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Introduction

Overview

- Genetics studies inheritance and variation of traits from parents to offspring.
- Inheritance: transfer of traits from parent to progeny; basis of heredity.
- Variation: differences between progeny and parents.
- Early agriculturists (8000-1000 BC) used natural variations in sexual reproduction for selective breeding.
- Domesticated varieties developed by crossing wild plants and animals (e.g., Sahiwal cows from wild ancestors).
- *Gregor Johann Mendel*: Father of genetics.
- William Bateson coined the term "genetics."

Memory Aids

- Mendel = Father of Genetics.
- Bateson = Term "Genetics."
- Selective breeding exploits natural variation.

Quick Checkpoints

- What is the difference between inheritance and variation?
- Who is known as the Father of Genetics?
- How did early agriculturists use variation?

Mendel's Laws of Inheritance

Key Points

- *Gregor Mendel* studied inheritance using garden pea (*Pisum sativum*) from 1856-1863.
- Studied traits with two contrasting forms (e.g., tall/dwarf, yellow/green seeds).
- Used true-breeding pea lines that show stable traits after self-pollination.

- Selected 14 true-breeding varieties differing in one contrasting trait each.
- Reasons for choosing pea plant:
  - Annual plant with short 2-3 month life cycle.
  - Many contrasting traits available.
  - Natural self-pollination present.
  - Artificial cross-pollination possible for hybridisation.
  - Easy to cultivate.
  - Produces many seeds.

Table : Characters studied by Mendel

	Trait studied	Dominant	Recessive
(i)	Plant height	Tall (T)	Dwarf (t)
(ii)	Flower position	Axillary (A)	Terminal (a)
(iii)	Pod colour	Green (G) or (Y)	Yellow (g) or (y)
(iv)	Pod shape	Full or Inflated (I) or (C)	Constricted (i) or (c)
(v)	Flower colour	Violet (V) or (W)	White (v) or (w)
(vi)	Seed shape	Round (R) or (W)	Wrinkled (r) or (w)
(vii)	Seed colour	Yellow (Y) or (G)	Green (y) or (g)

Memory Aids

- Pea plant traits: contrasting and easy to study.
- True-breeding lines ensure trait stability.
- Short life cycle speeds experiments.

Quick Checkpoints

- Why was *Pisum sativum* chosen for Mendel's experiments?
- What is a true-breeding line?
- What traits did Mendel study in peas?

Inheritance of One Gene

Observations and Results

- Mendel crossed tall and dwarf pea plants to study inheritance of one gene.
- Seeds from this cross grew into F□9 generation (first hybrid generation).
- All F□9 plants were tall, resembling one parent; dwarf trait was not seen.
- Self-pollination of F□9 tall plants produced F□2 generation with both tall and dwarf plants.
- In F□2, 3/4 plants were tall and 1/4 were dwarf.
- Traits showed no blending; offspring were either tall or dwarf.
- Similar results for other traits: F□9 resembled one parent, F□2 showed both traits in 3:1 ratio.

Memory Aids

- F□9 = first hybrid, shows dominant trait only.
- F□2 = second generation, shows 3:1 dominant to recessive ratio.
- No blending means traits remain distinct.

Quick Checkpoints

- What trait appears in F□9 generation?
- What is the phenotypic ratio in F□2 generation?
- Does blending occur in these traits?

Genetics Terminology

Key Terms

- **Alleles:** Different forms of the same gene.
- **Homozygous:** Organism with identical alleles (e.g., TT, tt).
- **Heterozygous:** Organism with different alleles (e.g., Tt).

- **Monohybrid cross:** Cross between homozygous tall (TT) and dwarf (tt) producing heterozygous tall (Tt).
- **Dominant allele:** Expresses in homozygous or heterozygous state (e.g., T for tall).
- **Recessive allele:** Expresses only in homozygous state (e.g., t for dwarf).
- **Phenotype:** Observable traits influenced by genotype and environment.
- **Genotype:** Genetic makeup of an organism for a trait.

Memory Aids

- Dominant allele masks recessive in heterozygotes.
- Homozygous = same alleles; heterozygous = different alleles.
- Phenotype = what you see; genotype = genetic code.

Quick Checkpoints

- Define homozygous and heterozygous.
- What is a monohybrid cross?
- Difference between dominant and recessive alleles?

Punnett Square Method

Concept and Usage

- Developed by Reginald C. Punnett to predict offspring genotypes.
- Graphical tool to calculate probability of genotypes in genetic crosses.
- **Test cross:** Cross between unknown genotype and recessive parent to determine genotype.
- Helps analyze progeny to predict genotype of dominant phenotype organism.

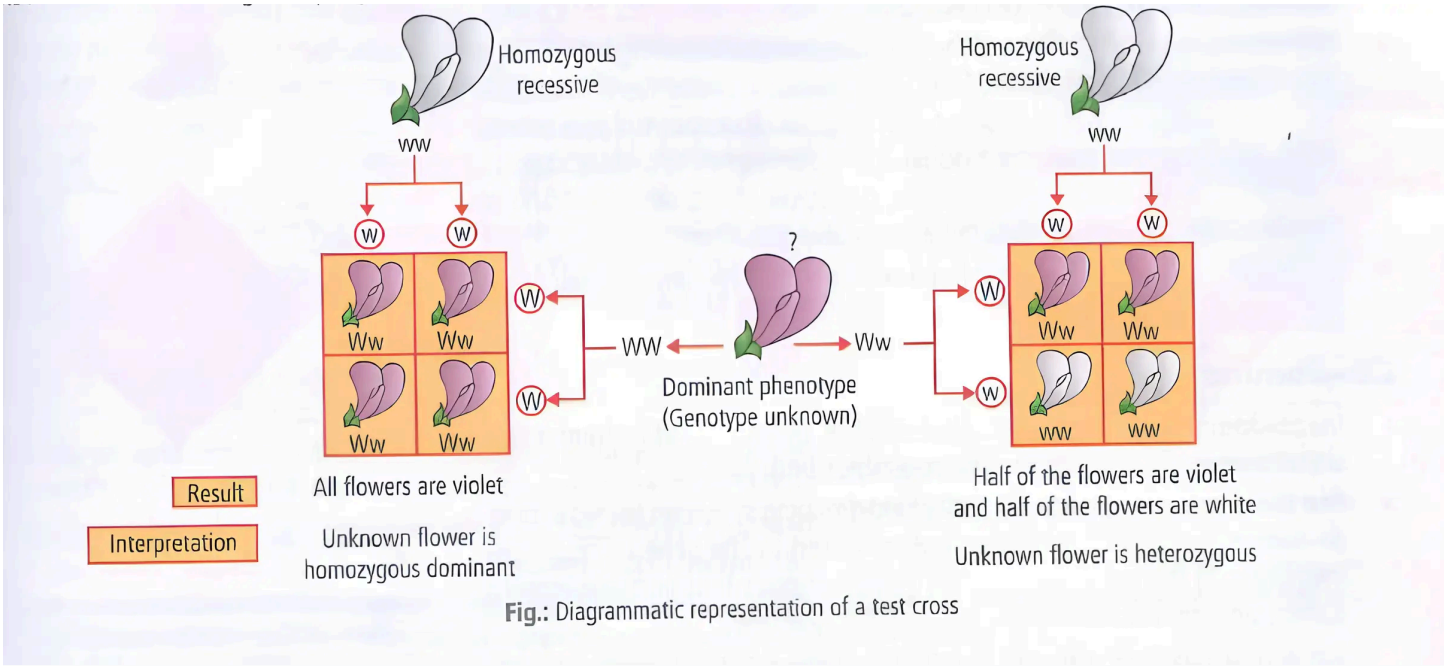


Fig.: Diagrammatic representation of a test cross

Memory Aids

- Test cross uses recessive parent to reveal unknown genotype.
- Punnett square = probability grid for offspring genotypes.

Quick Checkpoints

- What is the purpose of a Punnett square?
- How does a test cross help determine genotype?

Laws of Inheritance

Principles by Mendel

- First Law: Law of Dominance - dominant allele expresses in heterozygote.
- Second Law: Law of Segregation - alleles separate during gamete formation.
- Traits do not blend; recessive traits reappear in F□2 generation.

Memory Aids

- Dominance: one allele masks the other.
- Segregation: alleles separate into different gametes.

