FREE EDITION · NOTES + 3 SAMPLE MCQS

CUET · BIOLOGY · CLASS XII · CODE 304

Principles of Inheritance and Variation

CUET unit: Genetics and Evolution → Principles of Inheritance and Variation

By **UniDrill** · NCERT-grounded study material

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📌 Snapshot

- Builds the entire classical-genetics scaffold of CUET Biology: Mendel's three laws, monohybrid (3:1) and dihybrid (9:3:3:1) ratios, and the Punnett-square machinery for predicting genotypes/phenotypes.
- Extends Mendelism beyond simple dominance into incomplete dominance (Snapdragon, 1:2:1 phenotype), co-dominance with multiple alleles (ABO blood groups, I^A I^B i), pleiotropy (phenylketonuria), and polygenic inheritance (human skin colour, height).
- Anchors genes to chromosomes through Sutton–Boveri's Chromosomal Theory (1902) and Morgan's Drosophila work on linkage and recombination, including Sturtevant's genetic mapping using recombination frequency.
- Covers all four major sex-determination systems — XO (grasshopper), XY (humans, Drosophila), ZW (birds), and haplodiploidy (honey bees) — plus mutation (point mutations, chromosomal aberrations) and pedigree analysis symbols.
- Closes with disease genetics: Mendelian disorders (haemophilia, sickle-cell anaemia, phenylketonuria, thalassaemia, colour blindness) and chromosomal disorders with karyotypes (Down's — trisomy 21; Klinefelter — 47, XXY; Turner — 45, XO). The single most-tested chapter in CUET Biology.

📖 Detailed Notes

2.1 Core concepts

- Genetics is the branch of biology dealing with inheritance (transmission of characters from parent to progeny) and variation (degree by which progeny differ from parents) (NCERT §4 intro, p. 54).
- Gregor Mendel conducted hybridisation experiments on garden peas for seven years (1856–1863); he used large samples and applied statistical/mathematical logic to biology for the first time and selected 14 true-breeding lines as 7 pairs with contrasting traits (NCERT §4.1, p. 54).
- The seven pairs of contrasting traits Mendel studied: stem height (tall/dwarf), flower colour (violet/white), flower position (axial/terminal), pod shape (inflated/constricted), pod colour (green/yellow), seed shape (round/wrinkled), seed colour (yellow/green) (NCERT §4.1 Table 4.1, p. 55).

- A monohybrid cross (Tt × Tt) gives an F2 phenotypic ratio of 3:1 (tall:dwarf) and a genotypic ratio of 1:2:1 (TT:Tt:tt); the F1 is all Tt and phenotypically tall (NCERT §4.2, pp. 56–58).
- The Punnett square, developed by Reginald C. Punnett, graphically represents probabilities of all possible offspring genotypes by writing gametes on the top row and left column (NCERT §4.2, p. 57).
- A test cross is a cross between an organism showing dominant phenotype (unknown genotype) and the homozygous recessive parent; it is used to determine whether the dominant individual is homozygous (TT) or heterozygous (Tt) (NCERT §4.2, p. 58).
- **Law of Dominance** (Mendel's First Law): characters are controlled by discrete units called factors that occur in pairs; in a dissimilar pair one factor dominates the other; explains the 3:1 F2 phenotypic ratio (NCERT §4.2.1, p. 59).
- **Law of Segregation** (Second Law): alleles do not blend; during gamete formation the two alleles segregate so that each gamete carries only one of them; a heterozygote produces two kinds of gametes in equal proportion (NCERT §4.2.2, p. 59).
- **Incomplete dominance**: in snapdragon (Antirrhinum) / Mirabilis-type cross RR (red) × rr (white) → F1 Rr is pink, F2 is 1 red : 2 pink : 1 white; phenotypic ratio (1:2:1) equals genotypic ratio because R is not fully dominant over r (NCERT §4.2.2.1, p. 60).
- **Co-dominance**: in human ABO blood groups, gene I has three alleles I^A, I^B and i; I^A and I^B are both dominant over i but co-dominant with each other, so I^A I^B genotype expresses both A and B sugars on RBCs (AB blood type); 6 genotypes give 4 phenotypes (A, B, AB, O) (NCERT §4.2.2.2 Table 4.2, p. 61).
- **Multiple alleles**: ABO blood grouping illustrates more than two alleles for one gene in a population, though any individual carries only two (NCERT §4.2.2.2, p. 62).
- **Pleiotropy (incomplete-dominance angle)**: starch synthesis in pea seeds is controlled by one gene (B, b) — BB produces large round seeds, bb produces small wrinkled seeds, Bb produces round seeds with intermediate-sized starch grains; so the same gene shows complete dominance for shape but incomplete dominance for grain size (NCERT §4.2.2.2, p. 62).
- A dihybrid cross RrYy × RrYy gives an F2 phenotypic ratio of 9 round-yellow : 3 wrinkled-yellow : 3 round-green : 1 wrinkled-green (NCERT §4.3 and §4.3.1, pp. 63–64).
- **Law of Independent Assortment**: when two pairs of traits are combined in a hybrid, segregation of one pair is independent of the other; gametes formed by RrYy are RY, Ry, rY, ry each at 25% (NCERT §4.3.1, p. 64).
- **Chromosomal Theory of Inheritance**: Walter Sutton and Theodore Boveri (1902) noted that the behaviour of chromosomes during meiosis parallels the behaviour of

