FREE NOTES · NCERT + NEET LEVEL

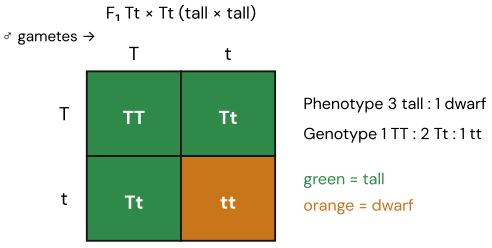
Principles of Inheritance & Variation

Mendel's experiments and laws, monohybrid and dihybrid crosses, incomplete dominance, co-dominance, multiple alleles, pleiotropy, polygenic inheritance, chromosomal theory, linkage, sex determination, mutation, pedigree analysis, Mendelian and chromosomal disorders.

01 Mendel and his laws
02 Dihybrid cross and deviations
03 Polygenic inheritance and chromosomal theory
04 Sex determination
05 Mutation and pedigree analysis
06 Genetic disorders
07 Extra + tricks
08 Important Q&A
09 Common mistakes
10 Practice (NEET style)
11 Quick revision

01 Mendel and his laws

- Gregor Mendel (1856–1863) on garden pea (*Pisum sativum*): 7 pairs of contrasting traits (stem height, flower colour, flower position, pod shape, pod colour, seed shape, seed colour). Used true-breeding lines and statistics.
- Factors (now genes) occur in pairs; alleles = alternative forms. Dominant, recessive. Genotype vs phenotype.
- **Law of dominance**: in a dissimilar pair, one factor dominates. **Law of segregation**: alleles separate during gamete formation (gametes are always pure).
- **Test cross**: F_1 × recessive parent; 1:1 ⇒ heterozygous.
- Monohybrid F_2: phenotype 3:1, genotype 1:2:1.



Punnett square of monohybrid cross

02 Dihybrid cross and deviations

- Dihybrid (round yellow RRYY × wrinkled green rryy): F_2 **9:3:3:1**. **Law of independent assortment**: segregation of one pair is independent of the other.
- Mendel's work published 1865, rediscovered in 1900 by de Vries, Correns, von Tschermak.
- **Incomplete dominance**: snapdragon/dog flower (*Antirrhinum*): red × white ⇒ pink F_1; F_2 1 red : 2 pink : 1 white (phenotype = genotype ratio).
- **Co-dominance**: ABO blood groups (gene I, alleles I^A, I^B, i): I^A and I^B both expressed in AB. Also example of **multiple alleles** (3 alleles, 6 genotypes, 4 phenotypes).
- **Pleiotropy**: one gene, many effects: phenylketonuria; starch synthesis gene in pea (large/small starch grains and round/wrinkled seeds).
- Dominance depends on product: in pea seed shape, B (dominant) enzyme converts sugar to starch.

03 Polygenic inheritance and chromosomal theory

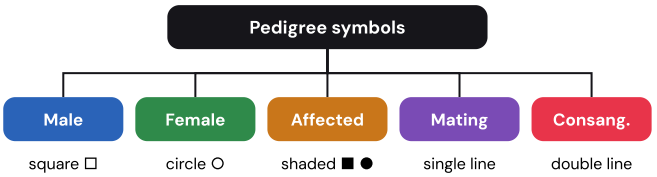
- **Polygenic**: trait controlled by 3 or more genes, each with additive effect; e.g. human skin colour, height. Gives a continuous range (bell curve).
- **Chromosomal theory of inheritance**: Sutton and Boveri (1902): chromosome movement explains Mendel's laws.
- **T.H. Morgan** on *Drosophila melanogaster* (fruit fly: completes life cycle in 2 weeks, many progeny, easily grown, clear male/female, many visible hereditary variations).
- **Linkage**: genes on same chromosome tend to be inherited together. **Recombination**: new combinations. Tightly linked genes: low recombination (white/yellow: 1.3%); loosely linked: higher (white/miniature: 37.2%).
- **Sturtevant** used recombination frequency to map genes on chromosomes (genetic maps).

04 Sex determination

System	Female	Male	Example
XX–XO	XX	XO (male heterogametic)	Grasshopper, many insects
XX–XY	XX	XY (male heterogametic)	Humans, *Drosophila*
ZW–ZZ	ZW (female heterogametic)	ZZ	Birds
Haplodiploidy	Diploid (fertilised egg)	Haploid (unfertilised egg, parthenogenesis)	Honey bee

Henking (1891) saw "X body" in insects. In humans, each pregnancy has 50% chance of boy or girl; sex is decided by the father's sperm.

