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Step-by-step solutions, alternate methods & exam tips for Class 12 Biology

Chapter 4: Principles of Inheritance and Variation

About this Chapter

This chapter introduces **Mendelian genetics** through pea plant experiments, the laws of **dominance, segregation and independent assortment**, and how phenotype emerges from genotype. We move on to deviations from Mendel: **incomplete dominance**, **co-dominance**, multiple alleles (ABO blood groups), **pleiotropy**, sex-linked inheritance, **linkage and recombination** (Morgan's flies), pedigree analysis, sex determination, and chromosomal disorders such as Down's, Klinefelter's and Turner's. By the end you can predict cross outcomes with Punnett squares, read pedigrees, and explain why traits sometimes break the 3:1 ratio.

Topics covered: Mendel's pea experiments • Laws of inheritance • Punnett squares • Test cross • Incomplete dominance • Co-dominance • ABO blood groups • Linkage and recombination • Pedigree analysis • Sex determination • Genetic disorders

Quick Formula Sheet

Monohybrid F_2 ratio:
Phenotype $3:1$, Genotype $1:2:1$

Dihybrid F_2 ratio:
Phenotype $9:3:3:1$

Gamete count for n heterozygous loci:
$N_{\text{gametes}} = 2^n$

Test cross ratio (heterozygote $\times$ recessive):
$1:1$ phenotype

Recombination frequency:
$\text{RF} = \dfrac{\text{Recombinants}}{\text{Total}} \times 100\%$

Also see for this chapter: Revision Notes | Formula Sheet | Exemplar Solutions

Exercises

Q 4.1 **Mention the advantages of selecting pea plant for experiment by Mendel.**

SOLUTION

Concept used. A genetic experiment needs an organism whose **breeding can be tightly controlled**, whose generations turn over fast, and whose traits show as clear either-or alternatives so that ratios can be counted without ambiguity. The garden pea (*Pisum sativum*) meets all three conditions, which is why Mendel's choice of organism mattered as much as his bookkeeping.

☞ **Why this question matters**

Mendel did not get lucky with his ratios. He got them because pea plants are an almost ideal genetic system: short life cycle, self-pollination by default, easy hand-crossing, and seven traits that are sharply contrasting.

Step 1. **Many sharply contrasting traits.** Mendel chose seven characters that occur in two clearly distinguishable forms each (e.g. tall *vs.* dwarf stem, round *vs.* wrinkled seed, yellow *vs.* green cotyledon, violet *vs.* white flower). Discrete alternatives make progeny easy to score; no measurement scale is needed.

Step 2. **Self-pollination by default, but cross-pollination possible.** Pea flowers are **bisexual** and the keel-shaped petals enclose the stamens and stigma, so the plants are usually self-pollinated and naturally produce true-breeding (homozygous) lines over generations. When Mendel wanted to make a hybrid, he simply emasculated the bud (removed anthers before pollen matured) and dusted pollen from the chosen male parent onto the stigma. So one species gave him both pure lines AND cross-bred lines.

Step 3. **Short life cycle and many offspring per cross.** A pea plant completes one generation in a single growing season and each cross yields a large number of seeds, so Mendel could repeat experiments quickly and apply statistics on big F_2 populations.

Step 4. **True-breeding pure lines were available.** Generations of self-pollination had already produced **homozygous** stocks of pea for each trait, so Mendel could start every cross knowing the genotype of both parents.

Step 5. **Hardy, easy to grow, no special equipment.** Pea is a garden crop, grows from seed in any monastery plot, and needs no specialised lab.

Final Answer: Pea has many sharply contrasting traits, is normally self-pollinating yet easy to cross-pollinate, produces many seeds per cross in one short season, has ready-made true-breeding lines, and is easy to grow, together making it almost ideal for genetic analysis.

☞ Exam Tip

NCERT examiners frequently ask this as a 3-mark question. To bag full marks, write FIVE distinct advantages (not three): contrasting traits + bisexual flowers + self-pollination default + short life cycle + many offspring. Always mention that crossing is also possible (some students forget this and lose a mark).

