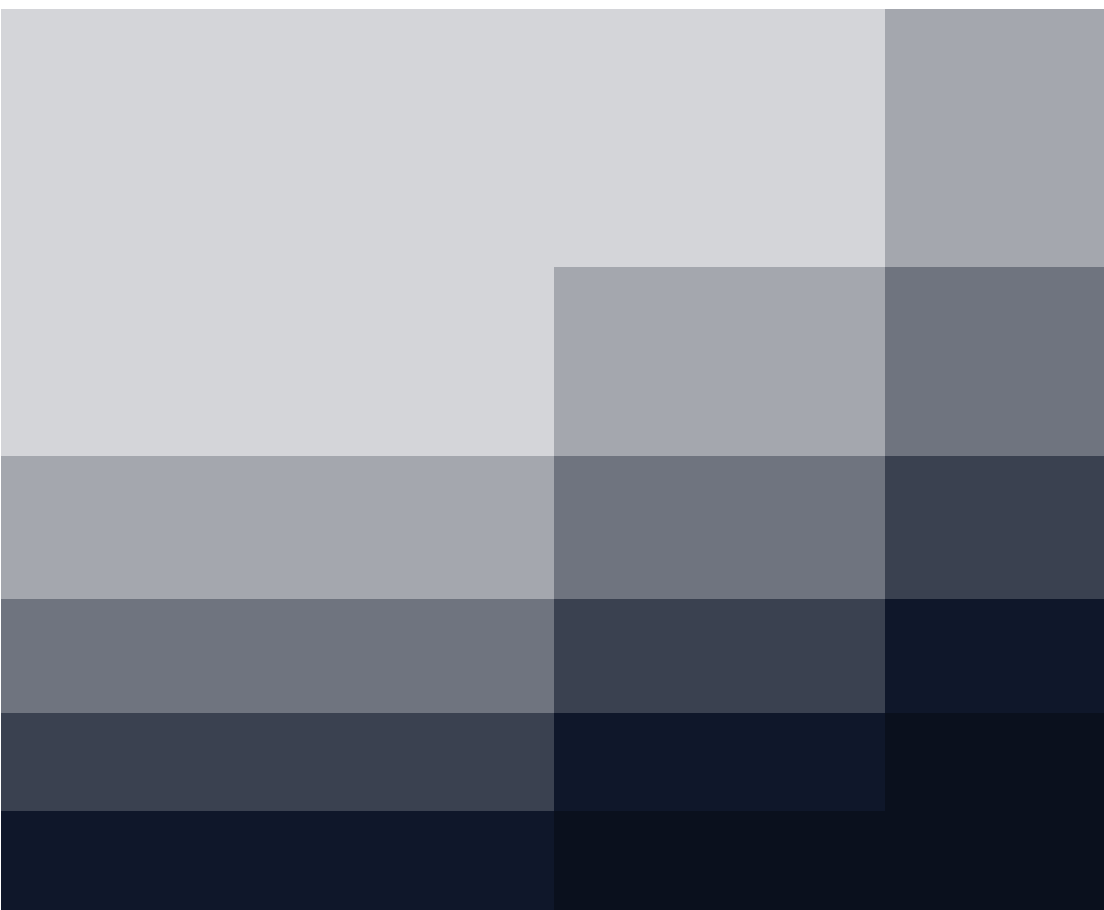




CHAPTER NOTES

Principles of Inheritance and Variation

Mendel, chromosomes and genetic disorders

SUBJECT

Biology

CLASS / COURSE

Class 12 • CBSE / NEET
foundation

PREPARED BY

Subject Teacher

How to Use These Notes

These notes cover Mendel's laws, chromosomal theory, sex determination and genetic disorders. Each chapter starts with a short overview, then lists the key definitions, explains the core concepts with examples, and ends with practice questions. Answers to the multiple-choice questions are at the end.

Study plan

- Read the overview and definitions first
- Work through the solved examples without looking at the answers
- Attempt the practice questions in timed conditions
- Revise using the summary sheet the night before the exam

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CHAPTER 1: MENDEL'S LAWS OF INHERITANCE

Gregor Mendel studied seven pairs of contrasting traits in garden pea and proposed that traits are controlled by factors (now called genes) passed from parents to offspring in predictable ratios.

