

CHAPTER 4 : PRINCIPLES OF INHERITANCE AND VARIATION

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Questions Asked : 67

2 QUESTION PER PAGE AT AN AVERAGE

BUT, LETS SEE WHICH PAGE HAS MAXIMUM WEIGHTAGE

- Which page has the maximum

60, 73 = 2 Page

27.3% QUESTIONS

2010-2024		Page No.
	0	52,53,63,66,68, 70,76,77,93
10,14,15,16(2), 18(2),19,,21(2),22(2)	1	56,57,58,62,64 70,71,74
11(2),17,20,22	2	54,69
10(2),11,12(2),14,15(3),16 17(2),18,19(2),20,21,22	4	55,59,60,75
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12(2),13,16(2), 18,20,22(2)	7	73
10,13(2),15(2), 16,17,20	8	60

PRINCIPLES OF INHERITANCE AND VARIATION //

1. **Genetics** deals with the inheritance as well as variation of characters from parents to offsprings.
2. **Inheritance** is the process progeny by which characters are passed on from parent to progeny.
3. **Variation** is the degree by which progeny differs from their parents.
4. Indian breeds (**Sahiwal cows**) develop in **Punjab**.
5. Gregor mendel conducted **hybridisation experiment on Garden Peas for seven year** (1856-1563) & proposed laws of inheritance in living orgnaisms.

[NEET 2017][NCERT 54]

6. Mendal selected **14 true**-breeding pea plant varieties.

[NEET 2020] [NCERT 54]

7. **Seven pairs** of contrasting traits studied by mendel on pea plants

[NEET 2015] [NCERT 70]

Character	Dominant traid	Recessive traid
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[NEET 2017,2015] [NCERT 55]

Seed shape	Round	Wrinkled
Seed colour	Yellow	Green
Flower colour	Voilet	White
Pod shape	Full	Constricted
Pod colour	Green	Yellow
Flower position	Axial	Terminal
Stem Height	Tall	Dwarf

PRINCIPLES OF INHERITANCE AND VARIATION //

8. **Genes** - unit of inheritance, contain the information that is required to express a particular trait.
9. Mendal called **factors** (Gene)
10. **Alleles** - Genes which code for a pair of contrasting traits, they are slightly different forms of the same gene.

[NEET 2024, 2022, 2015][NCERT 56]

11. **Punnett square** - Graphical representation to calculate the probability of all possible genotypes of offspring in a genetic cross.

- Developed by a british geneticist, Reginald C. Punnett.

12. **Monohybrid cross**

- Study of inheritance of **one character** at a time in an organism
- Phenotypic ratio = 3 : 1
- Genotypic ratio = 1 : 2 : 1

13. Based on his observation on monohybrid crosses mendal proposed two general rules. These rules are called principles **or** law of interitance.

1. Law of Dominance
2. Law of Segregation

14. **Law of Dominance**

[NEET 2024, 2018,2016] [NCERT 58]

1. Characters are controlled by discrete units called factors.
2. Factors occur in pairs
3. In a dissimilar pair of factors one member of the pair dominates & other recessive.

- Law of Dominance used to explain the expression of only one of the parental characters in monohybrid cross in F_1 & expression of both in F_2 .

15. Law of Segregation

- This law is based on the fact that the alleles do not show any blending & both character are recovered as such in the F_2 generation.
- The factors or alleles of a pair segregate from each other such that a gamete receive only one of the two factors.
- **Law of Dominance have two exceptions**
 1. Incomplete Dominance
 2. Co-dominance

[NEET 2019, 2016, 2012, 2018] [NCERT 59]

INCOMPLETE DOMINANCE //

- F_1 phenotype did not resemble either of the two parents & was in between the two
 - Phenotypic ratio change from 3 : 1 to 1 : 2 : 1
 - Phenotypic & Genotypic ratio = 1 : 2 : 1
 - Examples : Flower colour in the dog flower (**Snapdragon or Antirrhinum spl**) & Starch grain size

[RE-NEET-2024] [NEET 2024]

[NEET 2015, 2018, 2016] [NCERT 60]

16. Co-dominance

- F_1 Phenotype resembles both parents

[NEET 2013] [NCERT 61]

- Phenotypic & Genotypic ratio = 1 : 2 : 1
- Eg: AB blood group ($I^A I^B$)

[RE-NEET-2024] [NEET 2013] [NCERT 61]

17. ABO blood group in human Beings

[NEET 2020, 2018] [NCERT 61]

- ABO blood group controlled by the gene i .
- Gene i has three alleles I^A , I^B & i
- I^A & I^B produce slightly different form of sugar while alleles i doesn't produce any sugars.