EXPERT'S SOLUTION *: Aanya Reddy, M.Sc Botany, Delhi University*

Why this organism. Think of Mendel as a quantitative biologist before genetics existed. He needed a system where each cross was tractable mathematically. Three properties make pea fit that bill: (i) discrete phenotypes you can count, (ii) controllable mating to set up the cross you want, and (iii) enough progeny to see real ratios above sampling noise.

Step 1. **Discrete phenotypes.** Mendel found seven traits whose two alleles segregate cleanly: stem height, flower colour, flower position, pod shape, pod colour, seed shape, seed colour. No "tallish" or "yellowish-green" intermediate, and that absence of blending is what kept the 3:1 ratio visible.

Step 2. **Controllable mating.** Pea is normally **autogamous** (self-pollinating), so true-breeding lines exist in nature. To make a cross Mendel emasculated the female parent before anther dehiscence and applied pollen from the chosen male – clean experimental control.

Step 3. **Statistical power.** Each pod gives several seeds and each plant gives many pods. A single F_2 population can run into the hundreds, which is what lets a $3:1$ ratio show itself above random fluctuation.

Step 4. **Other practical pluses.** Short generation time ($\sim$3 months), easy garden cultivation, large flowers easy to manipulate by hand, and a clear distinction between seed traits (visible without growing the plant) and adult traits (require growing).

Why this matters. If Mendel had picked a plant with blending inheritance or with hidden hybrid vigour, the laws he discovered would have been invisible. The principles of inheritance are as much a story of good experimental design as of biology.

> **Final Answer:** Discrete contrasting traits, bisexual self-pollinating flowers that can also be cross-pollinated, large progeny per cross, short generation time, and ready true-breeding lines – pea was chosen because all five conditions hold simultaneously.

Q 4.2 Differentiate between the following:

(a) Dominance and Recessive
(b) Homozygous and Heterozygous
(c) Monohybrid and Dihybrid.

SOLUTION

Concept used. The vocabulary of Mendelian genetics rests on two ideas. First, an **allele** is one of the alternative forms of a gene; a diploid carries two alleles for each autosomal gene. Second, a **cross** is a controlled mating designed to track how alleles move from parent to offspring.

Step 1. **(a) Dominance and Recessive.**

- A **dominant** allele is one whose phenotype expresses itself even when only one copy is present in the diploid (heterozygous Aa shows the dominant trait). Conventionally written with a capital letter (T, R, Y).
- A **recessive** allele is one whose phenotype appears only when both copies present in the diploid are recessive (aa). Written in lower case (t, r, y).
- In a monohybrid F_2 population the dominant phenotype appears in $3/4$ of progeny and the recessive in $1/4$ (the $3{:}1$ ratio).

Step 2. **(b) Homozygous and Heterozygous.**

- **Homozygous** means an individual carrying two identical alleles at a locus: either TT (homozygous dominant) or tt (homozygous recessive). Such an individual breeds true on self-pollination.
- **Heterozygous** means an individual carrying two different alleles at a locus: Tt. Its phenotype matches whichever allele is dominant, but its progeny segregate on selfing in a $3{:}1$ ratio.

Step 3. **(c) Monohybrid and Dihybrid.**

- A **monohybrid cross** is between parents that differ in *one* character (e.g. $TT \times tt$). F_1 is uniform; F_2 shows a $3{:}1$ phenotypic ratio ($1{:}2{:}1$ genotypic).
- A **dihybrid cross** is between parents that differ in *two* unlinked characters (e.g. $RRYY \times rryy$). F_2 shows a $9{:}3{:}3{:}1$ phenotypic ratio when the two loci assort independently.

Final Answer: Dominance hides recessive; homozygote has two identical alleles, heterozygote has two different alleles; monohybrid tracks one trait (3:1 in F_2), dihybrid tracks two traits (9:3:3:1 in F_2).