05 Mutation and pedigree analysis



Common symbols used in pedigree charts

- **Mutation**: change in DNA sequence ⇒ change in genotype and phenotype. Chromosomal aberrations (deletion, insertion, duplication) often in cancer cells. **Point mutation**: single base change (sickle-cell anaemia). Frameshift: deletion/insertion of bases. Mutagens: UV radiation, chemicals.
- **Pedigree analysis**: study of a trait over generations in a family tree; helps trace inheritance of disorders (autosomal or sex-linked, dominant or recessive).

06 Genetic disorders

Disorder	Type	Key feature
Haemophilia	Sex-linked recessive	Clotting protein defect; small cut ⇒ non-stop bleeding; carrier females transmit to sons; royal family of Queen Victoria
Sickle-cell anaemia	Autosomal recessive	Glu → Val at 6th position of β-globin (GAG → GUG); Hb^sHb^s affected; RBCs sickle-shaped at low O_2
Phenylketonuria	Autosomal recessive (inborn error)	Lacks enzyme converting phenylalanine to tyrosine; mental retardation; phenylpyruvic acid in urine
Colour blindness	Sex-linked recessive	Red/green cones defective; ~8% males, 0.4% females
Thalassaemia	Autosomal recessive	Reduced synthesis of α (α-thal, genes HBA1, HBA2 on chr 16) or β globin (β-thal, HBB on chr 11); quantitative problem (sickle cell = qualitative)
Down's syndrome	Trisomy 21	Short stature, small round head, furrowed tongue, partially open mouth, palm crease, retarded development (Langdon Down, 1866)
Klinefelter's	47, XXY	Male, gynaecomastia, sterile, overall masculine development
Turner's	45, XO	Female, sterile (rudimentary ovaries), lack secondary sexual characters

Aneuploidy from failure of chromatid segregation; polyploidy (common in plants) from failure of cytokinesis.

07 Extra + tricks

Syndrome	Cause
Patau	Trisomy 13
Edwards	Trisomy 18
Down	Trisomy 21
Cri-du-chat	Deletion on chromosome 5 (cat-like cry)
Klinefelter	47, XXY
Turner	45, XO
Cystic fibrosis	Autosomal recessive; thick mucus in lungs, gut

- Number of gamete types = $\mathbf{2^n}$ (n = heterozygous pairs); F_2 genotypes = 3^n; F_2 phenotypes (complete dominance) = 2^n.
- *Drosophila* sex by X:A ratio: 1 = female, 0.5 = male, >1 = super female, <0.5 = super male, between = intersex.
- Pedigree tricks: affected child of unaffected parents ⇒ **recessive**; trait in every generation ⇒ often **dominant**; mostly males + passed via carrier mothers ⇒ **X-linked recessive**.

- Haemophilia A = factor VIII; B = factor IX (Christmas disease).

08 Important Q&A

Q1. A man of blood group A marries a woman of group B; their child is O. Give parents' genotypes.

Father I^Ai, mother I^Bi (both heterozygous); child ii.

Q2. Why do we see 1:2:1 phenotypic ratio in snapdragon F_2?

Incomplete dominance: heterozygote (Rr) is pink, intermediate, so each genotype has its own phenotype.

Q3. Why is haemophilia more common in males?

It is X-linked recessive; males have one X, so one defective allele causes disease; females need two.

09 Common mistakes

- Calling ABO only co-dominance (also multiple alleles).
- Writing dihybrid test-cross ratio as 9:3:3:1 (it is 1:1:1:1).
- Saying females are heterogametic in humans.
- Mixing thalassaemia (quantity) with sickle-cell (quality).

10 Practice (NEET style)

1. F_2 ratio of dihybrid cross?
2. Karyotype of Turner's syndrome?
3. Organism used by Morgan?
4. Sex determination in birds?
5. Trisomy 21 causes?

ANSWERS

1) 9:3:3:1 · 2) 45, XO · 3) *Drosophila* · 4) ZW female, ZZ male · 5) Down's syndrome

11 Quick revision: 5 minutes before the exam

MENDEL

- 3:1, 1:2:1
- 9:3:3:1
- Test cross 1:1

BEYOND MENDEL

- Snapdragon incomplete
- ABO co-dominant, multiple alleles
- Skin colour polygenic

CHROMOSOMES

- Sutton–Boveri theory
- Morgan linkage
- XO grasshopper, ZW birds

DISORDERS

- Haemophilia, colour blindness X-linked
- Sickle GAG→GUG
- Down 21, Klinefelter XXY, Turner XO

Have a doubt? Ask the AI Doubt Solver at padhaiadda.netlify.app, or take the mock test on this chapter.