Chapter 4: Principles of Inheritance and Variation - Back Exercises

NCERT Biology Class 11

4.1 Advantages of Pea Plant Selection

Question: Mention the advantages of selecting pea plant for experiment by Mendel.

Answer: (a) True-breeding varieties: Pea plants show stable trait inheritance through continuous self-pollination over several generations.

- **(b) Contrasting traits:** Clear either/or characteristics (tall/dwarf, yellow/green seeds) with no intermediate forms, making analysis straightforward.
- **(c) Easy artificial pollination:** Large flowers allow easy cross-pollination by removing anthers and transferring pollen manually.
- **(d) Short generation time:** Quick life cycle enables study of multiple generations within reasonable time.
- (e) High fertility: Large number of offspring provides statistically significant data for ratio analysis.
- (f) Bisexual flowers: Both self and cross-pollination possible, giving experimental flexibility.
- (g) Clear visibility: Contrasting traits easily observable without microscopic examination.

4.2 Basic Genetic Terminology

Question: Differentiate between the following:

(a) Dominance and Recessive

Aspect	Dominant	Recessive
Expression	Expressed in heterozygous condition	Only expressed in homozygous condition
Symbol	Capital letter (T for tall)	Small letter (t for dwarf)
Masking ability	Masks recessive allele expression	Masked by dominant allele
F ₁ appearance	Always appears in F ₁	Never appears in F₁
Molecular basis	Usually produces functional enzyme	Often produces non-functional enzyme
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(b) Homozygous and Heterozygous

Aspect	Homozygous	Heterozygous
Allele composition	Both alleles identical (TT or tt)	Both alleles different (Tt)
Gamete types	Produces only one type of gamete	Produces two types of gametes
Breeding behavior	True-breeding	Not true-breeding
F ₁ from selfing	All offspring similar to parent	Shows segregation in F ₁
Examples	TT (tall), tt (dwarf)	Tt (tall but carrier)
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(c) Monohybrid and Dihybrid

Aspect	Monohybrid	Dihybrid
Characters studied	Single character (e.g., height only)	Two characters (e.g., height and seed color)
F ₁ genotype	Tt	TtRr
F ₂ phenotypic ratio	3:1	9:3:3:1
F ₂ genotypic ratio	1:2:1	Complex (16 squares in Punnett square)
Gamete types in F ₁	2 types (T, t)	4 types (TR, Tr, tR, tr)
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4.3 Gamete Types in Heterozygous Organism

Question: A diploid organism is heterozygous for 4 loci, how many types of gametes can be produced?

Solution: For an organism heterozygous at n loci, number of different gamete types = 2^n

Given: Heterozygous for 4 loci (e.g., AaBbCcDd)

Number of gamete types = 2^4 = **16 different types**

Gamete types: ABCD, abcd

4.4 Law of Dominance Explanation

Question: Explain the Law of Dominance using a monohybrid cross.

Answer: Law of Dominance states:

- 1. Characters are controlled by discrete units called factors (genes)
- 2. Factors occur in pairs (alleles)
- 3. In a dissimilar pair, one member dominates (dominant) over the other (recessive)

Monohybrid Cross Example: Cross: Tall plant (TT) × Dwarf plant (tt)

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P Generation: TT (Tall) × tt (Dwarf)

Gametes: T t

F<sub>1</sub> Generation: All Tt (Tall phenotype)

F<sub>2</sub> Generation: TT: Tt: tt

1:2:1 (Genotypic ratio)

Tall: Dwarf

3:1 (Phenotypic ratio)
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Explanation:

- In F₁, all plants are Tt but show tall phenotype (dominance of T over t)
- In F_2 , 3/4 plants are tall (TT + Tt) and 1/4 are dwarf (tt)
- Recessive trait (dwarf) reappears in F2, proving factors don't blend
- Demonstrates that T (tall) is dominant over t (dwarf)

4.5 Test Cross Design

Question: Define and design a test-cross.

Definition: Test cross is a cross between an organism showing dominant phenotype (with unknown genotype) and the homozygous recessive parent.

Purpose: To determine the genotype of an organism showing dominant trait.

