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CLASS XII BIOLOGY NOTES

Key Notes and Important Questions with Answers

PRINCIPLES OF INHERITANCE AND VARIATION



Chapter - 5 Principles of Inheritance and Variation

Allele : Various or slightly different forms of a gene, having same position on the two homologous chromosomes.

Phenotype : The observable or external characteristics of an organism.

Genotype : The genetic constitution of an organism.

Monohybrid cross : A cross between two individuals of species, considering the inheritance of single pair of contrasting character, e.g. a cross between pure tall (TT) and Dwarf (tt)

Dihybrid cross : A cross between two individuals of a species, considering the inheritance of two pairs of contrasting traits/characters e.g., a cross between Round and Yellow (RRYY) and wrinkled and green (rryy) seed.

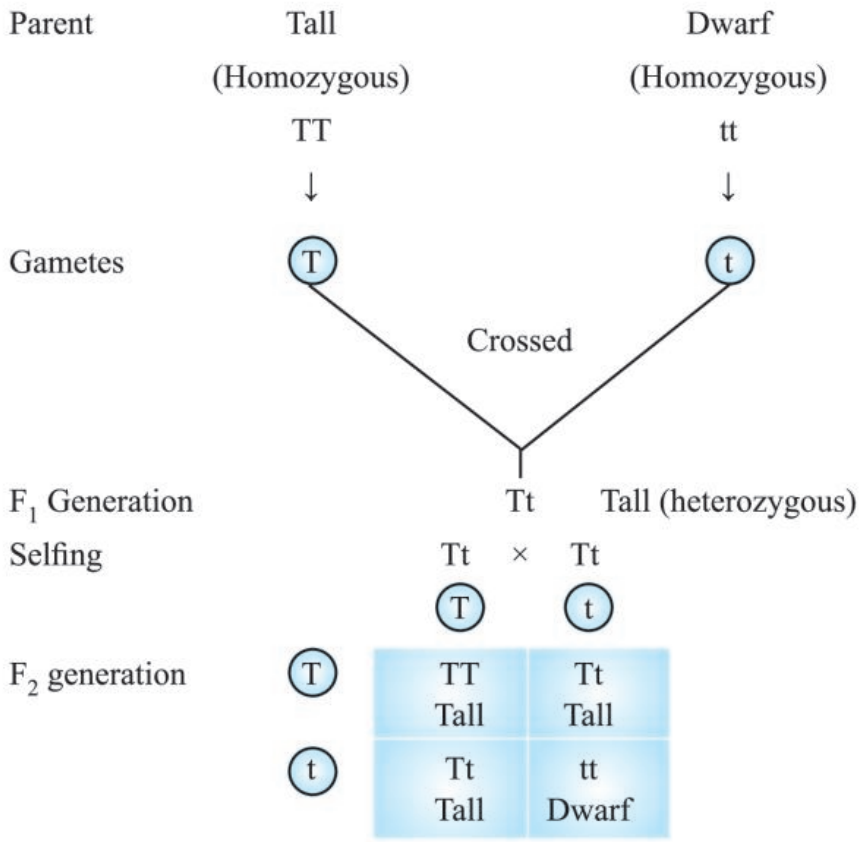
Aneuploidy : The phenomenon of gain or loss of one or more chromosome(s) that results due to failure of separation of homologous pair of chromosomes during meiosis.

Trisomy : The condition in which a particular chromosome is present in three copies in a diploid cell/nucleus.

Male heterogamety : When male produces two different types of gametes/ sperms e.g. In human beings X and Y.

Female Heterogamety : When female produces two different types of gametes/ova, e.g., female bird produces Z and W gametes.

Gregor Mendel Conducted controlled breeding experiment on garden pea (*Pisum sativum*) with a single trait. It is called monohybrid cross.



Tall homozygous (TT) – 25% Pure

Tall heterozygous (Tt) – 50% Hybrid.

Dwarf homozygous (tt) – 25% Pure.

Law of Dominance : When two individuals of a species differing in a pair of contrasting characters/traits are crossed, the trait that appears in the F_1 generation is dominant and the alternate form that remain hidden, is called recessive.