genes — both occur in pairs, both segregate at gamete formation, and independent pairs assort independently (NCERT §4.3.2 Table 4.3, p. 66).

- **Morgan & Drosophila**: T.H. Morgan used the fruit fly Drosophila melanogaster (simple medium, ~2-week life cycle, large progeny, distinguishable sexes) to experimentally verify the chromosomal theory (NCERT §4.3.2, p. 67).
- **Linkage and recombination**: when two genes lie on the same chromosome, parental combinations exceed non-parental (recombinant) combinations; Morgan coined "linkage" for physical association and "recombination" for generation of non-parental combinations. y–w showed 1.3% recombination (tightly linked) while w–m showed 37.2% (loosely linked); Sturtevant used recombination frequency to map gene positions (NCERT §4.3.3, pp. 67–68).
- **Polygenic inheritance**: traits like human skin colour and height are controlled by three or more genes with additive allelic effects and environmental influence; e.g., AABBCC = darkest, aabbcc = lightest skin (NCERT §4.4, p. 69).
- **Pleiotropy**: a single gene producing multiple phenotypic effects — phenylketonuria is caused by mutation in the gene coding for phenylalanine hydroxylase and manifests as mental retardation plus reduced hair and skin pigmentation (NCERT §4.5, p. 69).
- **Sex determination — XO type** (grasshopper, insects): males have only one X-chromosome (besides autosomes); females have two X-chromosomes; male-heterogamety (NCERT §4.6, p. 70).
- **XY type** (humans, Drosophila): males XY, females XX; male-heterogamety; both sexes have same total chromosome number; humans have 22 pairs of autosomes plus one pair of sex chromosomes (NCERT §4.6 / §4.6.1, p. 70–71).
- **ZW type** (birds): females ZW, males ZZ; female-heterogamety (NCERT §4.6, p. 71).
- **Haplodiploidy** (honey bees): females (queen/worker) develop from fertilised eggs and are diploid (32 chromosomes); males (drones) develop from unfertilised eggs by parthenogenesis and are haploid (16 chromosomes); drones have no father, cannot have sons, but have a grandfather and grandsons (NCERT §4.6.2, p. 71).
- **Mutation**: alteration of DNA sequence resulting in change in genotype and phenotype; arises from deletions/insertions/duplications of DNA segments (chromosomal aberrations, common in cancer cells) or single-base-pair changes (point mutations, e.g., sickle-cell anaemia); UV radiation is a mutagen (NCERT §4.7, p. 72).
- **Pedigree analysis**: study of inheritance of a trait across generations in a family tree, since controlled crosses are impossible in humans; uses standard symbols (NCERT §4.8.1, p. 72).
- **Colour blindness**: X-linked recessive defect in red or green cone; ~8% of males and ~0.4% of females are affected because the gene is on X chromosome; mother is unaffected carrier if heterozygous (NCERT §4.8.2, p. 73).

