Principles of Inheritance and Variation

Mendel's Laws and Chromosomal Theory

Heredity is the biological process by which characteristics are passed from parents to offspring through reproduction.

Variation refers to differences in traits among individuals of a species.

Mendel's Laws of Inheritance

Gregor Mendel conducted hybridisation experiments on garden pea plants (*Pisum sativum*) studying seven pairs of contrasting traits.

He observed that traits are controlled by factors (now called genes) that occur in pairs (alleles).

Monohybrid Cross: A cross between two plants differing in one trait, e.g., tall (T) and dwarf (t) stem height.

Steps in monohybrid cross include selection of contrasting plants, emasculation to prevent self-pollination, pollination, and observation of offspring.

F_1 generation hybrids (Tt) show dominant trait; F_2 generation shows a phenotypic ratio of 3 tall : 1 dwarf and genotypic ratio of 1 TT : 2 Tt : 1 tt.

Backcross: Crossing F_1 hybrid with a parent.

Test cross: Crossing F_1 hybrid with homozygous recessive to determine unknown genotype.

Mendel's Principles

Principle of Dominance

In heterozygous condition, one allele (dominant) masks the effect of the other (recessive).

Law of Segregation

Allelic pairs separate during gamete formation; each gamete receives one allele randomly.

Law of Independent Assortment

Genes for different traits assort independently during gamete formation, leading to new combinations in offspring.

Non-Mendelian Inheritance

Incomplete Dominance

Heterozygous offspring show intermediate phenotype, e.g., flower colour in Snapdragon.

Co-dominance

Both alleles express equally, e.g., ABO blood groups where alleles I^A and I^B are co-dominant.

Multiple Allelism

More than two alleles govern a trait in a population, e.g., ABO blood group alleles I^A, I^B, and i.

Pleiotropy

One gene influences multiple traits, e.g., a gene controlling seed texture also affects starch grain size in peas.

Chromosomal Theory of Inheritance

Proposed by Sutton and Boveri, it states that genes are located on chromosomes which segregate during meiosis.

Chromosomes occur in homologous pairs; genes are arranged linearly on chromosomes.

Thomas Hunt Morgan proved this theory using fruit flies (*Drosophila melanogaster*).

Linkage and Recombination

Linkage: Genes located close together on the same chromosome tend to be inherited together.

Recombination: Exchange of genetic material during meiosis producing new allele combinations.

Linked genes show deviation from the expected 9:3:3:1 dihybrid ratio.

Recombination frequency is used to map gene positions on chromosomes.

Solved Examples

Example 1

In pea plants, flowers may be axial (A) or terminal (a). Find the proportion of terminal flowers in the offspring of crosses:

(i) Aa x Aa

(ii) AA x Aa

Solution:

(i) Cross Aa x Aa:

Gametes from each parent: A and a

Offspring genotypes: AA, Aa, Aa, aa

Phenotypes: Axial (AA, Aa) and Terminal (aa)

Proportion terminal = 1/4 = 25%

(ii) Cross AA x Aa:

Gametes: AA parent produces A; Aa parent produces A and a

Offspring genotypes: AA, Aa

Phenotypes: All axial; no terminal flowers

Example 2

Show sex-linked inheritance of colour blindness when a colour-blind man (X^cY) marries a normal woman (XX).

Solution:

Man's gametes: X^c and Y

Woman's gametes: X and X

Possible offspring:

- XX^c (carrier daughter, normal vision)
- XY (normal son)

Thus, 50% daughters are carriers, 50% sons are normal.

Practice Set

Conceptual Questions

- **Level 1:** Define the Law of Segregation.
- **Level 2:** Explain the difference between incomplete dominance and co-dominance with examples.

Application-based Question

- **Level 3:** In a dihybrid cross of pea plants with traits seed shape (round R, wrinkled r) and seed colour (yellow Y, green y), what phenotypic ratio is expected in the F_2 generation? Explain the principle involved.

Answer Key

- **Level 1:** Law of Segregation states that alleles separate during gamete formation so that each gamete carries only one allele of each gene.
- **Level 2:** Incomplete dominance shows intermediate phenotype in heterozygotes (e.g., pink flowers from red and white), while co-dominance shows both alleles expressed equally (e.g., AB blood group).
- **Level 3:** Phenotypic ratio is 9 round yellow : 3 round green : 3 wrinkled yellow : 1 wrinkled green. This is due to the Law of Independent Assortment where genes for seed shape and colour assort independently.