Law of Segregation (law of purity of gametes) : The members of allelic pair that remained together in the parent, segregate/separate during gamete formation and only one of the factors enters a gamete.

Law of Independent Assortment : In the inheritance of two pairs of contrasting characters (dihybrid cross) the factors of each pair of characters segregate independently of the factors of the other pair of characters.

Test Cross : When offspring or individual with dominant phenotype, whose genotype is not known, is crossed with an individual which is homozygous recessive for the trait, this cross is known as test cross.

Test cross is done to determine whether the individual parent exhibiting dominant traits is homozygous or heterozygous.

Flower colour is → Violet (Dominant phenotype, Genotype is unknown)

Genotype may be WW or Ww

Example :

Violet × White

WW ww homozygous recessive

Case 1

	(w)	(w)
(W)	Ww Violet	Ww Violet
(W)	Ww Violet	Ww Violet

Here, all flowers are violet

If all the offspring dominant trait, it indicate that individual under test is homozygous (WW) for dominant trait.

Case 2

Violet × White

Ww ww homozygous recessive

	(w)	(w)
(W)	Ww Violet	Ww Violet
(w)	ww White	ww White

50% flowers are violet

50% flowers are white

ratio Ww : ww

1 : 1

If a test cross gives 50% offspring showing dominant trait and 50% showing recessive traits, it indicates that individual parent under test is heterozygous (Ww) for dominant trait.

Incomplete dominance : It is the phenomenon where none of the two contrasting alleles is dominant but express themselves partially when present together in a hybrid and somewhat intermediate stage is seen.

Example : In plant snapdragon or *Antirrhinum* species, when plants with red flower (RR) are crossed with plant having white flower (rr) the hybrid F_1 plants (Rr) bear pink flower. When the F_1 hybrid with pink flower are self pollinated or crossed themselves, they produce red, pink and white flower in ratio of 1 : 2 : 1 in F_2 generation.

P Red-flowered plant × White flowered plant

RR rr

Gametes (R) (r)

F_1 generation Rr All pink flower

Pink × Pink

Rr Rr

	(R)	(r)
(R)	RR Red	Rr Pink
(r)	Rr Pink	rr White

F2 generation

Phonotypic ratio	Red	:	Pink	:	White
	1	:	2	:	1
	RR	:	Rr	:	rr
Genotypic ratio	1	:	2	:	1

In incomplete dominance, phenotypic ratio is equal to the genotypic ratio.

Multiple Allelism : It is a phenomenon in which a single character is governed by more than two alleles.

Example :

- ABO blood groups are controlled by gene I
- 'I' has three alleles—I^A, I^B, and i.

I^A and I^B alleles produce slightly different form of sugar present on plasma membrane of red blood cells.

- In allele 'i' do not produce any sugar.
- In any diploid individual only two alleles can be found. So multiple alleles can be detected only in a population.

Co-dominance : The alleles which do not show dominance recessive relationship and are able to express themselves independently when present together are called co-dominant alleles and this phenomenon is known as codominance. Example : Human blood groups.

There are 3 different alleles, 6 different genotypes control 4 different type of sugar Phenotypes :

Blood Group	Genotype	Types of Sugar
A	I^AI^A, I^Ai	A
B	I^BI^B, I^Bi	B
AB	I^AI^B,	both A & B
O	ii	No sugar alleles

In humans, blood group AB shows co-dominance as both the alleles I^A and I^B express themselves fully in presence of each other.

Chromosomal Theory of Inheritance : Proposed by Sutton and Boveri. The pairing and separation of a pair of chromosomes would lead to the segregation of a pair of factors they carried. They united the knowledge of segregation with Mendelian principles.

- **Linkage-** is the tendency of genes on a chromosome to remain together.
- Linked genes occur in the same chromosome.
- They lie in linear sequence in the chromosome - There is a tendency to maintain the parental combination of genes except for accessional choosers.
- Strength of linkage between genes is inversely proportional to the distance between the two.

Recombination : is the generation of non-parental gene combinations to the offsprings.Tightly linked genes show very low recombination frequency. Loosely linked genes show higher recombination frequency.

The frequency of recombination between gene pairs on the same chromosome is a measure of distance between genes and is used to map the position of genes on the chromosome.

