Chapter 4: Principles of Inheritance and Variation

Exercises — Chapter-end Exercises

Q1 Mention the advantages of selecting pea plant for experiment by Mendel.

SOLUTION

Mendel selected garden pea (Pisum sativum) for his hybridisation experiments due to several advantageous features:

1. Pea plants have many distinguishable contrasting characters (7 pairs of traits such as tall/dwarf, round/wrinkled seeds, yellow/green seeds, etc.) that are easy to observe and score.
2. They are naturally self-pollinating, which ensures true-breeding (homozygous) lines are easy to maintain.
3. Cross-pollination can be performed easily by hand, allowing controlled hybridisation experiments.
4. They have a short generation time and produce a large number of offspring, enabling statistically significant data.
5. Plants are easy and inexpensive to grow and maintain in large numbers.
6. Traits are distinct with no intermediate forms (discontinuous variation), making scoring unambiguous.

Answer: Pea plants have clearly contrasting traits, are naturally self-pollinating (allowing true-breeding lines), permit easy cross-pollination, have a short generation time, and produce large numbers of offspring — all of which made them ideal for Mendel's experiments.

Q2 Differentiate between the following —
(a) Dominance and Recessive
(b) Homozygous and Heterozygous
(c) Monohybrid and Dihybrid.

SOLUTION

(a) Dominance and Recessive:
A dominant allele is one that expresses its phenotype even when only one copy (heterozygous condition) is present. In a pair of contrasting alleles, the dominant allele 'masks' the effect of the other allele. For example, tallness (T) is dominant over dwarfness (t) in pea plants — a plant with genotype Tt is tall.
A recessive allele is one whose expression is suppressed in the presence of the dominant allele. It is expressed only in the homozygous condition (tt). The recessive character reappears in the F_2 generation when both alleles are recessive.

(b) Homozygous and Heterozygous:
A homozygous organism has identical alleles for a given gene locus on both homologous chromosomes (e.g., TT or tt). Homozygous individuals breed true and produce only one type of gamete for that character.

A heterozygous organism has two different alleles at a given gene locus on homologous chromosomes (e.g., Tt). Such individuals do not breed true and produce two types of gametes (T and t) in equal proportions.

(c) Monohybrid and Dihybrid:
A monohybrid cross involves parents that differ in only one pair of contrasting characters (one gene locus). The F_2 ratio for a monohybrid cross is 3:1 (phenotypic) and 1:2:1 (genotypic). Example: TT × tt.
A dihybrid cross involves parents that differ in two pairs of contrasting characters (two gene loci). The F_2 phenotypic ratio is 9:3:3:1. Example: RRYY × rryy.

Answer: (a) Dominant allele expresses in heterozygous condition; recessive expresses only when homozygous. (b) Homozygous has identical alleles (TT/tt); heterozygous has dissimilar alleles (Tt). (c) Monohybrid cross involves one trait pair (F_2 ratio 3:1); dihybrid involves two trait pairs (F_2 ratio 9:3:3:1).

Q3 A diploid organism is heterozygous for 4 loci, how many types of gametes can be produced?

SOLUTION

For a diploid organism heterozygous at a single locus (e.g., Aa), two types of gametes are produced: A and a.

For each additional heterozygous locus, the number of gamete types doubles (assuming independent assortment).

The formula is: Number of gamete types = 2^n, where n = number of heterozygous loci.

Here, n = 4.

Number of gamete types = $2^4 = 16$.

Answer: 16 types of gametes ($2^4 = 16$, where n = 4 heterozygous loci).

Q4 Explain the Law of Dominance using a monohybrid cross.

SOLUTION

Law of Dominance states that: (i) characters are controlled by discrete units called factors (genes); (ii) factors occur in pairs; (iii) in a pair of dissimilar factors, one member (dominant) dominates the other (recessive) and is expressed.

Illustration using a monohybrid cross (tall × dwarf pea):

Parents: Tall (TT) × Dwarf (tt)
Gametes from tall parent: T only
Gametes from dwarf parent: t only

F_1 generation: All progeny are Tt (genotype) → All plants are Tall (phenotype).
Here, only the dominant character (tallness, T) is expressed; the recessive character

(dwarfness, t) is not seen, even though the t allele is present.

$F_1 \times F_1$ (Tt × Tt):
Gametes: T and t (from each parent)
F_2 genotypes: 1 TT : 2 Tt : 1 tt
F_2 phenotypes: 3 Tall : 1 Dwarf (phenotypic ratio 3:1)

The dominant character (tallness) is expressed in both TT and Tt plants; the recessive character (dwarfness) is expressed only in tt plants. The Law of Dominance explains why only one parental character is seen in F_1 and why both appear in F_2 in a 3:1 ratio.