- **Haemophilia**: X-linked recessive; one of the clotting-cascade proteins is defective causing non-stop bleeding from a simple cut; heterozygous female (carrier) transmits to sons; female homozygotes are extremely rare because the father has to be haemophilic; Queen Victoria's family pedigree shows multiple haemophilic descendants (NCERT §4.8.2, p. 74).
- **Sickle-cell anaemia**: autosomal recessive; controlled by alleles Hb^A and Hb^S; only Hb^SHb^S shows the disease, Hb^AHb^S are carriers; caused by substitution of Glutamic acid by Valine at the sixth position of the β-globin chain (single base substitution GAG → GUG at the sixth codon); mutant haemoglobin polymerises under low oxygen tension changing RBC shape from biconcave to sickle-like (NCERT §4.8.2, pp. 74–75).
- **Phenylketonuria**: autosomal recessive inborn error of metabolism; affected individual lacks the enzyme that converts phenylalanine to tyrosine; accumulation of phenylalanine and phenylpyruvic acid in brain causes mental retardation; excreted via urine (NCERT §4.8.2, p. 75).
- **Thalassaemia**: autosomal recessive; reduced synthesis of one of the globin chains; α-thalassaemia controlled by two closely linked genes HBA1 and HBA2 on chromosome 16; β-thalassaemia controlled by single gene HBB on chromosome 11; differs from sickle-cell anaemia in being a quantitative (too few globin molecules) rather than qualitative defect (NCERT §4.8.2, p. 75).
- **Chromosomal disorders**: caused by absence/excess/abnormal arrangement of chromosomes; aneuploidy from failure of segregation of chromatids during cell division (gain/loss of chromosome); polyploidy from failure of cytokinesis (whole set added) (NCERT §4.8.3, p. 75).
- **Down's syndrome**: trisomy of chromosome 21 (extra copy of chromosome 21, total 47); first described by Langdon Down (1866); short stature, small round head, furrowed tongue, partially open mouth, broad palm with characteristic crease, retarded physical/psychomotor/mental development (NCERT §4.8.3, p. 76).
- **Klinefelter's syndrome**: karyotype 47, XXY (extra X); overall masculine development but gynaecomastia (breast development); sterile (NCERT §4.8.3, p. 76).
- **Turner's syndrome**: karyotype 45, X0 (one X chromosome missing); sterile females with rudimentary ovaries; lack secondary sexual characters (NCERT §4.8.3, p. 76).

2.2 Definitions to memorise

Term	Definition	Page
Inheritance	Process by which characters are passed on from parent to progeny; basis of heredity	54
Variation	Degree by which progeny differ from their parents	54
True-breeding line	Line that, after continuous self-pollination, shows stable trait inheritance for several generations	54