Linkage and Recombination.

T. H. Morgan carried out several dihybrid crosses in *Drosophila melanogastor*. Two of them are given below :

Cross–I : Yellow-bodied and white eyed females crossed with brown-bodied, red eyed males (wild type)

- F_1 Progeny intercrossed and F_2 generation ratio deviated from 9 : 3 : 3 : 1 (two genes didn't segregate independently)
- The Parental combinations were 98.7% and recombinats were 1.3%

Conclusion : The two genes (body colour and eye-colour) are tightly linked ; results in less crossing over and less no. of non-parental progeny.

Cross-II : White bodied female with miniature wings and yellow-bodied male with normal wings (wild type) were crossed.

- The parental combinatios were 62.8% while the recombinants were 37.2%.

Conclusion : The two genes (body colour & wing's size) are loosely-linked ; results in more crossing over and more no. of non-parental progeny.

Chromosomal basis of sex Determination :

(i) XX-XY type - Female homogametic i.e. XX and male heterogametic i.e. XY in *Drosophila*, humans.

(ii) XX-XO type—All eggs bear additional X chromosome, Males have only one X chromosome besides autosomes whereas females have a pair of X chromosomes e.g., grasshoppers.

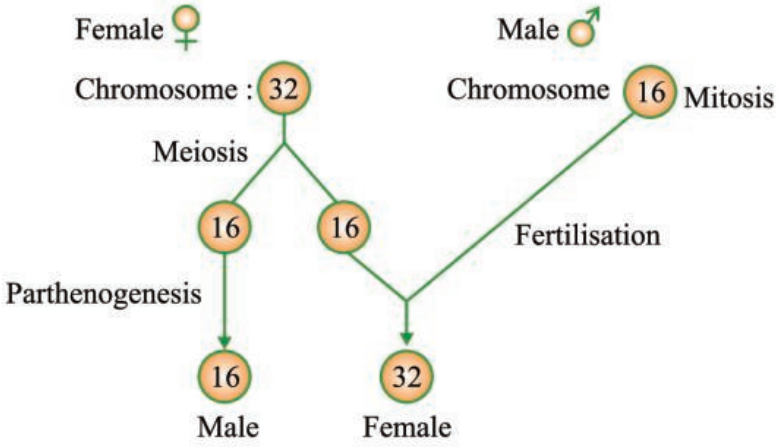
(iii) ZW-ZZ type—The females are heterogametic and have one Z and one W chromosome. The males are homogametic with a pair of Z chromosomes besides autosomes e.g., birds.

(iv) ZO-ZZ type—Females are heterogametic and produce 2 types of eggs - (A + Z) and (A + O). The males are homogametic with all the sperms having (A + Z) e.g. moths and butterflies.

here A = autosome

Sex determination in honey Bee : In Honey bee fertilized eggs develop into female (Queen or Worker) While unfertilized egg develops into male (drone)

by parthenogensis, the males have half no. of chromosomes than a female. The males are haploid (16-chromosomes), females are diploid (32-chromosomes).



There are three types of individuals :

1. Queen — diploid
   - developed from fertilized egg
   - functional female
2. Worker — diploid
   - developed from fertilized egg
   - non-functional female
3. Drone (male) — haploid
   - developed from unfertilized egg parthenogenetically
   - functional Male.

Pedigree Analysis

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

Usefulness of Pedigree Analysis

1. It is useful for genetic counsellors to advice intending couples about the possibility of having children with genetic defects like haemophilia, thalassemia etc.
2. It is helpful to study certain genetic trait and find out the possibility or absence or presence of that trait in homozygous or heterozygous condition in a particular individual.

Mendelian disorders

These are mainly determined by a alternation or mutation in single genes.

1. **Haemophilia :** Sex linked recessive disease which is transmitted from unaffected carriers female to male progeny. A single protein is affected which is a part of the cascade of proteins involved in the clotting of blood.

   $X^h Y$ = affected male

   $X^h X$ = carrier female

   The heterozygous female for haemophila may transmits the disease to her sons. The possibility of a female suffering from the disease is extremely rare (only when the mother of the female is a carrier is $X^h X$ and father is haemophilic i.e. $X^h Y$.