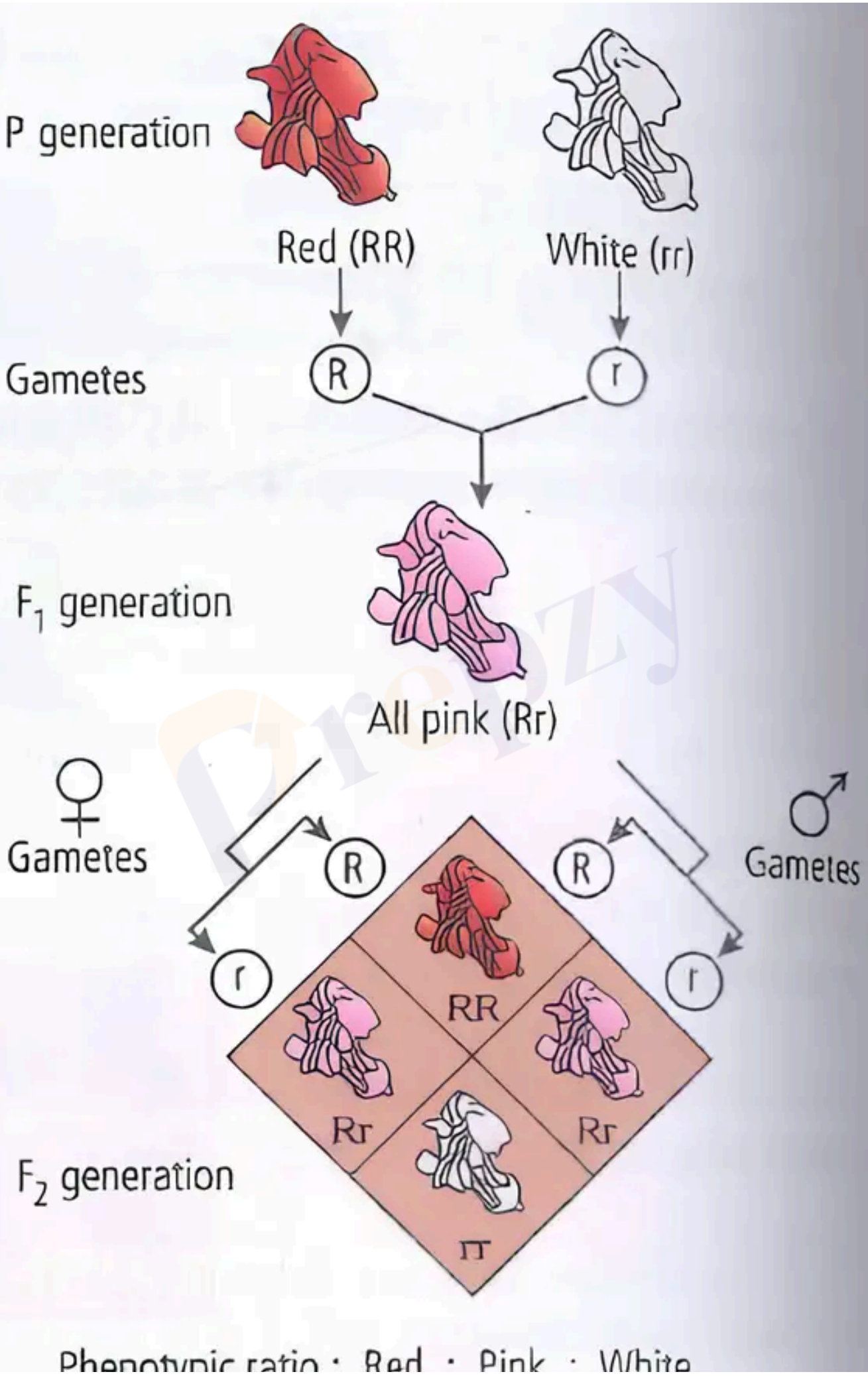
Quick Checkpoints

- What does the Law of Dominance state?
- Explain the Law of Segregation.
- Why do recessive traits reappear in F□2?

Incomplete Dominance

Definition and Example

- Neither allele is dominant; heterozygote shows intermediate phenotype.
- Example: Flower color in *Antirrhinum* (snapdragon).
- Cross of red (RR) and white (rr) flowers produces pink (Rr) flowers in F□9.
- F□2 generation shows 1 red : 2 pink : 1 white ratio in both genotype and phenotype.



Phenotypic ratio : Red : Pink : White

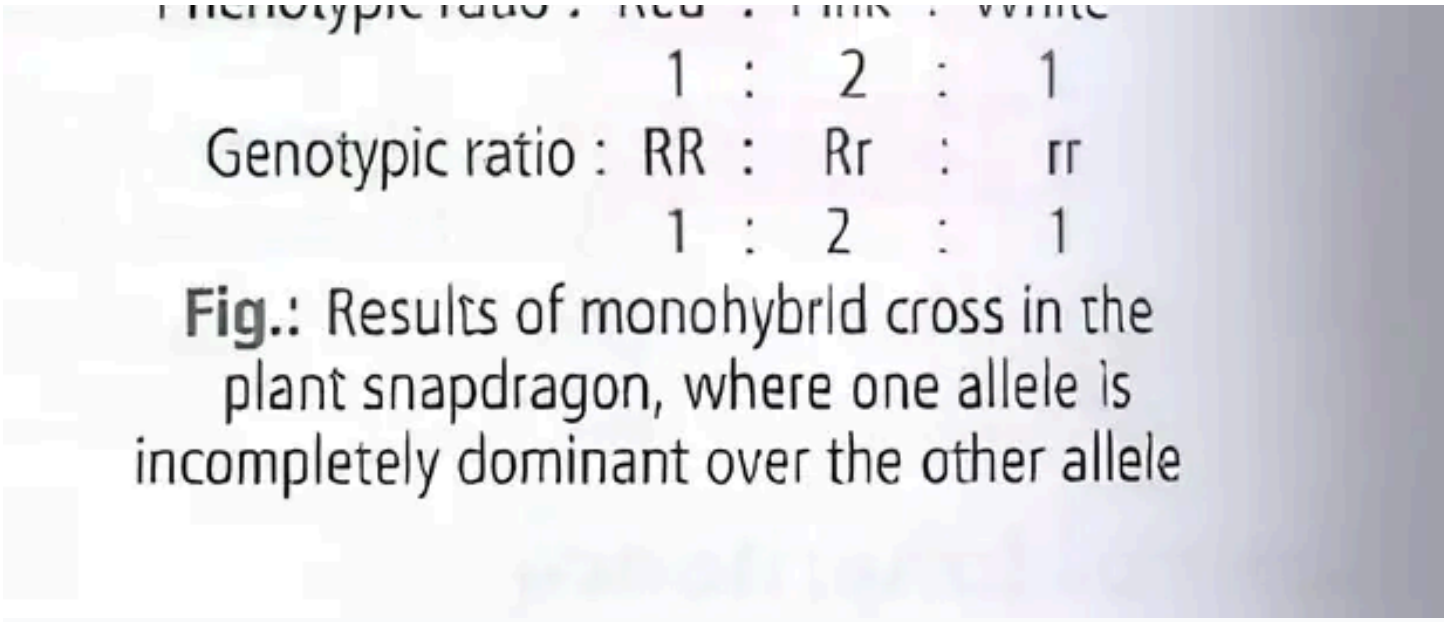


Fig.: Results of monohybrid cross in the plant snapdragon, where one allele is incompletely dominant over the other allele

Memory Aids

- Incomplete dominance = blend of traits in heterozygote.
- Phenotypic ratio equals genotypic ratio in incomplete dominance.

Quick Checkpoints

- What is incomplete dominance?
- How does flower color inheritance in snapdragon illustrate this?
- What is the phenotypic ratio in F□2 generation?

Co-dominance

Definition and Example

- Both alleles express simultaneously in heterozygote.
- Example: ABO blood group in humans controlled by gene with alleles I^A, I^B, and i.
- I^A and I^B are dominant over i; when together, both express (blood group AB).
- Multiple alleles: more than two alleles exist in population but individual has only two.
- Single gene can have multiple effects (pleiotropy), e.g., starch synthesis in pea seeds.

Table : Genetic basis of blood groups in human population

Allele from Parent 1	Allele from Parent 2	Genotype of offspring	Blood types of offspring
I^A	I^A	$I^A I^A$	A
I^A	I^B	$I^A I^B$	AB
I^A	i	$I^A i$	A
I^B	I^B	$I^B I^B$	B
I^B	i	$I^B i$	B
i	i	ii	O

Memory Aids

- Co-dominance = both alleles visible in phenotype.
- Multiple alleles exist in population, not in individual.
- Dominance depends on gene product and phenotype.