Answer: The Law of Dominance states that in a pair of contrasting alleles, the dominant allele masks the recessive. In a monohybrid cross (TT × tt), all F_1 are Tt and phenotypically tall (dominant). In F_2 (Tt × Tt), the phenotypic ratio is 3 Tall : 1 Dwarf, demonstrating that T dominates t.

Q5 Define and design a test-cross.

SOLUTION

Definition: A test cross is a cross between an organism showing a dominant phenotype (whose genotype is to be determined — could be homozygous dominant or heterozygous) and the homozygous recessive parent. The offspring ratio of the test cross reveals the genotype of the organism under test.

Design of a test cross:

Suppose we want to determine whether a tall pea plant (dominant phenotype) is TT or Tt.

Case 1 — If the tall plant is homozygous dominant (TT):
TT × tt → all offspring are Tt (all tall).
Result: All offspring show dominant phenotype (tall). ∴ The plant is TT.

Case 2 — If the tall plant is heterozygous (Tt):
Tt × tt → 1 Tt (tall) : 1 tt (dwarf).
Result: 50% offspring are tall and 50% are dwarf. ∴ The plant is Tt.

Inference: If all offspring of the test cross show the dominant phenotype, the organism is homozygous dominant. If offspring are in a 1:1 ratio of dominant to recessive, the organism is heterozygous.

Answer: A test cross is a cross of an organism of unknown genotype (showing dominant phenotype) with a homozygous recessive. If all offspring are dominant → organism is homozygous dominant (TT). If offspring are 1 dominant : 1 recessive → organism is heterozygous (Tt).

Q6 Using a Punnett Square, workout 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.

SOLUTION

Let the gene locus have alleles A (dominant) and a (recessive).
Homozygous female — could be AA or aa. We consider the general case where the homozygous parent is AA (homozygous dominant) and the heterozygous male is Aa.

Cross: Female AA × Male Aa

Gametes from female (AA): A, A
Gametes from male (Aa): A, a

Punnett Square:

	A (male)	a (male)
A (female)	AA	Aa
A (female)	AA	Aa

Genotypes of offspring: 2 AA : 2 Aa → 1 AA : 1 Aa
Phenotypes: All offspring show the dominant phenotype (since both AA and Aa express the dominant character).
Phenotypic ratio: 100% dominant (all show dominant trait).

If instead the homozygous female is aa (recessive) and male is Aa:
Gametes from female (aa): a, a
Gametes from male (Aa): A, a

Punnett Square:

	A (male)	a (male)
a (female)	Aa	aa
a (female)	Aa	aa

Genotypes: 1 Aa : 1 aa
Phenotypes: 50% dominant (Aa) : 50% recessive (aa).
Phenotypic ratio: 1 dominant : 1 recessive.

Answer: If homozygous female is AA and male is Aa: all F_1 offspring are either AA or Aa — 100% show the dominant phenotype. If homozygous female is aa and male is Aa: F_1 offspring are Aa and aa in 1:1 ratio — 50% dominant and 50% recessive phenotype.

Q7 When a cross is made between tall plant with yellow seeds (TtYy) and tall plant with green seed (Ttyy), what proportions of phenotype in the offspring could be expected to be
(a) tall and green.
(b) dwarf and green.

SOLUTION

Cross: TtYy × Ttyy

Consider each gene independently:

For height (Tt × Tt):
Offspring ratios: 1 TT : 2 Tt : 1 tt
Phenotype: 3 Tall (TT + Tt) : 1 Dwarf (tt)

For seed colour (Yy × yy):
Offspring ratios: 1 Yy : 1 yy
Phenotype: 1 Yellow (Yy) : 1 Green (yy)

Combined phenotypic expectations (using product rule, genes are on different chromosomes — independent assortment):
Total combinations = 4 × 2 = 8 parts (denominator = 8 for simplicity from 3:1 × 1:1).
But expressing as fractions: Tall = 3/4, Dwarf = 1/4, Yellow = 1/2, Green = 1/2.

(a) Tall and green: P(Tall) × P(Green) = 3/4 × 1/2 = 3/8

(b) Dwarf and green: P(Dwarf) × P(Green) = 1/4 × 1/2 = 1/8

Answer: (a) Tall and green: 3/8 of offspring. (b) Dwarf and green: 1/8 of offspring.

Q8 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?

SOLUTION

In a typical dihybrid cross (AaBb × AaBb) when the two genes are located on different chromosomes (independent assortment), the F_1 phenotypic ratio is 9:3:3:1.