2. **Sickle-cell anaemia :** This is an autosome linked recessive trait. This defect is caused by substitution of glutamic acid by valine at the 6th position of the beta globin chain of the haemoglobin molecule. The mutant Hb molecule undergoes polymerisation under low oxygen tension which results change in shape of RBC from biconcave disc to elongated sickle like structure. The disease is controlled by a pair of allele, Hb^A and Hb^S

   $Hb^A Hb^A$. Normal $Hb^S Hb^S$ sufferer

   $Hb^A Hb^S$. Apparently unaffected/carriers

   Example Hb^AHb^S (Carrier) × $Hb^A Hb^A$ (Normal Man)

	Hb^A	Hb^A
Hb^A	Hb^AHb^A	Hb^AHb^A
Hb^S	Hb^AHb^S	Hb^AHb^S

Phenylketonuria : Inbron error of metabolism, autosomal recessive trait. Affected individual lacks an enzyme that converts amino acid phenylalanine into tyrosine. Phenylalanine is accumulated and converted into phenylpyruvic acid which accumulates in brain resulting in mental retardation.

Thalassemia : Thalassemia is autosome linked recessive disease. This disorder caused by defers in the synthesis of globin chain. Thalassemia is of two types—Alpha (α) Thalassemia, Beta (β) Thalassemia.

- In alpha Thalassemia production of alpha globin chain is affected. This Thalassemia is controlled by genes HBA1 and HBA2 located on chromosome 16th of each parent. Thalassemia occurs due to mutation or deletion of one or more of the four genes.
- In Beta Thalassemia production of (β-globin chain is affected, this thalassemia is controlled by gene HBB located on 11th chromosome of each parent. It occurs due to one or both HBB genes
- In Thalassemia very few globin is synthesized and is quantitative problem whereas in sickle cell anaemia there is a synthesis of incorrectly functioning globin and is a qualitative disorder.

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

Colour blindness : Colour blindness is sex-linked recessive trait in which a person fails to distinguish red and green colour. The gene for normal vision is dominant. The normal genes and its recessive alleles are carried by X-chromosome.

$X^C X^C$ — Colour blind female

$X X^C$ — Carrier female

$X^C Y$ — Colour blind male

Y Chromosome of male do not carry any gene for certain vision.

Inheritance Pattern in Colour Blindness

Father		Mother		Son		Daughter	
Pheno-type	**Geno-type**	**Pheno-type**	**Geno-type**	**Pheno-type**	**Geno-type**	**Pheno-type**	**Geno-type**
Normal	XY	Carrier	X^CX	Normal	XY	Normal	XX
				Colour-blind	X^CY	Carrier	X^CX
Normal	XY	Colour-blind	X^CX^C	Colour-blind	X^CY	Carrier	X^CX
Colour-blind	X^CY	Normal	XX	Normal	XY	Carrier	X^CX
Colour-blind	X^CY	Carrier	X^CX	Colour-blind	X^CY	Colour-blind	X^CX^C
				Normal	XY	Carrier	X^CX

Inheritance Pattern in Haemophilia

Father		Mother		Son		Daughter	
Pheno-type	Geno-type	Pheno-type	Geno-type	Pheno-type	Geno-type	Phen-type	Geno-type
Normal	XY	Haemophilic	X^hX^h	Haemophilic	X^hY	Carrier	X^hX
Normal	XY	Carrier	X^hX	Normal	XY	Normal	XX
				Haemophilic	X^hY	Carrier	X^hX
Haemop-hilic	X^hY	Carrier	X^hX	Normal	XY	Carrier	X^hX
				Haemophilic	X^hY	Haemop-hilic	X^hX^h
Haemop-hilic	X^hY	Normal	XX	Normal	XY	Carrier	X^hX

Chromosomal Disorder :

1. **Down's syndrome :** Trisomy of chromosomes number 21 (2n + 1) Affected individual is short statured with small round head, furrowed tongue, partially open month, broad palm. Physical, psychomotor and mental development is retarded.
2. **Klinefelter's syndrome :** extra copy of X chromosome ; karyotype XXY. Affected individual has overall masculine development with famine characters like gynaecomastia (development of breast) and is sterile

   44 autosomes + xxy = 47 chromosomes
3. **Turner's syndrome :** has absence of one X chromosome i.e. 45 with XO. Affected females are sterile with rudimentary ovaries and lack secondary sexual characters.