Design: Unknown genotype (Tall plant) × Known recessive (tt)

Case 1: If unknown is TT (homozygous dominant)

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TT \times tt

Gametes: T \times t

F<sub>1</sub>: All Tt (100% tall)

Ratio: 1:0 (Tall:Dwarf)
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Case 2: If unknown is Tt (heterozygous)

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Tt × tt

Gametes: T,t × t

F<sub>1</sub>: Tt, tt (50% tall, 50% dwarf)

Ratio: 1:1 (Tall:Dwarf)
```

Interpretation:

- All tall offspring → Unknown parent was TT
- 1:1 ratio → Unknown parent was Tt

4.6 Punnett Square Analysis

Question: Using a Punnett Square, work out the distribution of phenotypic features in the first filial generation after a cross between a homozygous female and a heterozygous male for a single locus.

Cross: Homozygous female (tt) × Heterozygous male (Tt)

Punnett Square:

	Т	t
t	Tt	tt
t	Tt	tt
◄		

Results:

• **Genotypic ratio:** 1 Tt : 1 tt (1:1)

• Phenotypic ratio: 1 Tall : 1 Dwarf (1:1)

• Percentage: 50% tall plants, 50% dwarf plants

Interpretation: Equal probability of offspring showing either dominant or recessive trait due to heterozygous male contributing equal proportions of T and t gametes.

4.7 Dihybrid Test Cross

Question: When a cross is made between tall plant with yellow seeds (TtYy) and tall plant with green seeds (Ttyy), what proportions of phenotype in the offspring could be expected to be: (a) tall and green (b) dwarf and green

Cross: TtYy × Ttyy

Gametes:

• TtYy produces: TY, Ty, tY, ty (1:1:1:1)

• Ttyy produces: Ty, ty (1:1)

Punnett Square:

	Ту	ty
TY	ТТҮу	TtYy
Ту	ТТуу	Ttyy
tY	TtYy	ttYy
ty	Ttyy	ttyy
▲	•	>

Phenotypic Analysis:

• Tall, Yellow: TTYy, TtYy, TtYy = 3/8

• **Tall, Green:** TTyy, Ttyy, Ttyy = 3/8

• **Dwarf, Yellow:** ttYy = 1/8

• **Dwarf, Green:** ttyy = 1/8

Answers: (a) Tall and green: 3/8 or 37.5% (b) Dwarf and green: 1/8 or 12.5%

4.8 Linked Genes in Dihybrid Cross

Question: Two heterozygous parents are crossed. If the two loci are linked, what would be the distribution of phenotypic features in F_1 generation for a dihybrid cross?

Answer: When two loci are **linked** (located on same chromosome), they do **NOT** follow Mendel's Law of Independent Assortment.

Normal dihybrid (unlinked): AaBb × AaBb gives 9:3:3:1 ratio

Linked genes: Ratio deviates significantly from 9:3:3:1

Example: If A is linked to B, and a is linked to b

• Parental combinations: AB and ab (more frequent)

• Recombinant combinations: Ab and aB (less frequent)

Typical linked cross result:

• **Parental types:** Higher frequency (>25% each)

• **Recombinant types:** Lower frequency (<25% each)

• Exact ratio depends on: Distance between genes (recombination frequency)

Closer the genes → Lower recombination → Greater deviation from 9:3:3:1

4.9 T.H. Morgan's Contributions

Question: Briefly mention the contribution of T.H. Morgan in genetics.

T.H. Morgan's Major Contributions:

(a) Drosophila as Model Organism:

- Introduced fruit flies for genetic studies
- Established advantages: short life cycle, large progeny, clear sex differentiation

(b) Discovery of Linkage:

- Proved that genes located on same chromosome don't assort independently
- Showed deviation from Mendel's 9:3:3:1 ratio in dihybrid crosses

(c) Concept of Recombination:

- Explained generation of non-parental gene combinations
- Distinguished between parental and recombinant types

(d) Chromosomal Theory Support:

- Provided experimental evidence for chromosomal theory of inheritance
- Confirmed genes are located on chromosomes

(e) Sex-linked Inheritance:

- Discovered sex-linked genes (genes on X-chromosome)
- Showed different inheritance patterns for sex-linked traits

(f) Genetic Mapping:

- Student Alfred Sturtevant developed genetic mapping using Morgan's data
- Used recombination frequency to determine gene distances

Impact: Morgan's work established modern genetics and earned him Nobel Prize in 1933.

4.10 Pedigree Analysis

Question: What is pedigree analysis? Suggest how such an analysis can be useful.

Definition: Pedigree analysis is the study of inheritance patterns of specific traits through family trees across multiple generations.

Method:

- Uses standard symbols to represent individuals
- Tracks trait occurrence through generations
- Analyzes inheritance patterns

Standard Symbols:

Affected male

- Affected female
- O Normal male
- O Normal female
- Marriage line: —
- Offspring line: |

Uses of Pedigree Analysis:

(a) Determine Inheritance Pattern:

- Autosomal dominant/recessive
- Sex-linked inheritance
- Co-dominant traits

(b) Genetic Counseling:

- Predict probability of affected offspring
