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CHAPTER NOTES

Principles of Inheritance and Variation Class 12 Notes — CBSE Biology Chapter 5

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Principles of Inheritance and Variation Class 12 Notes — CBSE Biology Chapter 5

Chapter 5 — **Principles of Inheritance and Variation** — is the genetics chapter, one of the most important in Class 12 Biology with **10-12 marks** in Board exams. It covers Mendel's laws, chromosomal theory, linkage, sex determination, and genetic disorders. Master the crosses and pedigree analysis!

Key Concepts

Mendel's Laws of Inheritance

Law of Dominance: In a cross between two organisms with contrasting alleles, only the dominant trait appears in F₁.

Law of Segregation (Law of Purity of Gametes): During gamete formation, the two alleles of a gene segregate so each gamete receives only one allele.

Law of Independent Assortment: Alleles of different genes assort independently during gamete formation (applies when genes are on different chromosomes).

Monohybrid Cross ($Tt \times Tt$)

F₂ Genotype ratio: 1 TT : 2 Tt : 1 tt

F₂ Phenotype ratio: **3 Tall : 1 Dwarf**

Test cross ($Tt \times tt$): 1 Tall : 1 Dwarf (confirms heterozygosity)

Dihybrid Cross ($RrYy \times RrYy$)

F₂ Phenotype ratio: **9:3:3:1**

9 Round Yellow : 3 Round Green : 3 Wrinkled Yellow : 1 Wrinkled Green

Incomplete Dominance

Neither allele is completely dominant. F₁ shows **intermediate phenotype**.

Example: Snapdragon flower colour

RR (Red) $\times$ rr (White) $\rightarrow Rr$ (Pink) in F_1

F_2 ratio: 1 Red : 2 Pink : 1 White (1:2:1)

Codominance

Both alleles express themselves simultaneously.

Example: ABO blood groups

$I^A I^A$ or $I^A i$ $\rightarrow$ Blood group A

$I^B I^B$ or $I^B i$ $\rightarrow$ Blood group B

$I^A I^B$ $\rightarrow$ Blood group AB (codominance — both A and B antigens present)

ii $\rightarrow$ Blood group O

Sex Determination

System	Male	Female	Examples
XX-XY	XY (heterogametic)	XX (homogametic)	Humans, Drosophila
ZZ-ZW	ZZ (homogametic)	ZW (heterogametic)	Birds, some reptiles
XX-XO	XO	XX	Grasshoppers

Sex-Linked Inheritance

Colour blindness (X-linked recessive):

$X^c Y$ (affected male) $\times$ $X^C X^C$ (normal female) $\rightarrow$ all normal (carrier daughters)

$X^C X^c$ (carrier female) $\times$ $X^C Y$ (normal male) $\rightarrow$ 50% sons affected

Haemophilia (X-linked recessive): Same pattern as colour blindness

Males are more commonly affected (only need one defective allele on X)

Linkage and Recombination

- **Linkage:** Genes on the same chromosome tend to be inherited together (discovered by Morgan using Drosophila)
- **Recombination:** Crossing over breaks linkage $\rightarrow$ new combinations of genes

- Recombination frequency = (recombinant offspring / total offspring) × 100
- Tightly linked genes → low recombination frequency

Chromosomal Disorders

Disorder	Chromosomal Condition	Features
Down Syndrome	Trisomy 21 (47 chromosomes)	Mental retardation, short stature, broad face
Turner Syndrome	45, XO (monosomy)	Female, short stature, sterile, webbed neck
Klinefelter Syndrome	47, XXY (trisomy)	Male, gynaecomastia, sterile

Mendelian Disorders

Disorder	Inheritance	Feature
Sickle Cell Anaemia	Autosomal recessive	HbS (Glu→Val at position 6 of β-chain)
Phenylketonuria	Autosomal recessive	Cannot convert phenylalanine → tyrosine
Thalassemia	Autosomal recessive	Reduced synthesis of α or β chains of haemoglobin
Colour Blindness	X-linked recessive	Cannot distinguish red-green colours
Haemophilia	X-linked recessive	Blood doesn't clot properly

Solved Examples

Example 1: Monohybrid Cross

Q: In pea plants, tall (T) is dominant over dwarf (t). Cross a heterozygous tall plant with a dwarf plant. What is the expected phenotype ratio?

Solution: $Tt \times tt \rightarrow Tt, Tt, tt, tt \rightarrow 1 \text{ Tall} : 1 \text{ Dwarf}$ (test cross ratio)

Example 2: Blood Groups

Q: A mother has blood group A ($I^A i$) and father has blood group B ($I^B i$). What blood groups are possible in their children?

Solution: $I^A i \times I^B i \rightarrow I^A I^B$ (AB), $I^A i$ (A), $I^B i$ (B), ii (O). **All four blood groups are possible!**

Important Questions for Board Exams

1 Mark

1. State the law of segregation.
2. What is a test cross?
3. Why is haemophilia more common in males?

3 Mark

1. Explain codominance with ABO blood group example.
2. Describe the chromosomal basis of sex determination in humans.
3. What is Down syndrome? What is its chromosomal basis?

5 Mark

1. Describe Mendel's dihybrid cross. State the law of independent assortment. Give the F₂ phenotypic and genotypic ratios.
2. What is sex-linked inheritance? Explain the inheritance of colour blindness with a cross.

Quick Revision Points

- Mendel: 3 laws — dominance, segregation, independent assortment
- Monohybrid F₂: 3:1; Dihybrid F₂: 9:3:3:1; Test cross: 1:1
- Incomplete dominance: 1:2:1 (snapdragon); Codominance: ABO blood groups
- Sex determination: XX-XY (humans); ZZ-ZW (birds)
- Colour blindness & haemophilia: X-linked recessive → males more affected
- Down: trisomy 21; Turner: 45,XO; Klinefelter: 47,XXY
- Sickle cell: autosomal recessive, Glu → Val in β -globin
- Linkage: Morgan, Drosophila; Recombination frequency used for gene mapping



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