   44 autosomes + x = 45 chromosomes

Pleiotropy

The ability of a gene to have multiple phenotypic effects because it influences a number of characters simultaneously is known as pleiotropy. The gene having a multiple phenotypic effect because of its ability to control expression of a number of characters is called pleiotropic gene. E.g. in Garden Pea, the gene which controls the flower colour also controls the colour of seedcoat and presence of red spot in the leaf axis.

The disorder phenylketonuria shows pleiotropy.

Polygenic Inheritance

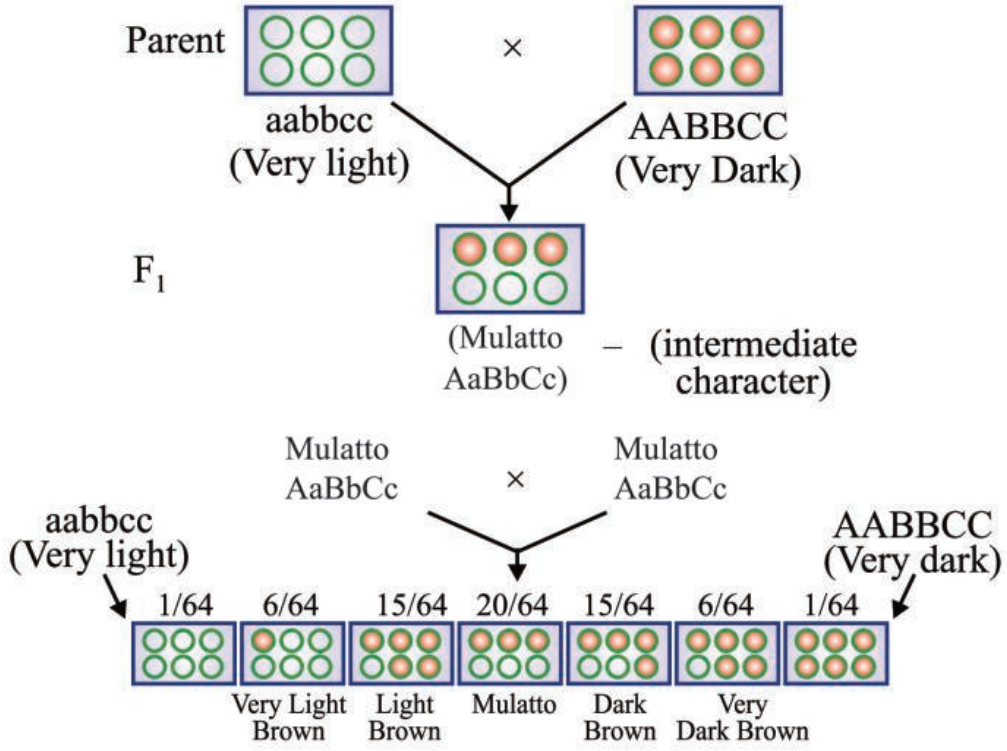
It is a type of inheritance controlled by three or more genes in which the dominant alleles have cumulative effect with each dominant allele expressing a part of the trait, the full being shown only when all the dominant alleles are present.

E.g., Kernel colour in wheat, skin colour in human beings, height in humans, cob length in maize etc.

In polygenic inheritance, a cross between two pure breeding parents produces an intermediate trait in F_1. In F_2 generation, apart from the two parental types, there are several intermediates (gradiations, show a bell shaped curve). F_1 hybrid form 8 kinds of gametes in each sex giving 64 combination in F_2 having 7 phenotypes.

Polygenic inheritance skin tone

3 loci : each has two possible alleles : Aa, Bb, Cc, each capital allele adds one unit of darkness, each lower case allele adds nothing. Parents produce F_1 offsprings with intermediate tone.



