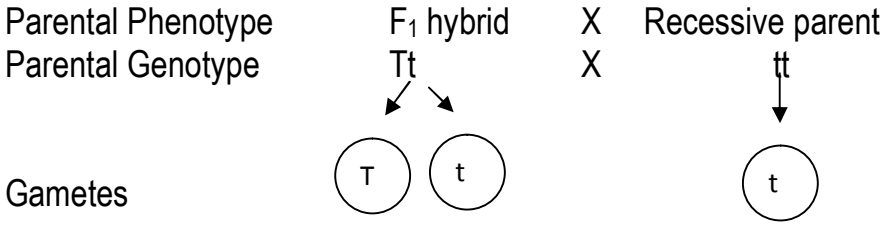
1. Write the comparison between the behavior of chromosomes and genes.

Chromosomes	Genes
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 segregates independently of another pair

for one gene inheritance. Write its significance.

Monohybrid Test Cross:

Crossing of F_1 monohybrid of unknown genotype with its recessive parent is called monohybrid test cross.



F_2 generation

	T	t
t	Tt Tall	tt Dwarf

Monohybrid Test cross Ratio 1 : 1

Significance:
Test Cross is conducted to know the genotype of F_1 hybrid .

3. a) In Drosophila melanogaster, the cross between
   i. yellow body, white eyed one with its wild type, the percentage of recombination is less.
   ii. white eyed, miniature winged one with its wild type, the percentage of recombination is high.
   Give reason for the above statements.
   b) Who studied the phenomenon of linkage in Drosophila?

a) i. because, the genes are tightly linked
   ii. because, the genes are placed part and crossing over has occurred
b) T.H.Morgan

4. Explain the experiment carried out by Morgan in Drosophila to demonstrate linkage. What is the observation made by him in that experiment?

Morgan hybridized yellow bodied, white eyed females to brown-bodied, red eyed male and intercrossed their F1 progeny. He observed that the two genes did not segregate independently of each other and the F2 ratio deviated very significantly from 9:3:3:1.

Morgan found that even when genes were grouped on the same chromosome, some genes were very tightly linked (showed very low recombination) while others were loosely linked (showed higher recombination).

5. What is codominance ? Explain with reference to human blood group.
It is the phenomenon of inheritance where both the alleles of heterozygote express themselves equally at a time.
Eg., In human the blood group AB having the alleles I^A and I^B express equally by producing both the sugars antigen A and antigen B.

6. What is polygenic inheritance ? Explain with an example.

Two or more genes influence a single character in a cumulative manner.

Eg., Human skin colour is regulated by three pairs of cumulative genes A,B and C.

AABBCC – dark skin (negro)

aabbcc – light skin (caucasoid)

AaBbCc – intermediate (mulattos)

7. What is haemophilia ? Mention the types.

   Inability of blood to clot or delay in clotting due to lack of clotting factors during bleeding.

   Types of haemophilia -

   Haemophilia A – deficiency of factor VIII.

   Haemophilia B – deficiency of factor IX.

   Haemophilia C – deficiency of factor XI

8. Briefly explain sickle cell anaemia.
   - Sickle cell anaemia is a condition of anaemia by the production of sickle cell RBCs.
   - Valine instead of glutamic acid in 6^{th} position by the gene Hb^S .
   - RBCs become sickle shape, do not carry sufficient O_2 and become stiff, crystaline and non-elastic.
   - Damage the capillaries of vital organs, internal bleeding and even death in severe condition.

9. Briefly explain phenylketon uria
   - Inborn error of phenyl alanine metabolism due to autosome linked recessive gene on chromosome 12.
   - Recessive gene pp fails to produce an enzyme phenyl alanine hydroxylase
   - No break down of phenyl alanine into tyrosine and its derivatives accumulates in the blood and CSF.
   - Mental retardation, light skin and hair and phenyalnine and its derivatives excreted through urine.

Five mark questions

1.a) Mention the features of Law of Dominance observed by Mendel .

I. Characters are uncontrolled by discrete unit called factors

II. Factors occur in pairs
III. In a dissimilar pair of factor one member of the pair dominates (dominant) the other (recessive).

b) Mention the possible genotypes of human A, B, AB and O blood groups

Blood types (Phenotypes)	**Genotypes**
A	I^AI^A / I^A i
AB	$I^A I^B$
B	I^BI^B/ I^Bi
O	i i

2. A red flowered snapdragon plant crossed with white flowered one produce a pink flowered plant, name and explain the inheritance pattern with schematic representation till the F2 generation.

Incomplete Dominance:-

A phenomenon where both the alleles of a character express incompletely producing a new intermediate phenotype in the heterozygous condition is called incomplete or partial dominance or blended inheritance.

Eg: Flower colour in Dog flower or snapdragon [Antirrhinum majus]

Explanation:-

Correns crossed homozygous red flowered plant **(RR)** with homozygous white flowered plant (**rr**), surprisingly in F_1 generation all hybrids were pink flowered plants (**Rr**). Because the dominant gene (R) fails to mask the recessive gene(r) completely.