Key Definitions

Term	Meaning
Gene	Unit of inheritance that controls a trait
Allele	Alternative form of a gene
Genotype	Genetic make-up of an organism, e.g. Tt
Phenotype	Observable character, e.g. tall
Homozygous	Having identical alleles (TT or tt)
Heterozygous	Having different alleles (Tt)

Why Garden Pea?

Pea plants have clear contrasting traits, a short life cycle, many seeds per generation and flowers that normally self-pollinate but can be cross-pollinated by hand.

Monohybrid and Dihybrid Crosses

A cross between tall (TT) and dwarf (tt) plants gives all tall F_1 plants; selfing F_1 gives tall and dwarf in a 3:1 phenotypic ratio and 1:2:1 genotypic ratio. In a dihybrid cross, the F_2 phenotypic ratio is 9:3:3:1.

- Law of dominance
- Law of segregation
- Law of independent assortment

Solved Examples

Q1. A heterozygous tall plant is crossed with a dwarf plant. Give the ratio of offspring.

$Tt \times tt$ gives Tt and tt in 1:1 — this is a test cross.

Q2. Why is the law of segregation also called the law of purity of gametes?

Each gamete receives only one allele of a pair, so gametes are always “pure” for a trait.

Exam tip

Draw Punnett squares neatly with gametes labelled on both axes — a correct square earns marks even if the ratio is miswritten.

Practice Questions

A. Multiple choice

- Phenotypic ratio of a monohybrid F_2 is
(a) 1:2:1 (b) 3:1 (c) 9:3:3:1 (d) 1:1
- A test cross is a cross with
(a) Dominant parent (b) Recessive parent (c) F_1 hybrid (d) Any parent
- Tt is
(a) Homozygous (b) Heterozygous (c) Recessive (d) Pure
- Dihybrid F_2 phenotypic ratio is
(a) 3:1 (b) 9:3:3:1 (c) 1:1 (d) 1:2:1

B. Short answer

- Define allele with an example.
- What is a test cross and why is it done?
- State the law of independent assortment.

C. Long answer

- Explain a monohybrid cross with a Punnett square up to F_2 .
- Explain a dihybrid cross and the law of independent assortment.

CHAPTER 2: DEVIATIONS FROM MENDELISM

Not all traits follow simple dominance. Incomplete dominance, co-dominance, multiple alleles and polygenic inheritance all produce ratios different from Mendel's.

Key Definitions

Term	Meaning
Incomplete dominance	Heterozygote shows an intermediate phenotype
Co-dominance	Both alleles express fully in the heterozygote
Multiple alleles	More than two alleles for a gene in a population
Pleiotropy	One gene affects several traits
Polygenic inheritance	Trait controlled by several genes, e.g. skin colour

Examples

In snapdragon, red $\times$ white gives pink F_1 , and F_2 shows red, pink and white in 1:2:1. Human ABO blood groups show both co-dominance (I^A and I^B) and multiple alleles (I^A , I^B , i).

Solved Examples

Q1. What blood groups are possible in children of $I^A i \times I^B i$ parents?

A, B, AB and O, each with a probability of 1/4.

Q2. Why is the F_2 ratio in snapdragon 1:2:1 for both phenotype and genotype?

Because the heterozygote has its own distinct (pink) phenotype.

Exam tip

For blood-group questions, write each parent's genotype first, then use a 2×2 Punnett square.