EXPERT'S SOLUTION *: Vivaan Iyer, M.Sc Biotechnology, AIIMS Delhi*

Structural angle. It helps to see these three pairs as answers to three different questions: what *shows up* (dominance vs. recessive), what is *inside* the individual (homozygous vs. heterozygous), and what the *cross is tracking* (monohybrid vs. dihybrid).

Step 1. **Phenotype-level pair: dominance vs. recessive.** These describe *alleles* by their behaviour. A dominant allele A expresses in both AA and Aa; a recessive allele a expresses only in aa. Tall (T) in pea is dominant over dwarf (t): TT and Tt are tall; only tt is dwarf.

Step 2. **Genotype-level pair: homozygous vs. heterozygous.** These describe *individuals*. TT and tt are homozygous (same two alleles), Tt is heterozygous (two different alleles). A homozygous plant on selfing always gives offspring of the same genotype; a heterozygous plant gives offspring in a $1{:}2{:}1$ genotypic split.

Step 3. **Cross-level pair: monohybrid vs. dihybrid.** These describe *crosses*. In a monohybrid we track one trait; F_2 phenotypes split $3{:}1$, genotypes $1{:}2{:}1$. In a dihybrid we track two traits; F_2 phenotypes split $9{:}3{:}3{:}1$ (which is the product $3{:}1 \times 3{:}1$, Mendel's law of independent assortment).

Why this matters. Each pair belongs to a different layer of description: allele behaviour, individual genotype, experimental design. Mixing the layers is a frequent slip in answers.

> **Final Answer:** Three layers: allele behaviour (dominant vs. recessive), individual make-up (homozygous vs. heterozygous), and cross design (monohybrid 3:1 vs. dihybrid 9:3:3:1).

♥ Connecting the three pairs

A heterozygous individual shows the dominant phenotype; a homozygous recessive individual shows the recessive phenotype. So homozygous/heterozygous *describes the genotype*, while dominant/recessive *describes the allele*. Don't mix them.

Q 4.3 A diploid organism is heterozygous for 4 loci, how many types of gametes can be produced?

SOLUTION

Concept used. If an organism is heterozygous at a single locus (say Aa), then during meiosis the two alleles segregate and the organism produces *two* kinds of gametes (A and a) in equal proportion. When the organism is heterozygous at n *independently assorting* loci, every gamete carries one allele from each locus, and the choices at the n loci are independent. So by the **multiplication principle of counting**:

$$N_{\text{gametes}} = 2 \times 2 \times \cdots \times 2 \ (n \text{ times}) \ = \ 2^n.$$

☞ **Why 2^n and not n^2**

The exponent counts how many *independent binary choices* are being made. Four heterozygous loci means four independent two-allele choices, so 2^4, not 4^2. The same logic gives $2^2 = 4$ gametes for a dihybrid $AaBb$, which is exactly what Mendel's 4×4 Punnett square uses on each axis.

Step 1. **Identify n.** The organism is heterozygous at four loci, so $n = 4$. Call the loci A/a, B/b, C/c, D/d; the organism's genotype is $AaBbCcDd$.

Step 2. **Apply the formula.**

$$N_{\text{gametes}} = 2^n = 2^4.$$

Step 3. **Compute.**

$$2^4 = 2 \times 2 \times 2 \times 2 = 16.$$

Step 4. **Sanity check by listing.** The 16 gametes are:

$$\begin{gathered} ABCD,\ ABCd,\ ABcD,\ ABcd, \\ AbCD,\ AbCd,\ AbcD,\ Abcd, \\ aBCD,\ aBCd,\ aBcD,\ aBcd, \\ abCD,\ abCd,\ abcD,\ abcd. \end{gathered}$$

That is exactly 16 entries – matching 2^4.

Final Answer: The organism can produce $\mathbf{2^4 = 16}$ types of gametes.