When F_1 pink flowering plants were self crossed, the F_2 generation produce 25% red flowered plants, 50% pink flowered plants and 25% white flowered plants in 1:2:1 ratio.

Parental Phenotype: Red flowered plant X White flowered plant

Parental genotype: RR X rr

Gametes: R, r

F_1 hybrid Rr – Genotype of F_1 **All pink flowered plants**

Selfing F_1 X F_1

Genotype Rr X Rr

Gametes

F_2 generation

Gametes	**R**	**r**
R	RR Red	Rr Pink
r	Rr Pink	rr White

From the above checker board we can observe that both F_2 **phenotypic & genotypic ratio in incomplete dominance** are same

i.e., 1 : 2 : 1

Homozygous Heterozygous Homozygous

Red Pink White

The Appearance of red and white flowered plants in F_2 generation indicates 2 important features.

- The genes for Red & white colour did not mix.
- Genes segregated in F_2 indicates that there is no specific gene for pink colour.

3. What is a monohybrid cross? Explain the inheritance of one gene, taking height of plant as a trait in Pisum sativum. Work out the cross upto F2 generation.

It is a cross made between two individuals of a species, considering the inheritance of the contrasting pair of a single character/trait

OR

Cross between two individuals differing in a pair of contrasting characters

- To study the inheritance of one gene, Mendel crossed tall and dwarf Pea plants.
- He collected the seeds produced as a result of this cross and grew them to generate plants of the first hybrid generation. This is also called F1 generation
- Mendel observed that all the F1 progeny plants were tall, like one of its parents; none were dwarf

- Mendel then self pollinated the tall F1 plants and to his surprise found that in the F2 generation some of the offspring were dwarf; the character that was not seen in the F1 generation was now expressed
- The proportion of plants that were dwarf were ¼ of the F2 plants while ¾ of the plants were tall

Parents Tall x Dwarf
Genotype TT x tt
Gametes T t

F1 generation Tt
Selfing of F1 plants Tall x Tall
Genotype Tt x Tt
Gametes T t T t

	T	t
T	TT	Tt
t	Tt	tt

F2 generation

Phenotypic ratio- Tall: Dwarf – 3:1
Genotypic ratio- TT: Tt : tt - 1:2:1

4. Explain the method of sex determination in humans.

The sex determining mechanism in case of humans is XY type. Out of 23 pairs of chromosomes present, 22 pairs are exactly same in both males and females; these are the autosomes.

A pair of X-chromosomes are present in the female, whereas the presence of an X and Y chromosome are determinant of the male characteristic. During spermatogenesis among males, two types of gametes are produced. 50 per cent of the total sperm produced carry the X-chromosome and the rest 50 per cent has Y-chromosome besides the autosomes.

Females, however, produce only one type of ovum with an X-chromosome. There is an equal probability of fertilisation of the ovum with the sperm carrying either X or Y chromosome. In case the ovum fertilises with a sperm carrying X-chromosome the zygote develops into a female (XX) and the fertilisation of ovum with Y-chromosome carrying sperm results into a male offspring. …1

Or

	Male		Female	
	44+XY		44+ XX	
Gametes	22+X	22+Y	22+X	22+X

44+ XX — Male progeny

44+XY — Female progeny

5. Explain two gene inheritances with a schematic representation.

Mendel used dihybrid cross to answer the inheritance of two characters. First he established true breeding pea plants and selected two different contrast characters of seven traits. When he crossed dominant round and yellow seeded (RRYY) pea plant with recessive wrinkle and green seeded (rryy) pea plant.

Phenotype: pure round yellow seed pea plant X pure wrinkled green seed pea plant

Genotype : RRYY rryy

Gametes: RY ry

F1 generation RrYy (hybrid round yellow)

RrYy X RrYy (inbred or self pollinated)

F2 generation

Gametes RY Ry rY ry RY Ry rY ry

	RY	Ry	rY	ry
RY	RRYY	RRY	RrYY	RrYy
Ry	RRYy	RRyy	RrYy	Rryy
rY	RrYY	RrYy	rrYY	rrYy
ry	RrYy	Rryy	rrYy	rryy

result ; phenotypic = 9:3:3:1 genotypic = 1 : 2 : 2 : 4 : 1 : 2 : 1 : 2 : 1

6. . a) Distinguish between Mendelian disorder and chromosomal disorder.

b) By taking starch synthesis in pea seeds as an example, explain how a single gene product may produce more than one effect.

a) Mendelian disorders are mainly determined by alteration or mutation in the single gene.

Chromosomal disorders caused due to absence or excess or abnormal arrangement of one or more chromosome.

b) A single gene product may produce more than one effect.

For example, starch synthesis in pea seeds is controlled by one gene. It has two alleles (B and b). Starch is synthesized effectively by BB homozygotes and therefore, large starch grains are produced. In contrast, bb homozygotes have lesser efficiency in starch synthesis and produce smaller starch grains. After maturation of the seeds, BB seeds are round and the bb seeds are wrinkled. Heterozygotes produce round seeds, and so B seems to be the dominant allele. But, the starch grains produced are of intermediate size in Bb seeds.