Practice Questions**A. Multiple choice**

1. Pink flowers in snapdragon show

(a) Dominance (b) Incomplete dominance (c) Co-dominance (d) Pleiotropy

2. AB blood group shows

(a) Incomplete dominance (b) Co-dominance (c) Recessiveness (d) Epistasis

3. Number of alleles for ABO gene

(a) 2 (b) 3 (c) 4 (d) 6

4. Skin colour in humans is

(a) Monogenic (b) Polygenic (c) Sex-linked (d) Co-dominant

B. Short answer

6. Distinguish incomplete dominance from co-dominance.

7. What is pleiotropy? Give an example.

8. Why is human skin colour called polygenic?

C. Long answer

9. Explain ABO blood groups as an example of multiple alleles and co-dominance.

10. Explain incomplete dominance with a cross in snapdragon up to F_2 .

CHAPTER 3: SEX DETERMINATION AND GENETIC DISORDERS

Chromosomes carry genes, and special sex chromosomes decide sex in many organisms. Changes in genes or chromosome number cause genetic disorders that can be traced through pedigrees.

Key Definitions

Term	Meaning
Autosomes	Chromosomes other than sex chromosomes
Pedigree analysis	Study of a trait across generations of a family
Mendelian disorder	Caused by mutation in a single gene
Chromosomal disorder	Caused by gain, loss or rearrangement of chromosomes
Aneuploidy	Gain or loss of one or a few chromosomes

Sex Determination

Humans show XX–XY determination: females produce only X eggs, while males produce X or Y sperm in equal numbers, so the father's sperm decides the sex of the child. Birds show ZW–ZZ, where the female is heterogametic.

Common Disorders

Haemophilia and colour blindness are X-linked recessive and appear more often in males. Sickle-cell anaemia is autosomal recessive. Down's syndrome results from trisomy of chromosome 21; Turner's (XO) and Klinefelter's (XXY) involve sex chromosomes.

Disorder	Type	Cause
Haemophilia	Mendelian, X-linked recessive	Defective clotting factor gene
Sickle-cell anaemia	Mendelian, autosomal recessive	Single base change in β -globin gene
Down's syndrome	Chromosomal	Trisomy 21
Klinefelter's syndrome	Chromosomal	XXY
Turner's syndrome	Chromosomal	XO

Table: Genetic disorders at a glance

Solved Examples

Q1. A colour-blind man marries a woman with normal vision whose father was colour blind. What fraction of sons will be colour blind?

The woman is a carrier (X^cX), so half of the sons will be colour blind.

Q2. Why are X-linked recessive disorders more common in men?

Men have only one X chromosome, so a single recessive allele is expressed.

Exam tip

Use standard pedigree symbols — squares for males, circles for females, shaded for affected — and label generations with Roman numerals.

Practice Questions

A. Multiple choice

1. Down's syndrome is due to trisomy of

- (a) 18 (b) 21 (c) 13 (d) X

2. Haemophilia is

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

3. Karyotype of Turner's syndrome

(a) XXY (b) XO (c) XYY (d) XXX

4. In birds, the female is

(a) ZZ (b) ZW (c) XX (d) XY

B. Short answer

11. Why is the father responsible for the sex of a child?

12. Name two X-linked recessive disorders.

13. What is aneuploidy?

C. Long answer

14. Describe sex determination in humans, birds and honeybees.

15. Explain sickle-cell anaemia and Down's syndrome with their causes and symptoms.

QUICK REVISION SHEET

Chapter	Term	Remember
Mendel's Laws of Inheritance	Gene	Unit of inheritance that controls a trait
Mendel's Laws of Inheritance	Allele	Alternative form of a gene
Mendel's Laws of Inheritance	Genotype	Genetic make-up of an organism, e.g. Tt
Deviations from Mendelism	Incomplete dominance	Heterozygote shows an intermediate phenotype
Deviations from Mendelism	Co-dominance	Both alleles express fully in the heterozygote
Deviations from Mendelism	Multiple alleles	More than two alleles for a gene in a population
Sex Determination and Genetic Disorders	Autosomes	Chromosomes other than sex chromosomes
Sex Determination and Genetic Disorders	Pedigree analysis	Study of a trait across generations of a family
Sex Determination and Genetic Disorders	Mendelian disorder	Caused by mutation in a single gene

ANSWER KEY

Chapter	Question	Answer
1	1	b

Chapter	Question	Answer
1	2	b
1	3	b
1	4	b
2	1	b
2	2	b
2	3	b
2	4	b
3	1	b
3	2	b
3	3	b
3	4	b