✗ Common Mistake

A common slip: writing $4^2 = 16$ "by accident". The result is the same number, but the reasoning is wrong, and the same wrong logic gives $5^2 = 25$ for $n = 5$ (the correct answer is $2^5 = 32$). Always write 2^n first.

EXPERT'S SOLUTION *: Karan Nair, M.Sc Biotechnology, AIIMS Delhi*

Quick reading. The formula is short, but the reasoning behind it is worth stating cleanly because it generalises to every n-locus question on the syllabus.

Step 1. **One locus, two alleles.** A heterozygous Aa during meiosis produces two kinds of gametes, A and a, by the law of segregation.

Step 2. **Add a second independently-assorting locus.** Genotype $AaBb$ produces gametes $AB,\ Ab,\ aB,\ ab$. That is $2 \times 2 = 4$ kinds because the choice at A/a is independent of the choice at B/b (Mendel's second law).

Step 3. **Generalise.** For n heterozygous loci, the **independence rule** gives

$$N = 2 \cdot 2 \cdots 2 \ (n \text{ factors}) = 2^n.$$

Step 4. **Plug in $n = 4$.**

$$N = 2^4 = 16.$$

Step 5. **Reality check.** If two of the four loci were linked on the same chromosome, the number would be *less* than 16 – at the extreme of complete linkage between two pairs, the count could drop to $2 \cdot 2 \cdot 2 = 8$ or even 4. The 2^n answer assumes independent assortment, as the question implicitly does.

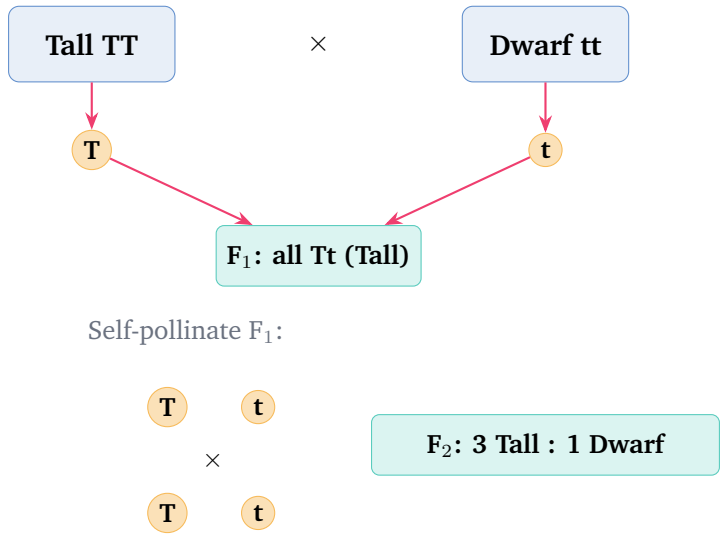
Why this matters. This is the same logic that gives the 2^{23} different chromosome-only gametes a human can produce. The genetic variety that drives evolution is, in large part, this combinatorial explosion.

> **Final Answer:** $\mathbf{2^4 = 16}$ gamete types, assuming the four loci assort independently.

Q 4.4 Explain the Law of Dominance using a monohybrid cross.

SOLUTION

Concept used. **Mendel's Law of Dominance** states three things, which we use a monohybrid cross to display: (1) characters are controlled by discrete units called **factors** (now alleles), which occur in pairs; (2) when the two factors in a pair are different, one (the **dominant** factor) expresses itself and masks the other (the **recessive** factor); and (3) the masked factor is not lost – it can reappear in the next generation. A **monohybrid cross** is a cross between parents that differ in one character. We use pea-plant height as the textbook example: true-breeding tall (TT) crossed with true-breeding dwarf (tt).



Step 1. **Parents (P).** Cross true-breeding tall (TT, only gamete T) with true-breeding dwarf (tt, only gamete t).

Step 2. **F_1 generation.** All offspring inherit one T and one t, so all F_1 are Tt. Their phenotype is tall – identical to one parent. The dwarf factor has not blended; it is simply masked. This is the visible meaning of dominance.