GENETIC BASIS OF BLOOD GROUPS IN HUMAN POPULATION //

Allele from Parent 1	Allele from Parent 2	Genotype of offspring	Blood types of offspring
I^A	I^A	$I^A I^A$	A
I^A	I^B	$I^A I^B$	AB
I^A	i	$I^A i$	A
I^B	I^A	$I^A I^B$	AB
I^B	I^B	$I^B I^B$	B
I^B	i	$I^B i$	B
i	i	$i i$	O

[NEET 2015,2024] [NCERT 61]

- ABO Blood grouping also provides a good example of multiple alleles

[RE-NEET-2024] [NEET 2015,2018][NCERT 62]

- Phenotype = 4, Genotypes=6, Alleles = 3

[NEET 2017] [NCERT 61]

- An individual only two alleles can present, multiple alleles can be found only when population studied are made.
- **Pleiotropy** : A single gene product may produce **more than one effect**

Eg: Starch synthesis in pea seeds

[NEET 2016] [NCERT 62]

18. Dihybrid cross

- Study of inheritance of two characters at a time.
- Phenotypic ratio = 9:3:3:1
- Genotypic ratio = 1:2:2:4:1:2:1:2:1

19. Law of Independent Assortment states that, “when two pairs of traits are combined in a hybrid, segregation of one pair of characters is independent of the other pairs of characters”.

[NEET 2022] [NCERT 64]

20. Important formula

- Type of gamete/type of phenotypes = 2^n (n = no. of Heterozygous.)
- Type of genotypes = 3^n

21. Test cross : It is used to find out the genotype of F₁ individual.

[NEET 2012, 2016, 2024] [NCERT 58]

- F₁ individual crossed with homozygous recessive parent.
- Test cross ratio : Monohybrid $\Rightarrow 1 : 1$
Dihybrid $\Rightarrow 1 : 1 : 1 : 1$
- Mendel published his work on inheritance of characters in **1865** but it remained unrecognised **till 1900** due to several reasons.

1. Communication was not easy in those days.
2. His concept of genes as stable & discrete unit
3. Use mathematics to explain biological phenomena

22. In 1900, de Vries, Correns & Von Tschermak independently rediscovered Mendel's results on the inheritance of characters.

23. **Walter sutton & Theodore Boveri** noted that behaviour of chromosomes was parallel to the behaviour of genes & used chromosomal movement in **1902**.

24. **Experimental verification** of the chromosomal theory of inheritance given by **Thomas Hunt Morgan & his colleagues**.

[NEET 2020] [NCERT 67]

25. **Morgan** work on fruit fly (**Drosophila melanogaster**) due to following reason

- Grow in simple synthetic medium in lab.
- Life cycle complete in two weeks
- Many type of hereditary variations

26. **Linkage & Recombination**

- Physical association or linkage of two genes called linkage & Recombination of describe the generation of non-parental gene combination.

[NEET 2015] [NCERT 67]

- In linkage, the proportion of parental gene combination are much **higher** than non-parental type.
- Tightly linked gene shows very low recombination while loosely linked genes shows higher recombination

27. **Alfred sturtevant** : Distance between genes & mapped their position of chromosome

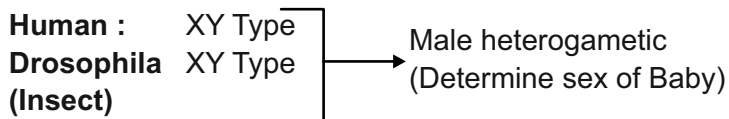
[NEET 2019,23] [NCERT 67]

- $\text{Linkage 1/Distance b/w genes 1/Recombination}$
- Recombination Crossing over

28. **Sex determination**

- Henking (1891) experiments carried out in insect & named **X-Body**. but on investigation by other scientist, concluded that **X-Body** of Henking are **X-chromosomes**.

29. Sex determination in -



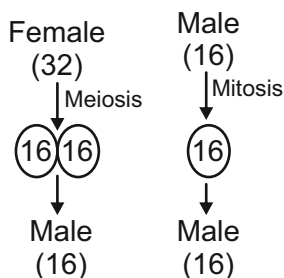
- **Most Insects (Grass hopper) : XO type** - Male heterogametic (Determine sex of baby)

[NEET 2022, 2018] [NCERT 70]

- **Birds : ZW type** - Female heterogametic (Determine sex of baby)
- It is also evident that in each pregnancy there is always **50%** probability of either a male or a female child.

30. Sex determination in honey bee

- Based on number of sets of chromosomes in individual receives.
- **Honey bee colony have -**



Queen, worker - Female (Diploid 32 chromosomes)

Drones - Male (16 chromosomes, Haploid)

- Known as **haplo-diploid sex-determination system**

MUTATION //

- Results in **alteration** of DNA sequences & results in changes in genotype & phenotype of an organism & leads to variation in DNA
- Mutation are due to **loss (Deletions) OR Gain (insertion/ (Duplication)** of a segment of DNA result in alteration in chromosomes.
- Chromosomal alteration result in abnormalities/Aberrations.
- Chromosomal aberrations are commonly observed in **Cancer cells**.

Point mutation - due to change in **single base pair of DNA**
eg - **Sickle cell Anaemia**.