However, when the two loci are linked — i.e., located on the same chromosome — they tend to be inherited together rather than independently.

If the two genes are tightly linked (complete linkage), no recombination occurs between them. The parental gene combinations (AB and ab) are inherited together. The offspring will show only two phenotypic classes resembling the two parental types, in approximately a 3:1 ratio (dominant-dominant : recessive-recessive), instead of four classes.

For example, if both dominant alleles are on one chromosome (AB/ab — cis configuration):
Gametes produced mainly: AB and ab (parental types), with very few or no recombinant types (Ab and aB).
F_1 phenotypes: mostly dominant for both traits (AB) and recessive for both traits (ab) — in ratio approximately 3 (dominant-dominant) : 1 (recessive-recessive) rather than the expected

9:3:3:1.

If the genes show partial linkage, some recombination occurs, giving all four phenotypic classes but in ratios that deviate from 9:3:3:1 — the parental type classes are more frequent than the recombinant type classes.

Morgan's experiments with Drosophila demonstrated this deviation from Mendel's Law of Independent Assortment when genes are linked.

Answer: When two loci are linked, the F_1 phenotypic ratio deviates from the expected 9:3:3:1. With complete linkage, only two parental phenotypic classes appear (in ~3:1 ratio). With partial linkage, all four classes appear but parental combinations are far more frequent than recombinant combinations.

Q9 Briefly mention the contribution of T.H. Morgan in genetics.

SOLUTION

Thomas Hunt Morgan made several landmark contributions to genetics:

1. Chromosomal Theory of Inheritance (Experimental Verification): Morgan provided the experimental evidence that genes are located on chromosomes, confirming the chromosomal theory proposed by Sutton and Boveri.

2. Sex-linked Inheritance: Morgan worked with Drosophila melanogaster and demonstrated that certain genes are sex-linked — carried on the sex chromosomes (X chromosome). He discovered the inheritance of white eye colour in Drosophila as an X-linked trait.

3. Linkage: Morgan discovered the concept of linkage — the physical association of genes located on the same chromosome. Linked genes do not assort independently, causing deviation from the 9:3:3:1 ratio expected in a dihybrid cross.

4. Recombination: Morgan coined the term 'recombination' to describe the generation of new, non-parental gene combinations due to crossing over during meiosis.

5. Genetic Maps: Morgan and his student Alfred Sturtevant used the frequency of recombination between gene pairs as a measure of the distance between genes and constructed the first genetic (linkage) maps of chromosomes.

6. Drosophila as a model organism: Morgan established Drosophila melanogaster as an ideal genetic model organism due to its short life cycle, large number of offspring, easily distinguishable sexes, and many heritable variations visible under a microscope.

Answer: Morgan provided experimental evidence for the chromosomal theory of inheritance using Drosophila. He discovered sex-linked inheritance, coined the terms 'linkage' and 'recombination', and along with Sturtevant, constructed the first genetic maps based on recombination frequencies.

Q10 What is pedigree analysis? Suggest how such an analysis can be useful.

SOLUTION

Pedigree Analysis is the study of the inheritance pattern of a particular trait, abnormality or disease across several generations of a family, represented diagrammatically as a family tree.

In pedigree analysis, standard symbols are used: squares represent males, circles represent females, horizontal lines between a male and female symbol indicate mating, vertical lines connect parents to offspring, and filled symbols indicate affected individuals.

Usefulness of Pedigree Analysis:

1. Tracing Inheritance Pattern: It helps determine whether a trait is dominant or recessive, and whether it is autosomal or sex-linked (X-linked).

2. Genetic Counselling: Pedigree analysis is a powerful tool in genetic counselling. By studying the family history, it is possible to predict the probability of a genetic disorder appearing in future offspring.

3. Identification of Carriers: It helps identify individuals who are carriers of a recessive trait (heterozygous) even though they do not show the phenotype themselves.

4. Understanding Mendelian Disorders: Since controlled crosses cannot be performed in humans (as in pea plants), pedigree analysis serves as an alternative method to study the inheritance of Mendelian disorders such as haemophilia, sickle-cell anaemia, colour blindness, and phenylketonuria.

5. Medical Diagnosis and Prevention: Pedigree analysis helps predict risk of genetic diseases in families where such disorders have previously occurred, allowing preventive measures or early diagnosis.

Answer: Pedigree analysis is the diagrammatic representation of inheritance of a trait across family generations. It is useful for determining if a trait is dominant/recessive and autosomal/sex-linked, identifying carriers, predicting risk in future offspring, and providing genetic counselling.

Q11 How is sex determined in human beings?