Quick Checkpoints

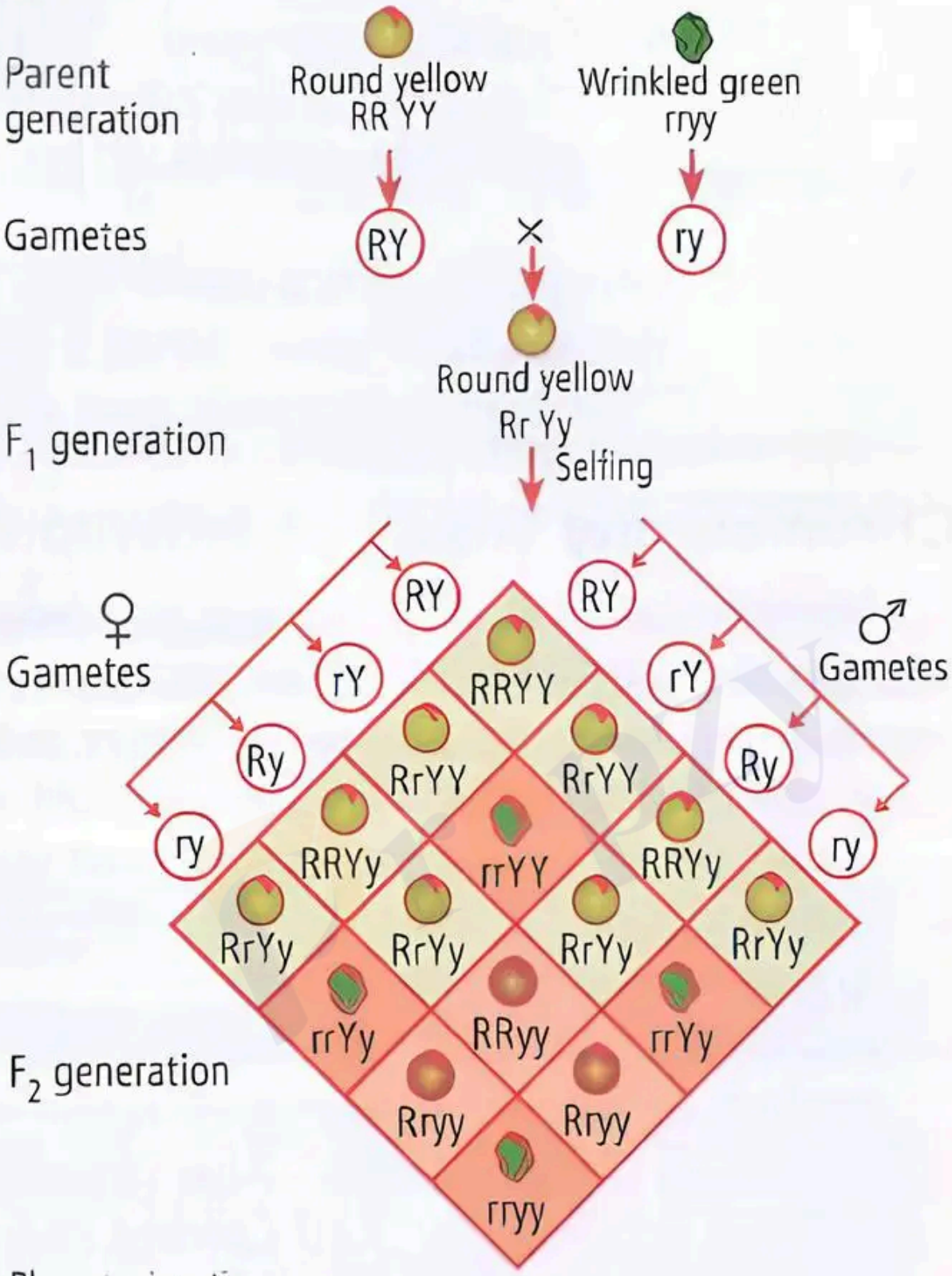
- What is co-dominance?
- Explain ABO blood group inheritance.
- What is multiple alleles?

Law of Independent Assortment

Explanation

- Mendel crossed pea plants differing in two traits: seed colour (yellow dominant over green) and seed shape (round dominant over wrinkled).
- Parental genotypes: RRYY (round yellow) × rryy (wrinkled green).

- F1 generation: All RrYy (round yellow) from RY and ry gametes.
- F2 generation phenotypic ratio: 9 round yellow : 3 wrinkled yellow : 3 round green : 1 wrinkled green.
- Segregation of one gene pair is independent of the other pair.
- Segregation of R and r gametes is independent of Y and y gametes.
- Applicable only to genes on different chromosomes or far apart on the same chromosome.



Phenotypic ratio :

Round yellow	Round green	Wrinkled yellow	Wrinkled green
9	3	3	1

Fig.: Result of a dihybrid cross where the two parents differed in two pairs of contrasting traits : seed colour and seed shape

Memory Aids

- Independent segregation = genes assort independently if on different chromosomes or far apart.
- Phenotypic ratio 9:3:3:1 in dihybrid cross.
- Use symbols: Y/y for seed colour, R/r for seed shape.

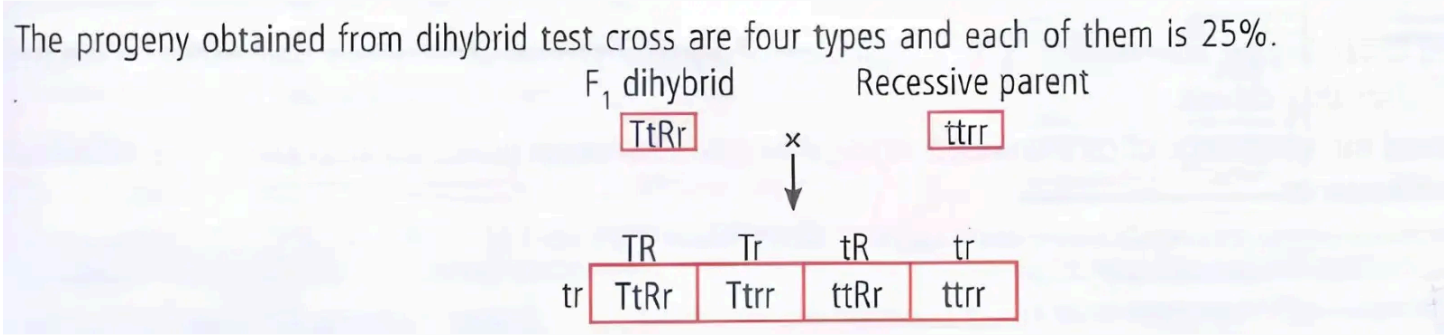
Quick Checkpoints

- What is the phenotypic ratio in a dihybrid cross?
- When does independent assortment apply?
- What are the parental genotypes in Mendel's dihybrid cross?

Dihybrid Test Cross

Explanation

- Cross between F1 dihybrid (TtRr) and recessive parent (ttrr).
- F1 produces four gametes: TR, Tr, tR, tr; recessive parent produces tr only.
- Offspring genotypes: TtRr, Ttrr, ttRr, ttrr in equal proportions.
- Phenotypic ratio: 1 : 1 : 1 : 1.



Memory Aids

- Test cross reveals genotype by crossing with recessive homozygote.
- Dihybrid test cross ratio always 1:1:1:1.

Quick Checkpoints

- What gametes does an F1 dihybrid produce?
- What is the phenotypic ratio in a dihybrid test cross?
- Why is the recessive parent used in test cross?

Intergenic Interaction

Explanation

- Effect of one gene modified by another non-allelic gene at different locus.
- Types:
  - Complementary genes: Two genes produce new trait together; F2 ratio 9:7.
  - Supplementary genes: One gene’s effect depends on presence of another; F2 ratio 9:3:4.
  - Epistasis: One gene masks expression of another.
  - Pleiotropy: One gene affects multiple phenotypes.

Memory Aids

- Complementary = both needed for trait.
- Supplementary = one gene supports another.
- Epistasis = gene masking.
- Pleiotropy = one gene, many effects.

Quick Checkpoints

- What is complementary gene interaction?
- Define epistasis.
- What is pleiotropy?