Term	Definition	Page
Allele	Slightly different forms of the same gene; code for a pair of contrasting traits	56
Genotype / Phenotype	Genetic constitution (TT/Tt/tt) vs descriptive appearance (tall/dwarf)	56
Homozygous / Heterozygous	Identical allelic pair (TT or tt) vs dissimilar (Tt)	56
Dominant / Recessive	In a dissimilar pair, the factor that dominates vs the one whose expression is masked	56
Monohybrid cross	Cross between two plants differing in one character (e.g., TT × tt)	57
Punnett square	Graphical tabular device by R.C. Punnett to calculate probabilities of offspring genotypes	57
Test cross	Cross of an organism showing dominant phenotype with the homozygous recessive parent to determine its genotype	58
Incomplete dominance	F1 phenotype is intermediate between the two parents (e.g., pink Rr)	60
Co-dominance	F1 expresses both parental phenotypes (e.g., AB blood from I^AI^B)	60–61
Multiple alleles	More than two alleles of a gene in a population (e.g., I^A, I^B, i)	62
Pleiotropy	A single gene producing multiple phenotypic effects (e.g., phenylketonuria)	69
Dihybrid cross	Cross between plants differing in two traits (RrYy × RrYy → 9:3:3:1)	64
Law of Independent Assortment	When two pairs of traits are combined in a hybrid, segregation of one pair is independent of the other	64
Chromosomal Theory of Inheritance	Sutton-Boveri synthesis: chromosomes carry the genes; their pairing/separation explains Mendel's laws	67
Linkage	Physical association of two or more genes on the same chromosome	67
Recombination	Generation of non-parental gene combinations through crossing over	67
Polygenic trait	Trait controlled by three or more genes, showing additive allelic effects and continuous variation	69
Point mutation	Change in a single base pair of DNA (e.g., GAG → GUG in sickle-cell anaemia)	72
Mutagen	Chemical/physical factor that induces mutation; UV radiation is a mutagen	72

Term	Definition	Page
Pedigree analysis	Study of inheritance of a particular trait in a family across generations	72
Aneuploidy	Gain or loss of a chromosome due to failure of segregation of chromatids	75
Polyploidy	Increase in a whole set of chromosomes due to failure of cytokinesis	75
Trisomy / Monosomy	Presence of an extra chromosome (2n+1) / absence of one chromosome (2n−1)	76

2.3 Diagrams / processes to remember

- Figure 4.1: Seven pairs of contrasting traits Mendel studied in pea (stem height, flower colour, flower position, pod shape, pod colour, seed shape, seed colour), p. 54.
- Figure 4.2: Steps in making a cross in pea — emasculation, dusting pollen, bagging, p. 55.
- Figure 4.3: Monohybrid cross diagrammatic representation (TT × tt → F1 all Tt tall → F2 3:1 phenotype, 1:2:1 genotype), p. 56.
- Figure 4.4: Punnett square for monohybrid cross between true-breeding tall and dwarf plants, p. 57.
- Figure 4.5: Test cross of violet (V) × white (v) showing 1:1 ratio, p. 59.
- Figure 4.6: Monohybrid cross in Snapdragon with incomplete dominance — F2 ratio 1 red : 2 pink : 1 white, p. 60.
- Figure 4.7: Dihybrid cross with seed colour and seed shape — F2 9:3:3:1 grid (16 squares), p. 63.
- Figure 4.8: Meiosis and germ-cell formation in a four-chromosome cell, p. 65.
- Figure 4.9: Independent assortment of chromosomes — two possibilities of metaphase alignment, p. 66.
- Figure 4.10: Drosophila melanogaster male and female, p. 67.
- Figure 4.11: Morgan's two dihybrid crosses showing tight linkage (y–w: 1.3% recombination) vs loose linkage (w–m: 37.2%), p. 68.
- Figure 4.12: Sex determination — (a) human XX/XY (b) Drosophila XX/XY (c) bird ZZ/ZW, p. 70.
- Figure 4.13: Sex determination in honey bee (haplodiploid; diploid females 32, haploid males 16), p. 71.
- Figure 4.13 (also): Symbols used in human pedigree analysis (male = square, female = circle, affected = shaded, mating = horizontal line, sibship = vertical descent), p. 72.
- Figure 4.14: Pedigrees of (a) autosomal dominant trait — myotonic dystrophy and (b) autosomal recessive trait — sickle-cell anaemia, p. 73.

- Figure 4.15: Micrograph of normal vs sickle-cell RBCs and the amino-acid composition of β-chain showing Glu→Val substitution at position 6, p. 74.
- Figure 4.16: Down's syndrome — flat back of head, broad flat face, big wrinkled tongue, palm crease, congenital heart disease; with trisomy-21 karyotype, p. 76.
- Figure 4.17: (a) Klinefelter's syndrome (tall, feminised character, gynaecomastia, 47 XXY); (b) Turner's syndrome (short stature, underdeveloped feminine character, 45 X0), p. 76.