Step 3. **F_2 generation (self-pollinated F_1).** The cross $Tt \times Tt$ is set up in a 2×2 Punnett square. The T and t gametes from each parent combine independently. Genotypes: $1\,TT : 2\,Tt : 1\,tt$. Phenotypes: 3 tall (genotype TT or Tt) and 1 dwarf (genotype tt).

Step 4. **Conclusions.** (a) Only one of the two factors in a heterozygote shows up – the dominant one (here T). (b) The masked recessive factor was not lost; it reappears in F_2 as tt dwarf. (c) The classic 3:1 phenotypic ratio in F_2 is the experimental signature of dominance.

> **Final Answer:** In $TT \times tt$, F_1 is uniformly tall (Tt); on selfing, F_2 gives Tall:Dwarf $= 3 : 1$. The dominant allele T masks the recessive t in the heterozygote but t is preserved intact and reappears.

♥ What dominance does NOT mean

Dominance does *not* mean the dominant allele is "stronger" or more common in the population. It only describes which allele's phenotype shows up in a heterozygote. The recessive allele is chemically untouched and can be passed on for many generations silently before resurfacing in a homozygote.

EXPERT'S SOLUTION *: Aditi Mehta, Ph.D Molecular Biology, NCBS Bangalore*

Picture-first angle. The cleanest way to talk about the Law of Dominance is to walk through the Punnett square explicitly, because every claim of the law shows up there.

Step 1. **Statement of the law.** Mendel's Law of Dominance has three parts: (i) characters are controlled by discrete factors (alleles) that occur in pairs in the diploid; (ii) when the two alleles in a pair are different, one (the dominant) expresses its phenotype and masks the other (the recessive); and (iii) the masked recessive factor is not lost or blended – it is preserved unchanged and can reappear in later generations.

Step 2. **Set up the parents.** Take pea-plant height. Tall is controlled by allele T, dwarf by allele t. True-breeding tall $= TT$; true-breeding dwarf $= tt$. Both parents are homozygous, so each produces only one type of gamete: the tall parent gives only T, the dwarf parent gives only t.

Step 3. **F_1 from $TT \times tt$.** Each parent contributes one allele. The only possible F_1 genotype is Tt. All F_1 plants are phenotypically tall, identical to the tall parent – the dwarf phenotype has vanished from this generation. *Inference 1:* T dominates t, and there is no blending – the F_1 is not "medium height".

Step 4. **Self the F_1: $Tt \times Tt$.** Each Tt parent makes $\frac{1}{2}\,T$ and $\frac{1}{2}\,t$ gametes. The 2×2 Punnett square is:

	T	t
T	TT	Tt
t	Tt	tt

Genotypic ratio $TT : Tt : tt = 1 : 2 : 1$; phenotypic ratio Tall : Dwarf $= 3 : 1$, because TT and Tt both show the dominant tall phenotype while only tt shows dwarf.

Step 5. **Read off the three claims of the law from the F_1 and F_2 data.**

- Factors come in pairs and segregate cleanly into gametes (the rows and columns of the square encode this).
- In the heterozygote, only the dominant factor's phenotype appears (F_1 is uniformly tall, never intermediate).
- Masked recessive factors are preserved and reappear whenever two recessive alleles meet (tt dwarfs in F_2 at frequency $1/4$, looking identical to the original dwarf grandparent).

Step 6. **Quantitative check.** Mendel counted 787 tall and 277 dwarf F_2 plants (~ 2.84:1, very close to the theoretical 3:1). The closeness of count to ratio is the decisive proof that segregation is particulate, not blending.

Why this matters. The 3:1 ratio is the smoking gun for particulate inheritance. A blending theory would predict that Tt is intermediate and that F_2 all looks similar;

instead we see sharp segregation and the original parental phenotypes reappearing in F_2. Mendel's quantitative count nailed this and made genetics a quantitative science.