Chromosomal Theory of Inheritance

Explanation

- Mendel’s work initially unrecognized due to communication, acceptance, and lack of physical proof.
- Rediscovered in 1900 by Correns, de Vries, and von Tschermak.
- Theory proposed by Sutton and Boveri in 1902; proved by T.H. Morgan in 1933.
- Chromosomes behave like Mendelian factors during meiosis.
- Key points:
  - Sperm and egg contribute equally.
  - Chromosomes carry hereditary traits.
  - Chromosomes retain number, structure, individuality.
  - Chromosomes and genes occur in pairs in diploid cells.
  - Alleles located on homologous chromosomes.
  - Chromosome pairs align independently during metaphase I.
  - Chromosome segregation explains gene segregation.
- Chromosomal explanation of law of segregation: Homologous chromosomes separate in meiosis I; chromatids separate in meiosis II.

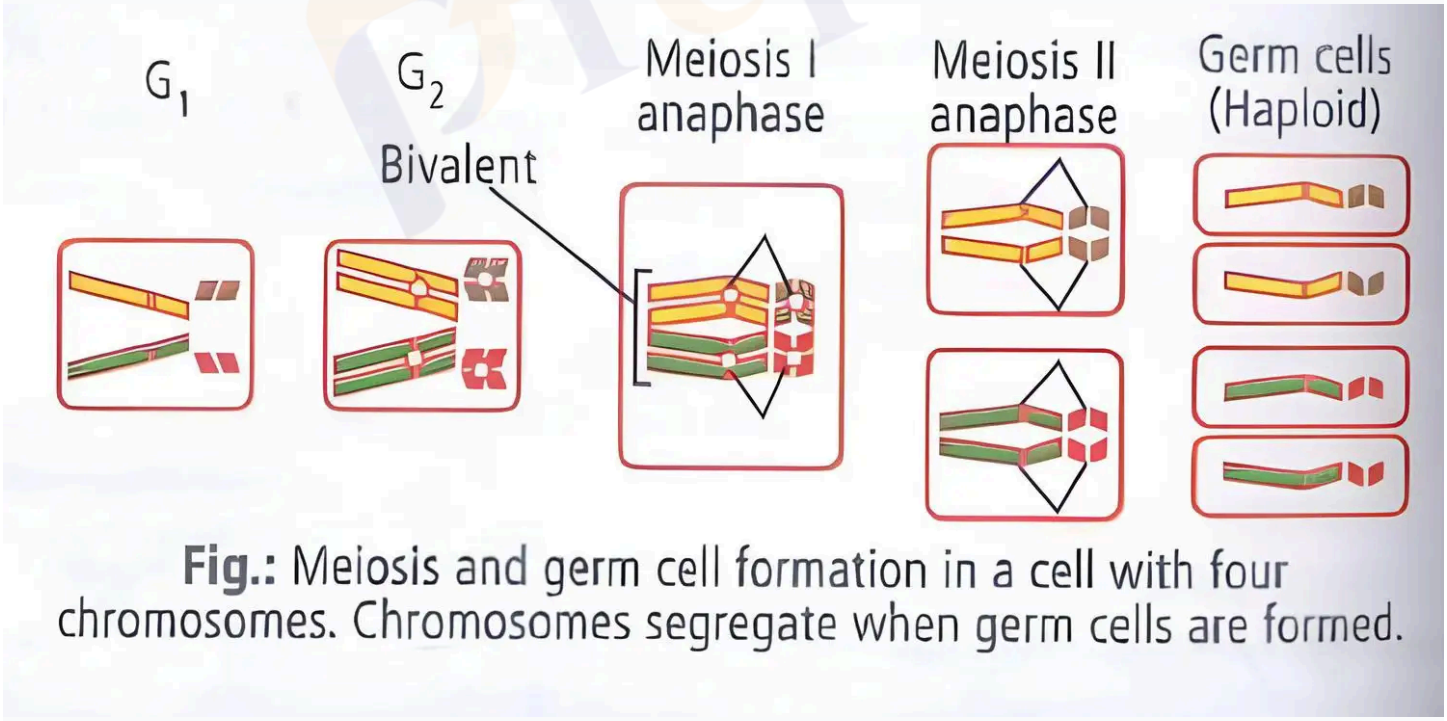


Fig.: Meiosis and germ cell formation in a cell with four chromosomes. Chromosomes segregate when germ cells are formed.

- Morgan used *Drosophila melanogaster* for genetic studies due to:
  - Easy lab culture.
  - Short life cycle (~2 weeks).

- Large progeny numbers.
- Distinct male and female flies.
- Visible hereditary variations.

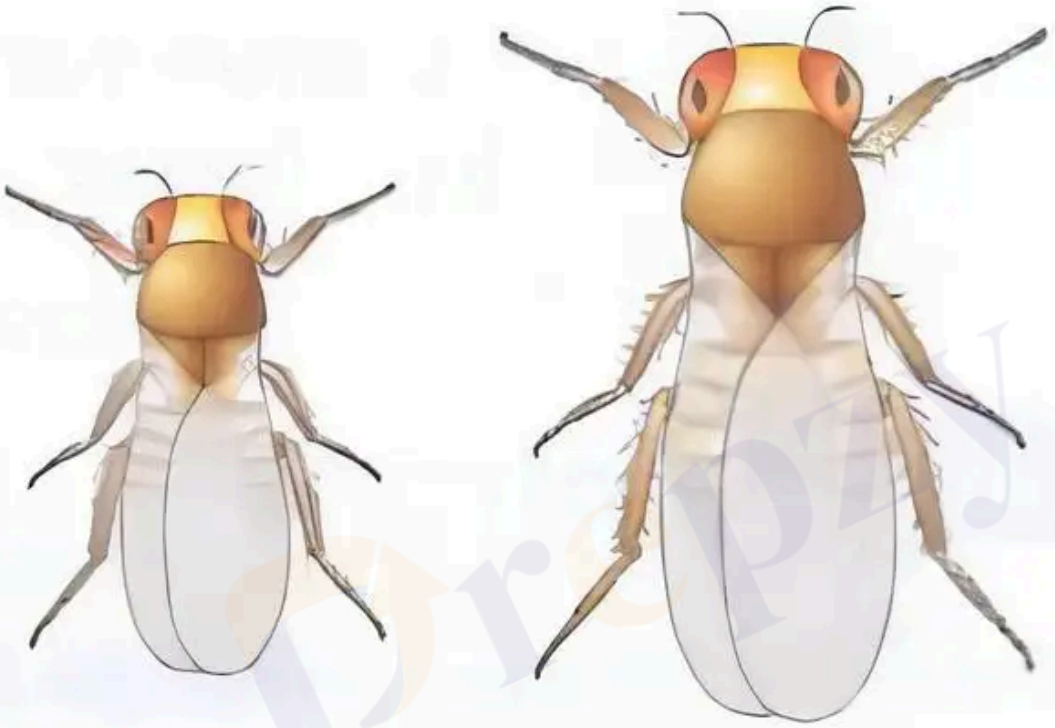


Fig.: *Drosophila melanogaster*
(a) Male (b) Female

Memory Aids

- Chromosomes = carriers of genes.
- Segregation explained by chromosome separation.
- *Drosophila* ideal for genetics due to fast breeding and visible traits.

Quick Checkpoints

- Who proposed the chromosomal theory of inheritance?
- How does chromosome behavior explain Mendel's laws?
- Why is *Drosophila melanogaster* used in genetics?

Linkage and Recombination

Explanation

- Morgan's dihybrid crosses in *Drosophila* showed deviation from 9:3:3:1 ratio.
- Genes on same chromosome tend to be inherited together (linkage).
- Linkage: physical association of genes on a chromosome.
- Recombination: production of non-parental gene combinations.
- Linkage groups = genes on same chromosome; number equals haploid chromosome number.
- Complete linkage: no crossing over, genes inherited together.
- Incomplete linkage: crossing over occurs, producing parental and recombinant types.
- Recombination frequency indicates gene distance.
- Example: white and yellow genes tightly linked (1.3% recombination); white and miniature wing loosely linked (37.2%).
- Sturtevant used recombination frequency to map gene positions.