2.4 Common confusions / NTA trap points

- **Phenotypic vs genotypic ratios.** In a normal monohybrid Tt × Tt cross, phenotypic is 3:1 but genotypic is 1:2:1; in incomplete dominance (Snapdragon RR × rr), both phenotypic and genotypic ratios are 1:2:1. NTA loves swapping these.
- **Co-dominance vs incomplete dominance.** Co-dominance — both alleles express their distinct phenotypes simultaneously (AB blood). Incomplete dominance — the heterozygote shows an intermediate/blended phenotype (pink Snapdragon).
- **Multiple alleles vs polygenic inheritance.** Multiple alleles = more than two alleles of the same gene (I^A, I^B, i for ABO). Polygenic = many different genes acting additively on the same trait (skin colour, height).
- **X-linked recessive disorders sex bias.** Colour blindness and haemophilia are far more common in males because they have only one X — a single recessive allele expresses. NTA distractors will swap the % frequencies (8% male / 0.4% female for colour blindness).
- **Sickle-cell anaemia substitution direction.** The mutation is GAG → GUG (codon change) and Glu → Val (amino-acid change) at the **sixth** position of the β-globin chain. Wrong-position (4th, 7th) or wrong-chain (α-chain) distractors are classic NTA traps.
- **Down's syndrome karyotype.** Trisomy of chromosome 21 → 47 chromosomes total (NOT 47, XX or 47, XY notation alone — the autosomal 21 is the extra one). Don't confuse with Klinefelter (47, XXY — sex-chromosome trisomy).
- **Turner vs Klinefelter karyotypes.** Turner = 45, X0 (female, missing X). Klinefelter = 47, XXY (male, extra X). Both are sterile, but one has a chromosome missing and the other has an extra one.
- **Honey bee haplodiploidy.** Males are haploid (16, from unfertilised egg), females are diploid (32). "Drones produce sperm by mitosis" and "drones have no father" are favourite assertion-reason hooks.
- **Linkage strength vs recombination frequency.** Tightly linked genes show low recombination frequency (y-w: 1.3%), loosely linked show high (w-m: 37.2%). Distractors will invert this relationship.

🎯 Practice MCQs

Q1. For how many years did Gregor Mendel conduct his hybridisation experiments on garden pea, and during which period?

A. Five years (1856–1861)

B. Seven years (1856–1863)

C. Eight years (1858–1866)

D. Ten years (1855–1865)

Q2. Which of the following pairs of contrasting traits was **NOT** among the seven studied by Mendel in pea plants?

A. Pod colour: green / yellow

B. Flower position: axial / terminal

C. Leaf shape: serrated / smooth

D. Seed shape: round / wrinkled

Q3. In a monohybrid cross between true-breeding tall (TT) and dwarf (tt) pea plants, the F2 progeny show the following ratios:

A. Phenotypic 1:2:1; Genotypic 3:1

B. Phenotypic 3:1; Genotypic 1:2:1

C. Phenotypic 9:3:3:1; Genotypic 1:1

D. Phenotypic 1:1; Genotypic 1:2:1

📊 PYQ Alignment

This chapter has been the single highest-weight chapter in CUET Biology 2023–25, contributing roughly 18–22 MCQs per paper across the section. NTA's favourite question types are: (i) numerical ratio problems on monohybrid/dihybrid/test crosses (3:1, 1:2:1,

9:3:3:1, 1:1), (ii) probability questions on dihybrid Punnett squares (e.g., proportion of tall-green offspring), (iii) ABO blood-group genotype-to-phenotype matching, (iv) sickle-cell mutation specifics (codon change GAG → GUG, amino-acid substitution Glu → Val at β-chain position 6), (v) sex-determination system matching (XO/XY/ZW/haplodiploid), and (vi) karyotype-to-disorder matching for Down's, Klinefelter's and Turner's. Pedigree-based questions on X-linked recessive inheritance (haemophilia, colour blindness) and assertion-reason items on incomplete vs co-dominance also recur every cycle.