- Risk assessment for couples
- Family planning guidance

(c) Medical Diagnosis:

- Identify carriers of genetic disorders
- Trace disease transmission
- Early detection possibilities

(d) Research Applications:

- Study rare genetic disorders
- Understand population genetics

• Identify new genetic syndromes

Limitations:

- Small family sizes reduce statistical power
- Environmental factors may confuse analysis
- Incomplete penetrance can complicate interpretation

4.11 Human Sex Determination

Question: How is sex determined in human beings?

Human Sex Determination - XY System:

Chromosomal Composition:

• Total chromosomes: 46 (23 pairs)

• Autosomes: 22 pairs (identical in males and females)

• **Sex chromosomes:** 1 pair (different in males and females)

Sex Chromosome Pattern:

• Females: 46, XX (homogametic)

• Males: 46, XY (heterogametic)

Mechanism:

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Male (XY) × Female (XX)
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Male Gametes: 50% X-bearing, 50% Y-bearing

Female Gametes: 100% X-bearing

Fertilization:

 $X (egg) + X (sperm) \rightarrow XX (Female)$ $X (egg) + Y (sperm) \rightarrow XY (Male)$

Result: 1:1 sex ratio (50% male, 50% female)

Important Points:

- Sperm determines sex of offspring, not egg
- Equal probability of male or female child in each pregnancy
- Y chromosome carries male-determining genes (SRY gene)
- Women are NOT responsible for child's sex (social misconception)

Clinical Significance:

- Sex-linked disorders show different patterns in males vs females
- Males more susceptible to X-linked recessive disorders
- Basis for understanding sex chromosome disorders

4.12 ABO Blood Group Genetics

Question: A child has blood group O. If the father has blood group A and mother blood group B, work out the genotypes of the parents and the possible genotypes of the other offspring.

Given:

• Child: Blood group O

• Father: Blood group A

• Mother: Blood group B

Analysis:

Child with O blood group: Must have genotype ii

• Receives one i allele from father

• Receives one i allele from mother

Therefore:

• Father (A blood group): Must be I^A i (heterozygous)

• Mother (B blood group): Must be I^B i (heterozygous)

Cross: $I^A i \times I^B i$

Punnett Square:

	I^A	i
I^B	I^A I^B	I^Bi
i	I^A i	ii
◄	'	

Results:

• **I^A I^B:** AB blood group (25%)

• **I^A i:** A blood group (25%)

• **I^B i:** B blood group (25%)

• **ii:** O blood group (25%)

Answer:

• Parent genotypes: Father = I^A i, Mother = I^B i

• **Possible offspring:** AB (25%), A (25%), B (25%), O (25%)

• Genotypic ratio: 1:1:1:1

4.13 Types of Dominance

Question: Explain the following terms with examples: (a) Co-dominance (b) Incomplete dominance

(a) Co-dominance:

Definition: Both alleles in heterozygous condition express themselves simultaneously without any blending.

Characteristics:

• Both parental traits visible in F₁

• No intermediate phenotype

• Both alleles equally dominant

Example: ABO Blood Groups

• **Cross:** I^A I^A (A group) × I^B I^B (B group)

• **F**₁: I^A I^B (AB group)

• **Result:** Both A and B antigens expressed simultaneously

• **F₂ ratio:** 1 A: 2 AB: 1 B

(b) Incomplete Dominance:

Definition: Neither allele is completely dominant; heterozygote shows intermediate phenotype between the two homozygotes.

Characteristics:

- F₁ phenotype intermediate between parents
- Genotypic ratio = Phenotypic ratio in F₂
- Both alleles contribute to phenotype

Example: Snapdragon Flower Color

- Cross: RR (Red) × rr (White)
- **F**₁: Rr (Pink) intermediate color
- **F₂ cross:** Rr × Rr
- **F₂ result:** 1 RR (Red) : 2 Rr (Pink) : 1 rr (White)
- **F₂ ratio:** 1:2:1 (both genotypic and phenotypic)

Comparison:

- Complete dominance: F₂ phenotypic ratio 3:1
- Incomplete dominance: F₂ phenotypic ratio 1:2:1
- **Co-dominance:** F₂ phenotypic ratio 1:2:1 (but both traits visible in heterozygote)

4.14 Point Mutation

Question: What is point mutation? Give one example.

Definition: Point mutation is a change in a single base pair (nucleotide) in the DNA sequence, resulting in alteration of genetic information.

Types of Point Mutations:

(a) Substitution:

- One base replaced by another
- May be silent, missense, or nonsense mutation

(b) Insertion:

- Addition of single nucleotide
- Causes frameshift mutation

(c) Deletion:

- Removal of single nucleotide
- Causes frameshift mutation

Example: Sickle Cell Anemia

Normal β-globin gene:

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DNA: ...CTG ACT CCT GAG GAG...
mRNA: ...GAG UGA GGA CUC CUC...
Amino acid: ...Glu-Thr-Gly-Leu-Leu...
(position 6 = Glutamic acid)
```