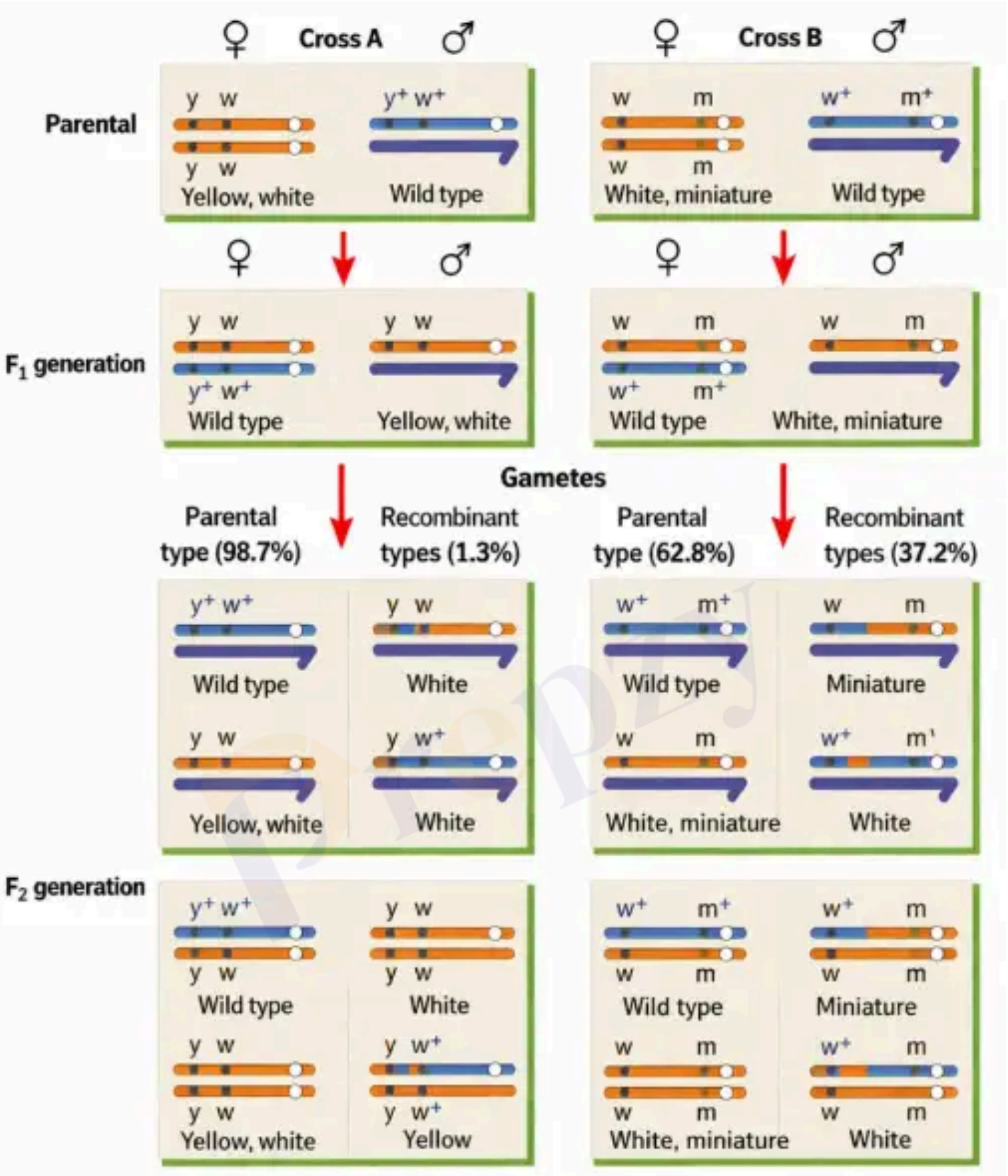


Fig.: Linkage: Results of two dihybrid crosses conducted by Morgan. Cross A shows crossing between genes y and w; Cross B shows crossing between genes w and m. Here, dominant wild type alleles are represented with (+) sign in superscript

Note: The strength of linkage between y and w is higher than w and m.

Memory Aids

- Linkage = genes close on chromosome inherited together.
- Recombination frequency = measure of gene distance.
- Complete linkage = no recombination; incomplete linkage = some recombination.

Quick Checkpoints

- What causes deviation from 9:3:3:1 ratio in dihybrid crosses?
- Define linkage and recombination.
- How is recombination frequency used in gene mapping?

Polygenic Inheritance

Explanation

- Inheritance controlled by three or more genes is polygenic inheritance.
- Phenotype reflects additive effect of each allele.
- Examples: human skin colour, height, wheat kernel colour.
- Human skin colour depends on melanin pigment quantity controlled by three gene pairs (A, B, C).
- Genotype AABBCC = darkest skin; all recessive alleles = lightest skin; mixed alleles = intermediate skin colour.
- Thomas Hunt Morgan's crosses show gene linkage on chromosomes (genes y, w, m).

Memory Aids

- Polygenic = many genes add up effects.
- Skin colour = sum of dominant alleles.
- More dominant alleles = darker skin.

Quick Checkpoints

- What defines polygenic inheritance?
- How does allele contribution affect phenotype?
- Which genes control human skin colour?
- What is the effect of dominant vs recessive alleles on skin colour?

Pleiotropy

Explanation

- Single gene influences multiple traits.
- Gene affects inter-related metabolic pathways producing different phenotypes.
- Example: Phenylketonuria caused by mutation in phenylalanine hydroxylase gene.
- Results in mental retardation and reduced hair and skin pigmentation.

Memory Aids

- Pleiotropy = one gene, many effects.
- Phenylketonuria = single gene mutation, multiple symptoms.

Quick Checkpoints

- What is pleiotropy?
- How does one gene affect multiple traits?
- What causes phenylketonuria?
- What are the symptoms of phenylketonuria?

Sex Determination

Overview

- Sex determination establishes male and female distinction in species.
- Determined by sex chromosomes (allosomes): X and Y chromosomes.
- Non-sex chromosomes are autosomes.
- Henking discovered X-body in 50% of sperm in some insects.

Memory Aids

- Sex chromosomes = Allosomes (X & Y).
- Autosomes = all other chromosomes.
- Henking's X-body = early sex chromosome discovery.

Quick Checkpoints

- What are sex chromosomes?
- Difference between autosomes and allosomes?
- Who discovered the X-body?

Types of Chromosomal Sex Determination

XX–XY type

- Female: homogametic (XX), produces one type of gamete (X).
- Male: heterogametic (XY), produces two types of gametes (X = gynosperm, Y = androsperm).
- Both sexes have same autosome number.
- Found in humans and *Drosophila*.

XX–XO type

- Males have one sex chromosome (X), heterogametic; some sperms have X, some none.
- Females are homogametic (XX).
- Fertilisation with X sperm = female (2A + XX); without X = male (2A + XO).
- Found in grasshopper, cockroach, roundworms.

ZW–ZZ type

- Female: heterogametic (ZW), produces Z and W gametes.
- Male: homogametic (ZZ), produces Z gametes.
- Found in birds and some reptiles.
- ZO–ZZ type: females (AA + Z), males (AA + ZZ), found in butterflies and moths.

Memory Aids

- XX–XY: Male heterogametic, female homogametic.
- XX–XO: Male heterogametic with single X.
- ZW–ZZ: Female heterogametic, male homogametic.

Quick Checkpoints

- Which sex is homogametic in XX–XY?
- What determines sex in XX–XO type?
- Where is ZW–ZZ system found?

Haploid–Diploid Mechanism of Sex Determination

Mechanism

- Observed in Hymenoptera (ants, honeybees, wasps).
- Sex depends on chromosome set number.
- Females (queen, workers) develop from fertilised (diploid) eggs.

- Males (drones) develop from unfertilised (haploid) eggs by parthenogenesis.
- Males have 16 chromosomes (haploid), females 32 (diploid).
- Male sperms formed by mitosis, no meiosis.
- Males have no father, but have grandfather; can have grandsons.

Memory Aids

- Haploid males from unfertilised eggs.
- Diploid females from fertilised eggs.
- No meiosis in male sperm formation.

Quick Checkpoints

- Which insects show haploid-diploid sex determination?
- How are male and female chromosome numbers different?
- How are male sperms produced?

Sex Determination in Humans

Mechanism

- Humans use XY system.
- 22 pairs autosomes identical in males and females.
- Females have XX; males have XY chromosomes.
- Males produce two sperm types: 50% X-chromosome, 50% Y-chromosome.
- Females produce only X-chromosome ova.
- Fertilisation with X sperm = female (XX); with Y sperm = male (XY).
- Sex determined by sperm chromosome.
- Equal chance of male or female child per pregnancy.
- Sex-linked inheritance: genes on sex chromosomes inherited with sex traits.
- Sex-linked traits more visible in males; females often carriers (e.g., colour blindness, haemophilia).
- Criss-cross inheritance: traits passed to same-sex grandchild via opposite-sex parent.

Memory Aids

- XY system: male heterogametic, female homogametic.
- Sperm chromosome determines sex.
- Sex-linked traits show criss-cross inheritance.

Quick Checkpoints

- What chromosomes determine human sex?
- How does sperm type affect sex?
- What is criss-cross inheritance?