CUET 2025 — Actual PYQs from this chapter

Q.12 (CUET 2025) Match List-I (Character) with List-II (Recessive trait). Character Recessive trait (A) Flower colour (i) Green (B) Seed colour (ii) Yellow (C) Pod colour (iii) Constrict ed (D) Pod shape (iv) White

- A) [option not extracted — see source]
- B) [option not extracted — see source]
- C) [option not extracted — see source]
- D) [option not extracted — see source]

Tests: aligns with §2 (principles of inheritance) Answer: Not in extracted key — verify against official NTA key

Q.14 (CUET 2025) Which of the following disorders are the results of aneuploidy? (A) Haemophilia (B) Down’s syndrome (C) Thalassemia (D) Turner’s syndrome

- A) [option not extracted — see source]
- B) [option not extracted — see source]
- C) [option not extracted — see source]
- D) [option not extracted — see source]

Tests: aligns with §2 (principles of inheritance) Answer: Not in extracted key — verify against official NTA key

Q.15 (CUET 2025) Which option expresses intermediate skin colour in an individual?

- A) AABBCC
- B) aabbcc
- C) AaBbCc
- D) aaBBcc

Tests: aligns with §2 (principles of inheritance) Answer: Not in extracted key — verify against official NTA key

CUET 2024 — Actual PYQs from this chapter

Q.7 (CUET 2024) Which pair of contrasting traits was not studied by Mendel?

- A) Pink and white flowers
- B) Inflated and constricted pods
- C) Axial and terminal flowers

- D) Green and yellow pods

Tests: aligns with §2 (principles of inheritance) Answer: Not in extracted key — verify against official NTA key

Q.8 (CUET 2024) Failure of chromatids to segregate during cell division results in:

- A) Polyploidy
- B) Euploidy
- C) Aneuploidy
- D) Autopolyploidy

Tests: aligns with §2 (principles of inheritance) Answer: Not in extracted key — verify against official NTA key

Q.9 (CUET 2024) Select correctly matched pair about sickle cell anaemia. Genotype – Phenotype pairs given.

- A) [option not extracted — see source]
- B) [option not extracted — see source]
- C) [option not extracted — see source]
- D) [option not extracted — see source]

Tests: aligns with §2 (principles of inheritance) Answer: Not in extracted key — verify against official NTA key

Q.10 (CUET 2024) Match scientists with discoveries. Scientists Discovery Sutton & Boveri Chromosomal theory Sturtevant Genetic maps Henking X-body Griffith Transformation

- A) [option not extracted — see source]
- B) [option not extracted — see source]
- C) [option not extracted — see source]
- D) [option not extracted — see source]

Tests: aligns with §2 (principles of inheritance) Answer: Not in extracted key — verify against official NTA key

CUET 2023 — Actual PYQs from this chapter

Q.10 (CUET 2023) Which of the following is not a Mendelian disorder?

- A) Haemophilia
- B) Sickle-cell anaemia
- C) Down's syndrome
- D) Phenylketonuria

Tests: aligns with §2 (principles of inheritance) Answer: Not in extracted key — verify against official NTA key

Q.11 (CUET 2023) Match List-I with List-II List-I (A) Metabolic disorder (B) Pedigree analysis (C) Thalassemia (D) Trisomy of 21st chromosome List-II (I) Family tree over generations (II) Blood disease (III) Phenylketonuria (IV) Down's syndrome

- A) A-III, B-IV, C-III, D-I
- B) A-I, B-III, C-II, D-IV
- C) A-III, B-I, C-II, D-IV
- D) A-III, B-II, C-IV, D-I

Tests: aligns with §2 (principles of inheritance) Answer: Not in extracted key — verify against official NTA key