Mutated β-globin gene:

```
DNA: ...CTG ACT CCT GUG GAG... (A→U)
mRNA: ...GAG UGA GGA GUG CUC...
Amino acid: ...Glu-Thr-Gly-Val-Leu...
(position 6 = Valine)
```

Consequences:

• **Molecular level:** GAG → GUG (single base change)

• **Protein level:** Glutamic acid → Valine substitution

• **Cellular level:** Hemoglobin polymerization under low oxygen

• **Physiological level:** Sickle-shaped RBCs, anemia, pain crises

Significance: Demonstrates how single nucleotide change can cause severe genetic disorder.

4.15 Chromosomal Theory of Inheritance

Question: Who had proposed the chromosomal theory of inheritance?

Answer: Walter Sutton and Theodore Boveri (1902) proposed the Chromosomal Theory of Inheritance.

Background:

- By 1900, Mendel's laws were rediscovered
- Advances in microscopy allowed chromosome observation
- Chromosome behavior during meiosis was understood

Key Observations: Sutton and Boveri noted parallel behavior between chromosomes and Mendel's factors (genes):

Chromosomes	Genes (Mendel's Factors)
Occur in pairs	Occur in pairs
Segregate during meiosis	Segregate during gamete formation
One from each pair goes to each gamete	One allele from each pair goes to each gamete
Independent pairs assort independently	Independent gene pairs assort independently
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Theory Statement: "Genes are located on chromosomes and the behavior of chromosomes during meiosis accounts for Mendel's laws of inheritance."

Supporting Evidence:

- Thomas Hunt Morgan (1910): Experimental verification using Drosophila
- **Sex-linkage studies:** Proved genes are on specific chromosomes
- **Linkage mapping:** Showed genes on same chromosome travel together

Significance:

- Provided physical basis for Mendel's abstract "factors"
- United cytology with genetics
- Foundation for modern chromosome mapping and genomics

4.16 Autosomal Genetic Disorders

Question: Mention any two autosomal genetic disorders with their symptoms.

(1) Sickle Cell Anemia

Type: Autosomal recessive disorder

Genetic Basis:

• **Gene:** β-globin gene (chromosome 11)

• **Mutation:** Point mutation GAG → GUG

• **Inheritance:** Both parents must be carriers (Hb^A Hb^S)

Symptoms:

• **Anemia:** Due to rapid destruction of sickle cells

• Pain crises: Blocked blood vessels by sickle-shaped RBCs

• Organ damage: Heart, kidney, liver, spleen complications

• **Growth retardation:** Poor oxygen delivery

• Infections: Increased susceptibility due to spleen damage

• Stroke: Due to blocked cerebral blood vessels

Molecular Basis: Glutamic acid \rightarrow Valine substitution at 6th position of β -globin chain causes hemoglobin polymerization

(2) Phenylketonuria (PKU)

Type: Autosomal recessive disorder

Genetic Basis:

• **Gene:** Phenylalanine hydroxylase gene (chromosome 12)

• **Defect:** Enzyme deficiency or absence

• Inheritance: Both parents must be carriers

Symptoms:

• Mental retardation: Due to phenylalanine accumulation in brain

• Reduced pigmentation: Light skin and hair color

- Musty odor: Due to phenylpyruvic acid in urine
- Behavioral problems: Hyperactivity, seizures
- Growth retardation: Poor physical development
- **Eczema:** Skin problems

Biochemical Basis:

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Phenylalanine \rightarrow (Phenylalanine hydroxylase) \rightarrow Tyrosine \downarrow (Enzyme deficient) Phenylpyruvic acid + other toxic metabolites
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Management: Early detection through newborn screening and phenylalanine-restricted diet can prevent symptoms.

KEY FORMULAS AND CONCEPTS REFERENCE

Genetic Ratios:

- Monohybrid F₂: 3:1 (phenotypic), 1:2:1 (genotypic)
- Dihybrid F₂: 9:3:3:1 (phenotypic)
- Test cross: 1:1 (if heterozygous parent)
- Incomplete dominance: 1:2:1 (phenotypic = genotypic)

Gamete Types:

- Heterozygous for n loci: 2ⁿ gamete types
- AaBbCc produces: $2^3 = 8$ different gametes

Hardy-Weinberg Principle:

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$$p^2 + 2pq + q^2 = 1$$

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$$p + q = 1$$

Sex-linked Inheritance:

- X-linked recessive: More males affected
- Carrier females can transmit to sons
- Male to male transmission impossible

Answer Key Complete - All exercise problems solved with detailed explanations for NEET/JEE preparation.