Offspring can have tone darker or lighter than either parent

Questions

VSA (I Mark)

1. Name the base change and the amino acid change, responsible for sickle cell anaemia.
2. Name the disorder with the following chromosome complement.
   (i) 22 pairs of autosomes + X X Y
   (ii) 22 pairs of autosomes + 21st chromosome + XY.
3. A test is performed to know whether the given plant is homozygous dominant or heterozygous. Name the test and phenotypic ratio of this test for a monohybrid cross.

SA-I (2 Marks)

4. Identify the sex of organism as male or female in which the sex chromosome are found as (i) ZW in bird (ii) XY in Drosophila (iii) ZZ in birds, (iv) XO in grasshopper.
5. The human male never passes on the gene for haemophilia to his son. Why is it so ?
6. Mention four reasons why Drosophila was chosen by Morgan for his experiments in genetics.
7. Differentiate between point mutation and frameshift mutations.

SA-II (3 Marks)

8. A woman with O blood group marries a man with AB blood group
   (i) Work out all the possible phenotypes and genotypes of the progeny.
   (ii) Discuss the kind of dominance in the parents and the progeny in this case.
9. Give reasons for success of Mendel.
10. In Mendel's breeding experiment on garden pea, the offspring of F_2 generation are obtained in the ratio of 25% pure yellow pod, 50% hybrid green pods and 25% green pods State (i) which pod colour is dominant (ii) The Phenotypes of the individuals of F_1 generation, (iii) Workout the cross.

LA **(5 Marks)**

11. A dihybrid heterozygous round, yellow seeded garden pea (*Pisum sativum*) was crossed with a double recessive plant.
    (i) What type of cross is this?
    (ii) Work out the genotype and phenotype of the progeny.
    (iii) What principle of Mendel is illustrated through the result of this cross?

Answers

VSA **(1 Mark)**

1. GAG changes GUG, Glutamic acid is substituted by valine.
2. (i) Klinefetter's Syndrome (ii) Down's syndrome
3. Test cross 1 : 1.

SA-I **(2 Marks)**

4. (i) Female (ii) Male (iii) Female (iv) Male
5. The gene for haemophilia is present on X chromosome. A male has only one X chromosome which he receives from his mother and Y chromosome from father. The human male passes the X chromosome to his daughters but not to the male progeny (sons).
6. (i) Very short life cycle (2-weeks)
    (ii) Can be grown easily in laboratory
    (iii) In single mating produce a large no. of flies.
    (iv) Male and female show many hereditary variations
    (v) It has only 4 pairs of chromosomes which are distinct in size and shape.
7. **Point Mutations :** Arises due to change in a single base pair of DNA e.g., sickle cell anaemia.

    Frame shift mutations : Deletion or insertion/duplication/addition of one or two bases in DNA.

SA-II **(3 Marks)**

8. (i) Blood group AB has alleles as I^A, I^B and O group has ii which on cross gives the both blood groups A and B while the genotype of progeny will be I^Ai and I^Bi.

   (ii) I^A and I^B are equally dominant (co-dominant). In multiple allelism, the gene I exists in 3 allelic forms, I^A, I^B and *i*.

9. (i) He used large samples for his experiments.

   (ii) He selected only pure breeding varieties.

   (iii) He choose the character which had distinctive contrasts.

   (iv) He selected pea plant which can be cross-bred as well as self bred.

   (v) Use of statistical methods and law of probability.

10. (i) Green pod colour is dominant

    (ii) Green pod colour

    (iii) Parents GG (green) X gg (yellow)

    Gametes (G) (g)

    F1 generation Gg (Hybrid green)

    Gametes (G) (g) X (G) (g)

    F2 generation GG Gg Gg gg

    Phenotypic ratio 3 : 1

    Genotypic ratio 1 : 2 : 1

LA **(5 Marks)**

11. (i) It is a dihybrid test cross

    (ii) Parent RrYy (Round Yellow) rryy (Wrinkled green)

    Gametes (RY), (Ry), (rY), (ry), × (ry)

Gametes	RY	Ry	rY	ry
F1 progeny	RrYy Round, Yellow	Rryy Round and green	rrYy wrinkled, yellow	rryy wrinkled, green.

Phenotypic ratio	1	:	1	:	1	:	1
Genotopic ratio	1	:	1	:	1	:	1

F_2 Progeny

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

(iii) Principle of Independent Assortment.