> **Final Answer:** Cross $TT \times tt \rightarrow F_1$ all Tt (tall); selfing gives F_2 in $3:1$ tall:dwarf and $1{:}2{:}1$ genotype, demonstrating that T is dominant, t is recessive, and the recessive factor is preserved unchanged through the F_1 heterozygote.

Q 4.5 Define and design a test-cross.

SOLUTION

Concept used. A dominant phenotype can hide either of two genotypes: *homozygous* dominant (TT) or *heterozygous* (Tt). Looking at the plant tells you the phenotype but not which of the two genotypes it carries. A **test cross** is the controlled mating designed to expose this: cross the individual of unknown genotype with a known *homozygous recessive* (tt), and read off the genotype from the progeny phenotypes.

Step 1. **Definition.** A test cross is a cross between an individual showing the dominant phenotype (genotype TT or Tt – to be determined) and an individual that is homozygous recessive (tt).

Step 2. **Logic of the design.** The homozygous recessive parent contributes only t gametes. Therefore the phenotype of every offspring is determined entirely by the allele the test parent contributed.

- If the test parent is TT, every gamete is T, so every offspring is Tt – *all tall*.
- If the test parent is Tt, gametes are half T and half t, so offspring are half Tt (tall) and half tt (dwarf) – a $1{:}1$ ratio.

Step 3. **Design (worked example with pea).** Suppose you have a tall pea plant whose genotype is unknown. Cross it with a dwarf pea plant (tt). Collect at least ~20 seeds (more is better for statistics), grow them and score the heights.

Step 1. **Read off the result.**

- If *all* offspring are tall, the test parent was TT.
- If offspring are *half tall, half dwarf* ($1{:}1$), the test parent was Tt.

> **Final Answer:** A test cross is "dominant phenotype $\times$ homozygous recessive". All-dominant progeny $\Rightarrow$ test parent is TT; $1{:}1$ dominant:recessive progeny $\Rightarrow$ test parent is Tt.

☞ Exam Tip

Two-mark NCERT favourite. To pick up both marks, your answer needs (i) a one-line *definition* ("cross with homozygous recessive") and (ii) the two *outcomes* that distinguish TT from Tt (all-dominant vs. $1{:}1$). A Punnett square for either case bags a bonus mark in board exams.

EXPERT'S SOLUTION *: Pranav Banerjee, M.Sc Botany, Delhi University*

Strategic angle. The test cross is the original phenotype-to-genotype assay. Today's molecular tools (PCR, sequencing) can read genotype directly, but the test cross still gets asked because it shows the logic without needing a gel.

Step 1. **Why tt and not just any plant.** The point of choosing a tt tester is that t is "silent" in the offspring: the recessive parent never adds a dominant allele of its own to muddy the read-out. Every offspring's phenotype directly tells you which allele the *other* parent gave.

Step 2. **Probabilistic reading.** If the test parent is Tt, you expect $1{:}1$ but you may see, say, $11{:}9$ in a small sample – still a Tt. If you see 20 tall and 0 dwarf, the parent is almost certainly TT (the chance of $Tt \times tt$ giving 20 talls in a row is $(1/2)^{20} \approx 10^{-6}$, so a TT call is statistically safe).

Step 3. **Beyond one locus.** A test cross also works for dihybrid analysis. $TtRr \times ttrr$ gives the four phenotypes in $1{:}1{:}1{:}1$ ratio if loci assort independently, and a skewed ratio if they are linked. So the test cross is also Morgan's tool for detecting linkage and measuring map distance.

Step 4. **Worked design (pea height).** You hold a tall pea ($T?$, genotype unknown). To diagnose: emasculate flowers on a known dwarf (tt) tester and dust the tall plant's pollen onto each emasculated stigma. Sow at least $20{-}40$ resulting seeds, grow them through to flowering, and score height. The observed ratio reveals the parent's genotype: $\approx 1{:}1$ tall:dwarf $\Rightarrow Tt$; all tall $\Rightarrow TT$.