SOLUTION

Sex determination in human beings is of the XY type.

Chromosomal Basis:
Humans have 46 chromosomes (23 pairs). Of these, 22 pairs are autosomes (same in both sexes) and one pair are sex chromosomes.
Females have two X chromosomes (XX) — they are homogametic.
Males have one X and one Y chromosome (XY) — they are heterogametic.

Mechanism:
During spermatogenesis in males, two types of sperms are produced in equal proportions:

- 50% of sperms carry the X chromosome (besides autosomes)
- 50% of sperms carry the Y chromosome (besides autosomes)

Females produce only one type of ovum: each ovum carries one X chromosome.

At fertilisation:
- If the ovum is fertilised by an X-bearing sperm → XX zygote → female offspring.
- If the ovum is fertilised by a Y-bearing sperm → XY zygote → male offspring.

Since there is an equal probability of fertilisation by an X-bearing or Y-bearing sperm, the expected sex ratio is 1:1 (50% male : 50% female) in each pregnancy.

It is the genetic makeup of the sperm (X or Y) that determines the sex of the child — NOT the ovum or the mother. Therefore, blaming women for the sex of the child is scientifically incorrect and socially unjust.

Answer: Sex in humans is determined by the XY mechanism. Females are XX (homogametic); males are XY (heterogametic). Males produce two types of sperms (X-bearing and Y-bearing) in equal proportion. Fertilisation of the egg by an X-bearing sperm gives a female (XX); by a Y-bearing sperm gives a male (XY). It is the sperm, not the egg, that determines the sex of the child.

Q12 A child has blood group O. If the father has blood group A and mother has blood group B, work out the genotypes of the parents and the possible genotypes of the other offsprings.

SOLUTION

ABO blood groups are controlled by the gene I, which has three alleles: I^A, I^B, and i.
I^A and I^B are both dominant over i; I^A and I^B show co-dominance with each other.

Blood group O genotype is always: ii (homozygous recessive).

The child has blood group O → genotype ii.
The child must have received one i allele from each parent.

Father has blood group A and must have contributed allele i to the child.
→ Father's genotype: I^Ai (heterozygous A)
(He cannot be I^AI^A because he contributed i to the O child.)

Mother has blood group B and must have contributed allele i to the child.
→ Mother's genotype: I^Bi (heterozygous B)
(She cannot be I^BI^B because she contributed i to the O child.)

Cross: I^Ai (father) × I^Bi (mother)

Possible gametes from father: I^A, i
Possible gametes from mother: I^B, i

Punnett Square:

	I^A	i
I^B	I^AI^B (AB)	I^Bi (A)

i Ii (B) ii (O)

Possible genotypes of offspring: II, Ii, Ii, ii
Possible phenotypes: AB, A, B, O — each in 1/4 proportion.

So the other offspring could have blood groups A, B, AB, or O.

Answer: Father's genotype: Ii (blood group A); Mother's genotype: Ii (blood group B). Possible genotypes of offspring: II (AB), Ii (A), Ii (B), ii (O) — each in 1:1:1:1 ratio. All four blood groups are possible.

Q13 Explain the following terms with example
(a) Co-dominance
(b) Incomplete dominance

SOLUTION

(a) Co-dominance:
Co-dominance is a pattern of inheritance in which the F_1 heterozygote simultaneously expresses both parental phenotypes — neither allele is dominant or recessive, and both are expressed equally and independently.

Example: ABO Blood Groups in humans.
Blood group is controlled by three alleles of gene I: I^A, I^B, and i.
When both I^A and I^B alleles are present in an individual (genotype I^AI^B), both A-type and B-type sugar molecules are produced on the red blood cell surface, resulting in blood group AB.
Neither I^A nor I^B masks the other — both are fully expressed. This is co-dominance.

(b) Incomplete Dominance:
Incomplete dominance is a pattern of inheritance where neither allele is completely dominant over the other. The F_1 heterozygote shows a phenotype that is intermediate (blended) between the two parental phenotypes — though genotypically the alleles remain unchanged.

Example: Flower colour in Snapdragon (Antirrhinum sp. / dog flower).
When true-breeding red-flowered plants (RR) are crossed with true-breeding white-flowered plants (rr), the F_1 plants (Rr) have pink flowers — an intermediate phenotype.
On self-pollination of F_1 (Rr × Rr), the F_2 ratio is:
1 Red (RR) : 2 Pink (Rr) : 1 White (rr)
The phenotypic ratio (1:2:1) mirrors the genotypic ratio, unlike in complete dominance (3:1 phenotypic).
Here the R allele is not completely dominant over r, so one copy of R produces only enough pigment for a pink (intermediate) colour.