Mutation

Types and Effects

- Mutation alters DNA sequences, changing genotype and phenotype.
- Both recombination and mutation cause DNA variation.
- Loss or gain of DNA segments alters chromosomes, causing abnormalities.
- Chromosomal aberrations are common in cancer cells.
- *Chromosomal mutations* include:
  - **Deletion:** Loss of chromosome segment (terminal or intercalary).
  - **Duplication:** Repetition of gene blocks in haploid set.
  - **Translocation:** Exchange of segments between non-homologous chromosomes.
  - **Inversion:** Chromosome segment breaks, rotates 180°, and rejoins reversed.
  - **Euploidy:** Gain or loss of complete chromosome sets; types: monoploidy, polyploidy.
  - **Aneuploidy:** Gain or loss of individual chromosomes; types: trisomy, tetrasomy, monosomy, nullisomy.
- **Gene mutations** occur naturally or internally.
- **Point mutation:** Change in single base pair (e.g., sickle cell anaemia).
- **Frame-shift mutation:** Caused by insertions or deletions of base pairs.
- **Mutagens:** Chemical or physical agents inducing mutations (e.g., UV radiation).

Memory Aids

- Deletion = DNA segment lost.
- Duplication = DNA segment repeated.
- Translocation = Swap between non-homologous chromosomes.
- Inversion = Segment flipped 180°.
- Euploidy = Whole set change; Aneuploidy = Single chromosome change.
- Point mutation = Single base change.
- Frame-shift = Insertions/deletions shift reading frame.
- Mutagens = Mutation-causing agents.

Quick Checkpoints

- What causes chromosomal aberrations?
- Define deletion and duplication mutations.
- Explain translocation and inversion.
- Differentiate euploidy and aneuploidy.
- What is a point mutation?
- How do frame-shift mutations occur?
- Give examples of mutagens.

Genetic Disorders Pedigree Analysis

Explanation

- **Pedigree** is a chart showing inheritance of traits across generations in humans or animals.
- It helps deduce genotypes from phenotypes and study inheritance patterns of dominant/recessive traits or diseases.
- Genetic disorders arise from altered genes or chromosomes.
- Two main categories: Mendelian disorders and Chromosomal disorders.

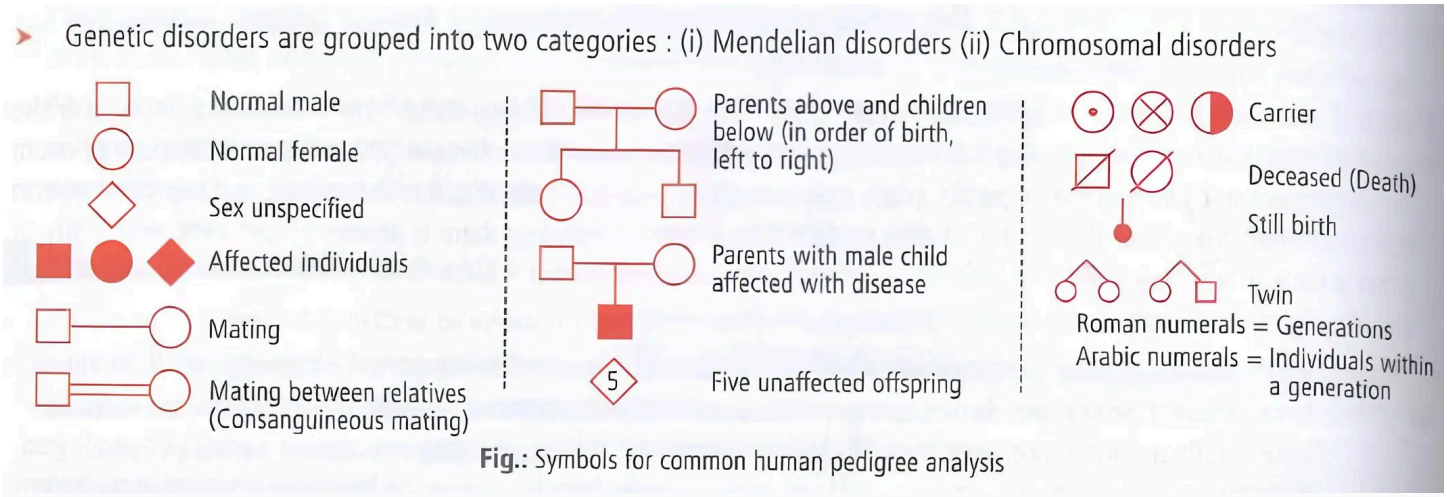


Fig.: Symbols for common human pedigree analysis

- Symbols in pedigree: squares (males), circles (females), diamonds (unspecified sex).
- Filled shapes indicate affected individuals.
- Single line = mating; double line = mating between relatives.
- Vertical lines connect parents to children.
- Special symbols for carriers, deceased, stillbirths, twins.
- Roman numerals for generations; Arabic numerals for individuals.

Memory Aids

- Square = Male, Circle = Female, Filled = Affected
- Single line = Mating, Double line = Consanguineous mating
- Roman numerals = Generations, Arabic numerals = Individuals

Quick Checkpoints

- What does a filled circle represent?
- How is consanguineous mating shown?
- What information does a pedigree provide?

Mendelian Disorders

Explanation

- Caused by mutation in a single gene.

- Inheritance pattern traced by pedigree analysis.
- Examples: haemophilia, cystic fibrosis, sickle-cell anaemia, colour blindness, phenylketonuria, thalassemia.
- Can be dominant or recessive; some are sex-linked (e.g., haemophilia is X-linked recessive).
- Carrier females transmit X-linked recessive traits to male offspring.

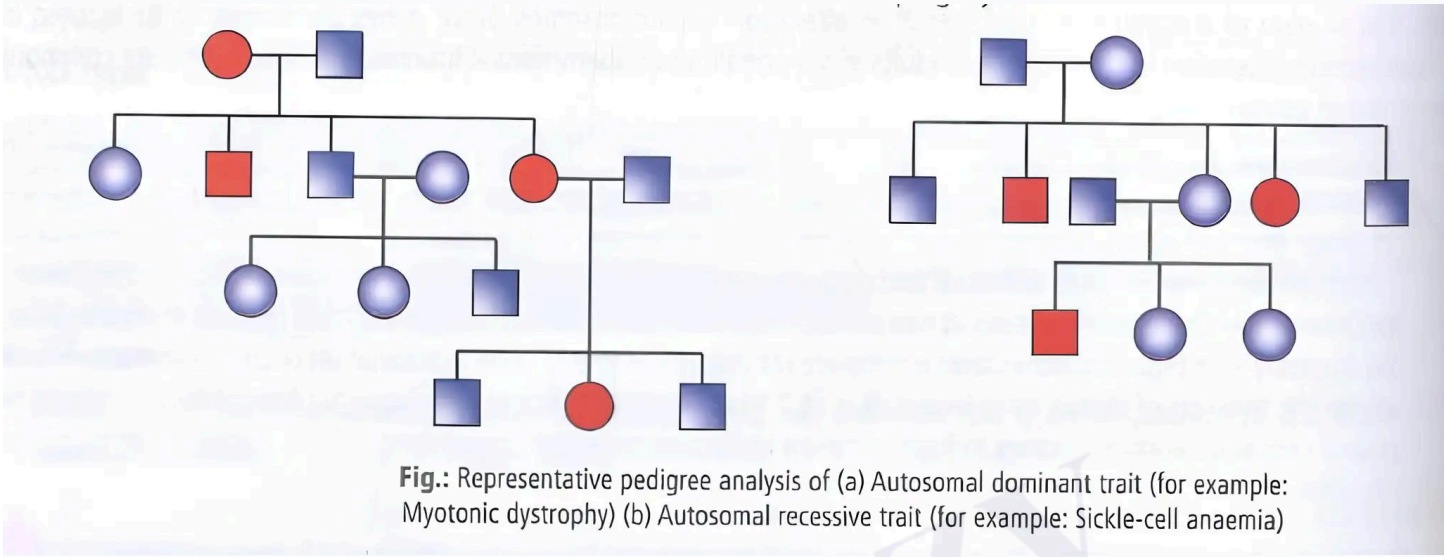


Fig.: Representative pedigree analysis of (a) Autosomal dominant trait (for example: Myotonic dystrophy) (b) Autosomal recessive trait (for example: Sickle-cell anaemia)

- Autosomal dominant: trait appears every generation if one parent affected.
- Autosomal recessive: trait appears when both parents carry gene, may skip generations.