Step 5. **Why the recessive tester is critical.** A TT tester would give all tall progeny regardless of the parent's genotype, so it provides zero diagnostic information. Only the homozygous recessive tester lets the parent's gametes write themselves directly onto the offspring's phenotype.

Why this matters. A test cross converts an invisible question (which genotype?) into a visible one (which phenotype ratio?). That trick of designing an experiment to make the

unknown visible is the heart of genetics.

> **Final Answer:** Test cross = unknown $\times tt$. Progeny phenotypes distinguish TT (all dominant) from Tt (1:1 split), and the same design generalises to multi-locus crosses for linkage analysis.

Q 4.6 **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

Concept used. A **Punnett square** is a grid that sets the gametes of one parent along one axis and those of the other parent along the other axis; each interior cell shows one possible offspring genotype, weighted equally because each gamete combination is equally likely. To use it we need each parent's gamete types.
The question is open about *which* homozygote the female is; the most-cited NCERT reading is "homozygous recessive" so we illustrate that case (Mendel's standard test-cross configuration). The dominant-homozygote case is included below as a check.

Step 1. **Pick a locus.** Use pea-plant height: tall (T, dominant) and dwarf (t, recessive).

Step 2. **Identify the parents' genotypes.**

- Homozygous female (recessive case): tt.
- Heterozygous male: Tt.

Step 3. **List each parent's gametes.**

- Female (tt): all gametes are t.
- Male (Tt): half gametes are T, half are t.

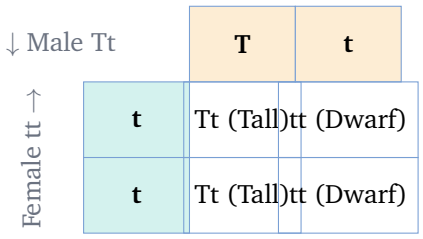
Step 4. **Build the Punnett square.**

	T	t
t	Tt	tt
t	Tt	tt

Four equally-likely cells: $2\,Tt$ and $2\,tt$.

Step 5. **Genotypic ratio.** $Tt : tt = 2 : 2 = 1 : 1$.

Step 6. **Phenotypic ratio.** Tt is tall (dominant shows), tt is dwarf. So Tall : Dwarf $= 1 : 1$, i.e. 50% tall, 50% dwarf.



Final Answer: F_1 from tt (female) $\times Tt$ (male) shows **1 Tall : 1 Dwarf** phenotypic ratio (50% : 50%); genotypic ratio is also $1\,Tt : 1\,tt$.

☞ Exam Tip

If the examiner reads "homozygous" as homozygous *dominant* (TT), the cross becomes $TT \times Tt$, gametes are T, T from the female and T, t from the male, the square fills with $\frac{1}{2}\,TT + \frac{1}{2}\,Tt$, and *all* F_1 are tall phenotypically (genotypic ratio $1\,TT : 1\,Tt$, phenotypic ratio $1 : 0$). Mention this case in your answer if the question is ambiguous – it shows you've covered both readings.

EXPERT'S SOLUTION *: Riya Joshi, M.Sc Botany, Delhi University*

Reading the question. "Homozygous female" can mean TT or tt. The standard NCERT model answer uses the recessive homozygote (tt) because that turns the cross into a **test cross** of the male – the most informative interpretation. We illustrate that case here and note the alternative below.

Step 1. **Identify parental genotypes.** Female: homozygous, taken as tt (the standard recessive-homozygote reading). Male: heterozygous, Tt. We track one trait, plant height, with allele convention T = tall (dominant), t = dwarf (recessive).

Step 2. **Gametes from each parent.** The female tt produces a single kind of gamete carrying t, with probability 1. The male Tt produces two kinds of gametes: T with probability $1/2$ and t with probability $1/2$ (Mendel's law of segregation operating on his two alleles during meiosis).