Answer: (a) Co-dominance: both alleles in a heterozygote are fully and simultaneously expressed. Example: II genotype gives AB blood group. (b) Incomplete dominance: neither allele fully dominates; the heterozygote shows an intermediate phenotype. Example: red (RR) × white (rr) snapdragon gives pink (Rr) F_1; F_2 ratio is 1 red : 2 pink : 1 white.

Q14 What is point mutation? Give one example.

SOLUTION

Point mutation is a type of mutation that involves a change (substitution, insertion, or deletion) in a single base pair (nucleotide) in the DNA sequence of a gene. Such a small change can alter the codon and consequently the amino acid specified, leading to a change in the protein product and potentially in the phenotype.

Example: Sickle-cell anaemia.
Sickle-cell anaemia is caused by a point mutation in the gene encoding the beta (β) globin chain of haemoglobin. A single base change in the sixth codon of the β-globin gene — from GAG (codes for Glutamic acid) to GUG (codes for Valine) — results in the substitution of Glutamic acid (Glu) by Valine (Val) at the sixth position of the β-globin chain.
This single amino acid change causes the haemoglobin molecules to polymerise under low oxygen conditions, distorting the red blood cells from their normal biconcave disc shape into an elongated sickle shape, causing sickle-cell anaemia.

Answer: Point mutation is a change in a single base pair of DNA. Example: sickle-cell anaemia — a single base substitution (GAG → GUG) in the β-globin gene causes replacement of Glutamic acid by Valine at position 6 of the β-globin chain, leading to abnormal haemoglobin and sickle-shaped RBCs.

Q15 Who had proposed the chromosomal theory of the inheritance?

SOLUTION

The Chromosomal Theory of Inheritance was proposed by Walter Sutton and Theodore Boveri in 1902.

Sutton and Boveri independently observed that the behaviour of chromosomes during meiosis is parallel to the behaviour of Mendel's 'factors' (genes). They noted that:
- Chromosomes occur in pairs, just as genes occur in pairs.
- Homologous chromosomes segregate during meiosis, similar to the segregation of alleles.
- Non-homologous chromosomes assort independently, just as different gene pairs assort independently.

Sutton united this knowledge of chromosomal segregation with Mendelian principles and called it the Chromosomal Theory of Inheritance.

Later, Thomas Hunt Morgan and his colleagues provided experimental verification of this theory through their work on Drosophila melanogaster.

Answer: The chromosomal theory of inheritance was proposed by Walter Sutton and Theodore Boveri (1902), who noted the parallel behaviour of chromosomes and Mendelian factors (genes) during meiosis. It was later experimentally verified by T.H. Morgan.

Q16 Mention any two autosomal genetic disorders with their symptoms.

SOLUTION

Two autosomal genetic disorders and their symptoms:

1. Sickle-cell Anaemia (Autosomal Recessive):
Caused by a point mutation in the β-globin gene (GAG → GUG), leading to substitution of Glutamic acid by Valine at the 6th position of the β-globin chain of haemoglobin.
Symptoms:
- Red blood cells (RBCs) become elongated, sickle-shaped under low oxygen conditions due to polymerisation of abnormal haemoglobin (HbS).
- Anaemia (reduced oxygen-carrying capacity of blood).
- Chronic fatigue and weakness.
- Episodes of severe pain (sickle-cell crisis) due to blockage of blood vessels by sickle-shaped cells.
- Susceptibility to infections.
Only homozygous individuals (Hb^sHb^s) show the diseased phenotype; heterozygous ($HbHb^s$) individuals are carriers (sickle-cell trait).

2. Phenylketonuria / PKU (Autosomal Recessive):
This is an inborn error of metabolism caused by a mutation in the gene coding for the enzyme phenylalanine hydroxylase.
Symptoms:
- The enzyme deficiency leads to inability to convert phenylalanine to tyrosine.
- Phenylalanine accumulates and is converted to phenylpyruvic acid and other derivatives.
- Accumulation of these substances in the brain results in mental retardation.
- Reduction in hair and skin pigmentation (since tyrosine is a precursor for melanin).
- Phenylpyruvic acid is excreted in urine due to poor kidney reabsorption.

Answer: 1. Sickle-cell anaemia (autosomal recessive): sickle-shaped RBCs, anaemia, pain crises, fatigue — caused by a point mutation in the β-globin gene (Hb^sHb^s genotype). 2. Phenylketonuria (autosomal recessive): mental retardation, reduced pigmentation — caused by deficiency of phenylalanine hydroxylase, leading to accumulation of phenylpyruvic acid.