Memory Aids

- Dominant traits appear in every generation.
- Recessive traits may skip generations.
- X-linked recessive traits mostly affect males.

Quick Checkpoints

- How to distinguish dominant vs recessive traits in pedigree?
- What is the inheritance pattern of haemophilia?
- Which disorders are Mendelian?

Colour Blindness

Explanation

- Sex-linked recessive disorder due to mutation in genes on X chromosome affecting red or green cones.
- Causes inability to distinguish red and green colors.
- Occurs in ~8% males, ~0.4% females due to males having one X chromosome.
- Carrier mothers have 50% chance of passing gene to sons.
- Daughters affected only if mother is carrier and father is colour blind.

Memory Aids

- Colour blindness gene is X-linked recessive.
- Males affected more due to single X chromosome.
- Carrier females usually unaffected.

Quick Checkpoints

- Why are males more affected?
- How is colour blindness inherited?
- What cones are affected?

Sickle-cell Anaemia

Explanation

- Autosomal recessive disorder caused by mutation in beta globin gene (Hb^A and Hb^s alleles).
- Only homozygous Hb^sHb^s individuals show disease; heterozygous are carriers.
- Mutation substitutes glutamic acid by valine at 6th position of beta globin chain.
- Causes hemoglobin polymerisation under low oxygen, changing RBC shape to sickle.

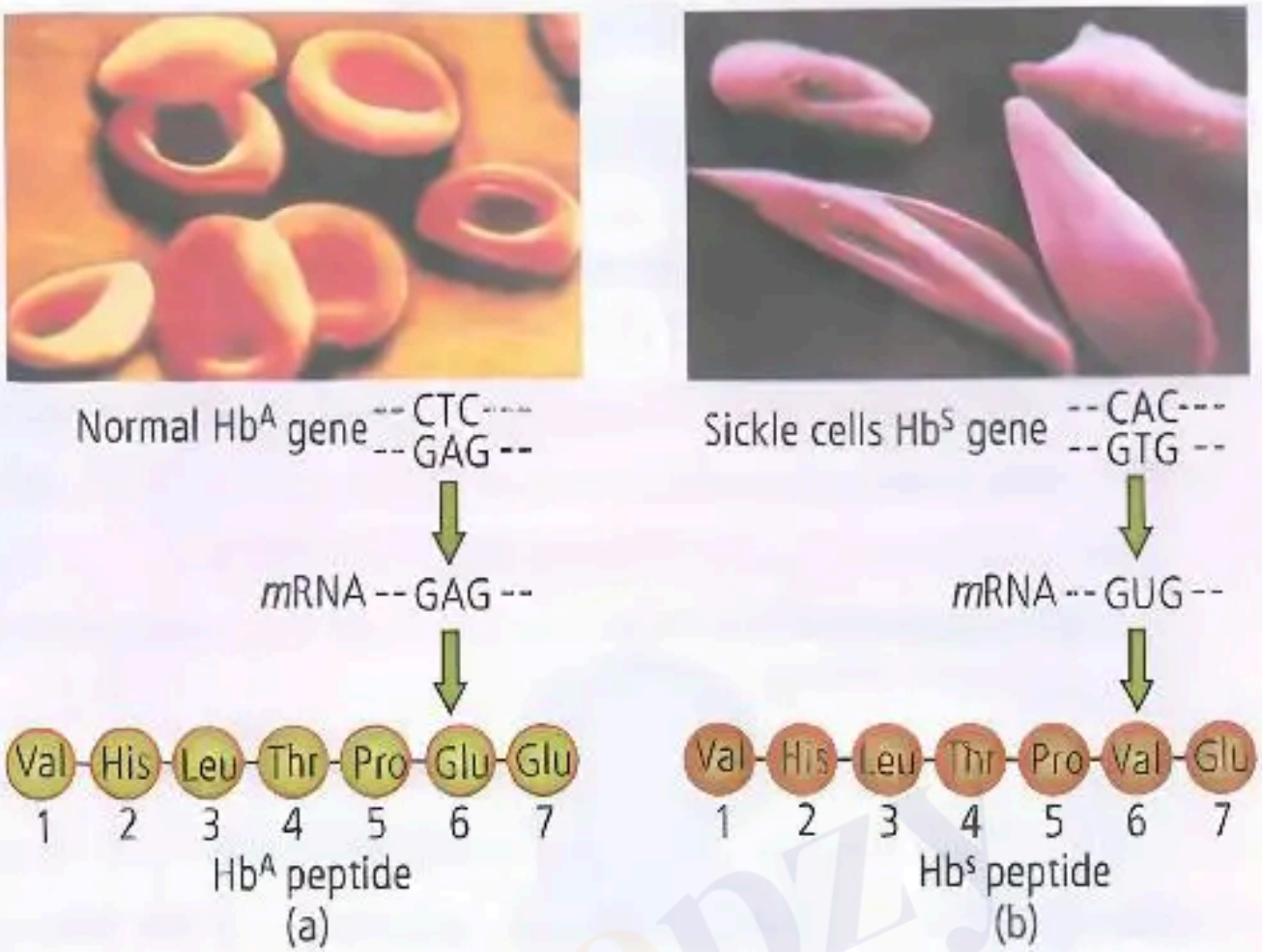


Fig.: Micrograph of the red blood cells and the amino acid composition of the relevant portion of β-chain of haemoglobin : (a) From a normal individual; (b) From an individual with sickle-cell anaemia

- Normal RBCs are round and flexible; sickle cells are crescent-shaped and rigid.
- DNA change: normal gene CTC to sickle gene CAC.
- mRNA changes from GAG to GUG causing amino acid change Glu to Val.
- Results in sickle-cell anaemia symptoms.

Memory Aids

- Hb^s allele causes sickle shape.
- Glu to Val substitution at 6th amino acid.
- Carriers are heterozygous, affected are homozygous.

Quick Checkpoints

- What amino acid substitution causes sickling?
- Which genotype shows disease?
- How does mutation affect RBC shape?

Haemophilia

Explanation

- Sex-linked recessive disorder affecting blood clotting protein.
- Carrier females transmit disease to male offspring.
- Causes prolonged bleeding from minor injuries.
- Female haemophilia is rare, requires carrier mother and affected father.
- Queen Victoria's pedigree shows haemophilia transmission.

Memory Aids

- X-linked recessive inheritance.
- Carrier females unaffected but transmit to sons.
- Causes bleeding due to clotting protein defect.

Quick Checkpoints

- How is haemophilia inherited?
- Why are males mostly affected?
- What causes bleeding in haemophilia?

Phenylketonuria

Explanation

- Autosomal recessive metabolic disorder.
- Lack of enzyme converting phenylalanine to tyrosine.
- Phenylalanine accumulates, converted to phenylpyruvic acid causing brain damage.
- Excess excreted in urine due to poor kidney absorption.

Memory Aids

- Autosomal recessive inheritance.
- Enzyme deficiency blocks phenylalanine metabolism.
- Causes mental retardation if untreated.

Quick Checkpoints

- Which enzyme is deficient?
- What accumulates in the body?
- How does it affect the brain?

Thalassemia

Explanation

- Autosomal recessive blood disorder from mutation or deletion affecting globin chain synthesis.
- Causes reduced synthesis of α, β, or δ globin chains leading to abnormal hemoglobin and anemia.
- Types: α-thalassemia (chromosome 16), β-thalassemia and δ-thalassemia (chromosome 11).
- Different from sickle-cell anemia: thalassemia is quantitative defect; sickle-cell is qualitative.

Memory Aids

- α-thalassemia: defective α-globin genes on chromosome 16.
- β-thalassemia: defective β-globin genes on chromosome 11.
- δ-thalassemia: defective δ-globin gene on chromosome 11.

Quick Checkpoints

- Which chromosomes carry globin genes?
- How does thalassemia differ from sickle-cell anemia?
- What causes anemia in thalassemia?

Chromosomal Disorders

Explanation

- Caused by absence, excess, or abnormal arrangement of chromosomes.
- Aneuploidy: gain or loss of chromosomes due to segregation failure (e.g., Down's syndrome trisomy 21, Turner's syndrome monosomy X).
- Polyploidy: failure of cytokinesis causing extra whole chromosome sets, common in plants.
- Normal human cells have 46 chromosomes (23 pairs): 22 autosomes + 1 pair sex chromosomes.
- Trisomy: extra chromosome; Monosomy: missing chromosome; both cause serious effects.