Sex Determination and Chromosomal Disorder

Sex Determination

Sex determination is the process by which the sex of an organism is established at fertilisation.

Autosomes are chromosomes other than sex chromosomes; they carry genes for body traits.

Sex chromosomes determine the sex; examples include X and Y chromosomes in humans.

Mechanisms of Sex Determination

Chromosomal mechanisms:

- XX-XO: Male is XO (heterogametic), female XX (homogametic), e.g., grasshoppers.
- XX-XY: Male XY (heterogametic), female XX (homogametic), e.g., humans, Drosophila.
- ZZ-ZW: Male ZZ (homogametic), female ZW (heterogametic), e.g., birds.
- ZO-ZZ: Female ZO, male ZZ, e.g., some insects.

Environmental sex determination: Sex determined by environmental factors like temperature, e.g., turtles.

Genetic balance mechanism: Sex determined by ratio of X chromosomes to autosomes, e.g., Drosophila.

Cytoplasmic sex determination: Determined by cytoplasmic factors, e.g., some bacteria.

Sex Determination in Humans

- Humans have 23 pairs of chromosomes: 22 autosomes and 1 pair of sex chromosomes.
- Females have XX; males have XY chromosomes.
- Males produce two types of sperm (X or Y chromosome); females produce eggs with X chromosome only.
- Sex of offspring depends on whether sperm carrying X or Y fertilises the egg.

Genetic Disorders

Genetic disorders are caused by mutations or chromosomal abnormalities.

Mendelian Disorders

Caused by mutation in a single gene; can be dominant or recessive.

Examples: Haemophilia, Cystic fibrosis, Sickle cell anaemia, Colour blindness, Phenylketonuria, Thalassaemia.

Colour Blindness

Recessive sex-linked trait where eyes cannot distinguish red and green.

Carried on X chromosome; females need two copies to express, males only one.

Haemophilia

Sex-linked recessive disorder affecting blood clotting.

Carriers are heterozygous females; males with the gene show disease.

Chromosomal Disorders

Caused by changes in chromosome number or structure.

Aneuploidy: Gain or loss of chromosomes.

- Nullisomy (2n-2): Loss of a homologous pair.
- Monosomy (2n-1): Loss of one chromosome.
- Trisomy (2n+1): Extra chromosome.
- Tetrasomy (2n+2): Two extra chromosomes.

Polyploidy (Euploidy): Increase in sets of chromosomes beyond diploid.

- Triploids (3n), tetraploids (4n), etc.
- Autopolyploidy: Multiple sets of same genome.
- Allopolyploidy: Sets from different species.

Chromosomal aberrations include deletion, inversion, duplication, and translocation.

Examples of Chromosomal Disorders

Down's Syndrome: Trisomy 21; features include short stature, flat face, mental retardation.

Klinefelter's Syndrome: Males with extra X chromosome (XXY); features include sterility, breast development.

Turner's Syndrome: Females with single X chromosome (XO); features include sterility, dwarfism.

Solved Examples

Example 3

Show the inheritance pattern of colour blindness when a colour-blind man (X^cY) marries a normal woman (XX).

Solution:

Man's gametes: X^c, Y

Woman's gametes: X, X

Offspring genotypes:

- XX^c (carrier daughter)
- XY (normal son)

50% daughters are carriers; 50% sons are normal.

Practice Set

Conceptual Questions

- **Level 1:** What is the difference between autosomes and sex chromosomes?
- **Level 2:** Explain the chromosomal basis of sex determination in humans.

Application-based Question

- **Level 3:** Describe the genetic cause and features of Down's syndrome.

Answer Key

- **Level 1:** Autosomes are chromosomes that determine body traits; sex chromosomes determine the sex of an organism.
- **Level 2:** Humans have 23 pairs of chromosomes; females have XX, males XY. Males produce sperm with X or Y chromosome; fertilisation determines sex.
- **Level 3:** Down's syndrome is caused by trisomy of chromosome 21 (extra copy). Features include short stature, flat face, mental retardation, and heart defects.

Quick Reference Table

Common Mistakes and Misconceptions

Glossary