Frame shift mutation - Deletions **or** insertions of base pairs of DNA.

31. Pedigree Analysis

- Study of the family history about inheritance of a particular trait provides an alternative.
- Pedigree analysis provides a strong tool, utilised to trace the inheritance of a specific trait, abnormality or disease.

32. Symbols used in human pedigree analysis.



: Male



: Female



: Sex unspecified



: Affected Individuals



: Mating



: Mating between relatives (consanguineous mating)

[NEET 2023][NCERT 72]

Genetic Disorders

Mendelian Disorder

- Colour blindness
- Haemophilia
- Sickle cell anaemia
- Phenylketonuria
- Thalassemia
- Cystic Fibrosis

Chromosomal Disorder

- Down syndrome
- Klinefelter's syndrome
- Turner's syndrome

33. **Colour blindness : sex-linked recessive disorder** due to defect in either red or green cone of eye resulting in failure to discriminate between red & green colour

[2022, 2012] [NCERT 73]

- Occurs in about **8 percent of males** & only about **0.4 percent of female**.

[NEET 2016, 2014] [NCERT 73]

34. **Haemophilia : sex-linked recessive disorder**, transmission from unaffected carrier female to some of the male progeny

[NEET 2022, 2020, 2016, 2013] [NCERT 74]

- A single protein that is a part of cascade of proteins involved in the clotting of blood is affected.
- Possibility of female becoming a haemophilic is extremely rare.
- Queen victoria - carrier of the disease.

35. **Sickle-cell Anaemia : Autosomal linked recessive disease**.

[NEET 2022, 2021, 2020, 2016] [NCERT 74]

- Disease occurs when both partner are heterozygous (carrier)

- **Three pair of Alleles**

$Hb^A Hb^A \Rightarrow$ Normal

$Hb^A Hb^S \Rightarrow$ Carrier

$Hb^S Hb^S \Rightarrow$ Disease

- Defect is caused by **substitution** of Glutamic acid (Glu) by valine (val) at the **six** position of beta globin chain of Hb molecules.
- Change takes place from **GAG to GUG**
- Shape of RBCs changes from **Biconcave disc to elongated sickle shaped** due to low oxygen tension.

36. **Phenylketonuria** : Inborn error of metabolism, Autosomal recessive trait.

- Affected individual **lack** the enzyme **phenylalanine hydroxylase** (that convert phenylalanine into tyrosine)
- Due to **lack** of this enzyme phenylalanine \Rightarrow Phenyl pyruvic acid
- Mental retardation, Hair loss & skin pigmentation takes place.

37. **Thalassemia** : **Autosomal linked recessive** blood disease

[NEET 2024, 2022, 2020, 2013] [NCERT 75]

- Disease takes place when both partner are carrier
- Defect could due to **either deletion/Mutation** of globin molecule.
- **Anaemia** is the characteristic of this disease.
- **Two types**
 1. Thalassemia (α globin chain affected)
 2. Thalassemia (β globin chain affected)
- α **Thalassemia** - 16th chromosomes, controlled by two closely linked gene. HBA1 & HBA2

- β **Thalassermia** - 11th chromosomes, controlled by one linked gene HBB.
- Thalassemia differs from sickle cell anaemia is that thalassemia is quantitative problem & sickle cell anaemia is qualitative problem of Haemoglobin molecules.

[NEET 2017][NCERT 75]

38. Chromosomal disorders -

- Caused due to **absence or excess or abnormal** arrangement of one **or** more chromosomes.
- **Aneuploidy** : Gain **or** loss of chromosomes due to failure of segregation of chromatids. [RE-NEET-2024]

Down's syndrome : First described by **Langdon down** (1866)

- Trisomy of 21th chromosomes

[NEET 2016, 2013, 2023, 2024] [NCERT 76]

- **Affected individual shows** - Short statured with small round head, furrowed tongue, partially open mouth, palm broad with crease.

congenital heart disease physical, psychomotor & mental retardation takes place.

39. Klinefelter's syndrome

[NEET 2019, 2016, 2023. 2024] [NCERT 76]

- Additional copy of X-chromosomes (**47,XXY**)
- **Such individual has** -

Overall masculine development

Feminine development (Gynacomastia)

Sterile individuals.

40. Turner syndrome

[NEET 2019][NCERT 76]

- Absence of one of the X-chromosomes. (**45+XO**)
- Such female are **sterile**
- Rudimentary ovary
- Lack of secondary sexual characters.

41. Polygenic inheritance

[NEET 2016] [NCERT 69]

- Single trait controlled by **three or more genes**
- Dark skin colour (**Dominant**) controlled by three genes - AAB \overline{B} CC
- Light skin colour (**Recessive**) controlled by three genes - aabbcc
- Intermediate skin colour also present
- **Examples** : Human height & Human skin colour.

[RE-NEET-2024]

42. Single gene affecting multiple phenotypic expression.

[NEET 2023] [NCERT 69]