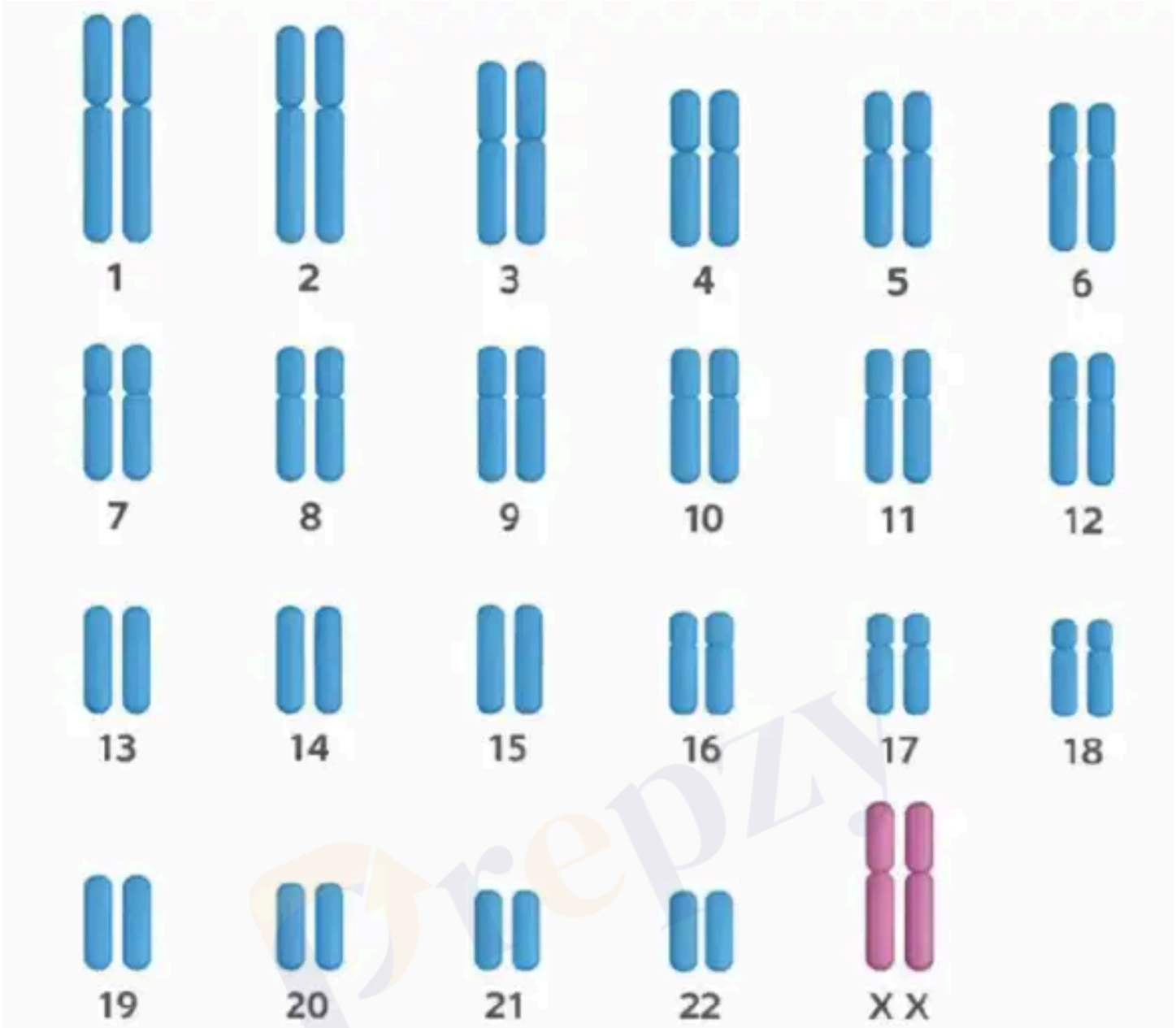
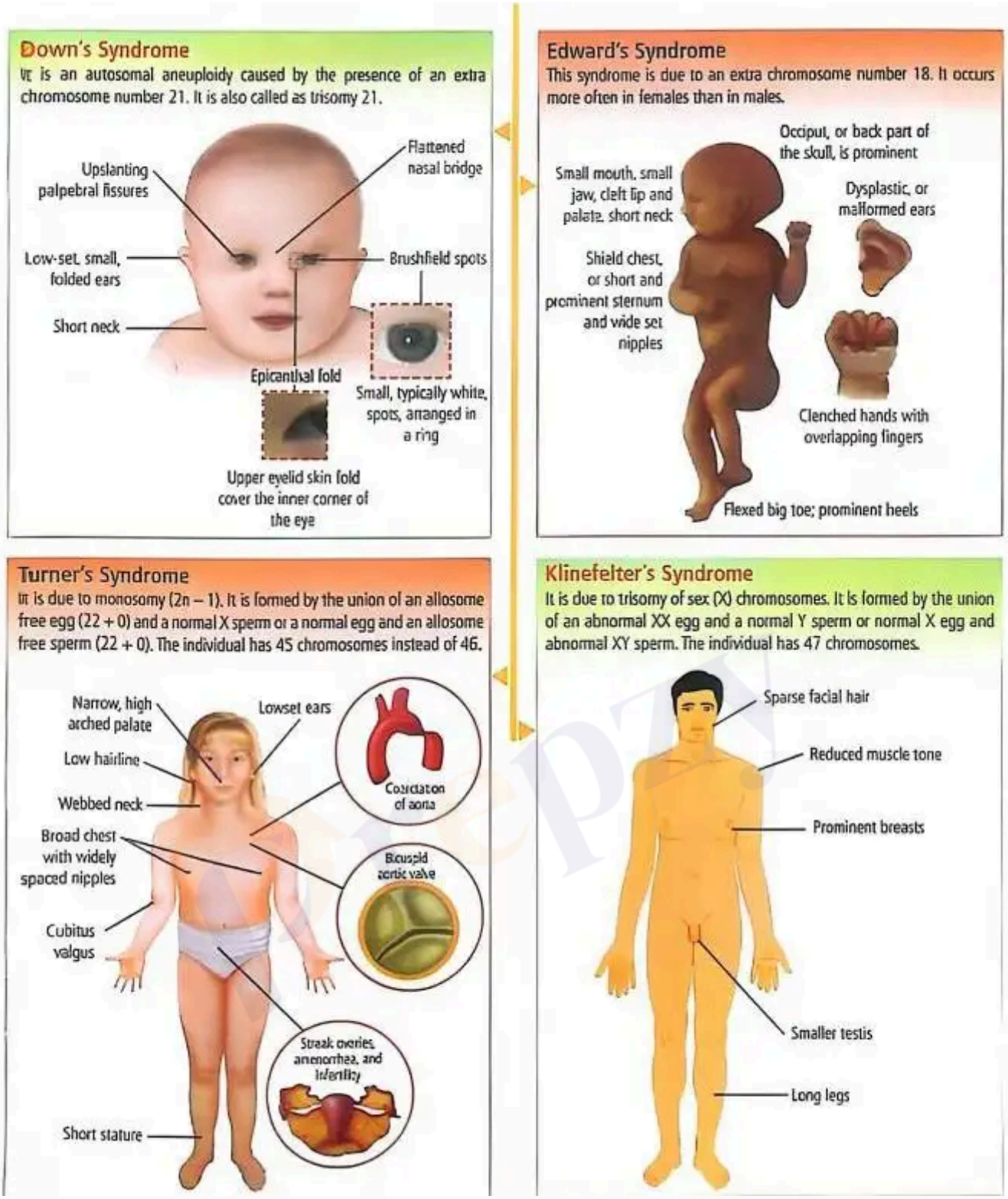


Fig.: A representative figure showing a karyotype of individual inflicted with Down's syndrome.

- Karyotype shows three copies of chromosome 21 in Down's syndrome.
- Extra chromosome causes developmental and physical traits of syndrome.



- Down's Syndrome: trisomy 21, features include flat nasal bridge, small ears, short neck.
- Edward's Syndrome: trisomy 18, features include small mouth, clenched hands, abnormal skull.
- Turner's Syndrome: monosomy X, features include short height, broad chest, webbed neck.
- Klinefelter's Syndrome: extra X in males, features include sparse hair, small testes, breast development.

Memory Aids

- Aneuploidy = abnormal chromosome number.
- Trisomy = extra chromosome; Monosomy = missing chromosome.
- Down’s = trisomy 21; Turner’s = monosomy X; Klinefelter’s = XXY males.

Quick Checkpoints

- What is aneuploidy?
- Which chromosome is extra in Down’s syndrome?
- What causes Turner’s syndrome?

Prepzy