Step 3. **Build the Punnett square (2×2).** Place female gametes (t, t – both rows are t because she is tt) on the rows, and male gametes (T, t) on the columns:

	T	t
t	Tt	tt
t	Tt	tt

The four equally-likely cells contain Tt, tt, Tt, tt, i.e. two Tt and two tt offspring out of four.

Step 4. **Genotypic ratio.** $Tt : tt = 2 : 2 = 1 : 1$, so half the offspring are heterozygous tall and the other half are homozygous dwarf.

Step 5. **Phenotype mapping.** A Tt offspring carries the dominant T allele and so expresses the tall phenotype; a tt offspring expresses dwarf. Phenotypic ratio is therefore 1 tall : 1 dwarf, or 50% tall and 50% dwarf.

Step 6. **Alternative reading.** If the question is read as the female being homozygous *dominant* (TT), then the cross $TT \times Tt$ gives gametes T, T from the female and T, t from the male, yielding $1\,TT : 1\,Tt$ (genotype) and all-tall (phenotype, ratio $1{:}0$). Worth mentioning in your answer; the test-cross reading ($tt \times Tt$) is the one most textbooks adopt because it produces a useful $1{:}1$ ratio rather than a uniform phenotype.

Why this matters. The $1{:}1$ split from a heterozygote crossed to a homozygous recessive is exactly Mendel's test-cross prediction, and is the simplest way to identify a heterozygote in the lab. The same logic generalises to dihybrid test crosses (which give $1{:}1{:}1{:}1$) and underpins all of classical genetic analysis.

> **Final Answer:** Punnett square of $tt \times Tt$ gives $1\,Tt : 1\,tt$, so phenotypically 1 Tall : 1 Dwarf in F_1. (If the female were TT instead, the cross $TT \times Tt$ would give all tall progeny.)

Q 4.7 **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

Concept used. For two genes that assort independently (Mendel's second law), the probability that an offspring has a specific *combination* of phenotypes equals the *product* of the probabilities for each gene considered separately. So instead of building a 16-cell Punnett square, we split the dihybrid cross into two monohybrid crosses and multiply. Conventions: T = tall (dominant), t = dwarf (recessive); Y = yellow seed (dominant), y = green seed (recessive).

Step 1. **Split into two monohybrid crosses.**

- Height: $Tt \times Tt$.
- Seed colour: $Yy \times yy$.

Step 2. **Resolve the height cross $Tt \times Tt$ (Punnett).**

	T	t
T	TT	Tt
t	Tt	tt

Genotype ratio $TT : Tt : tt = 1 : 2 : 1$; phenotype ratio Tall : Dwarf $= 3 : 1$. So:

$$P(\text{Tall}) = \tfrac{3}{4}, \quad P(\text{Dwarf}) = \tfrac{1}{4}.$$

Step 3. **Resolve the seed-colour cross $Yy \times yy$ (Punnett, this is a test-cross of the heterozygote).**

	Y	y
y	Yy	yy
y	Yy	yy

Phenotype ratio Yellow : Green $= 1 : 1$. So:

$$P(\text{Yellow}) = \tfrac{1}{2}, \quad P(\text{Green}) = \tfrac{1}{2}.$$

Step 4. **Combine by independence (Mendel's second law).** For unlinked loci,

$$P(\text{combo}) = P(\text{height}) \times P(\text{seed colour}).$$

Step 5. **Part (a): Tall and Green.**

$$P(\text{Tall AND Green}) = \tfrac{3}{4} \times \tfrac{1}{2} = \tfrac{3}{8}.$$

That is, 3 out of every 8 offspring (or 37.5%) are expected to be tall with green seeds.

Step 6. **Part (b): Dwarf and Green.**

$$P(\text{Dwarf AND Green}) = \tfrac{1}{4} \times \tfrac{1}{2} = \tfrac{1}{8}.$$

That is, 1 out of every 8 offspring (or 12.5%) are expected to be dwarf with green seeds.

Step 7. **Sanity check.** Adding all four combinations:
$\frac{3}{8}(\text{TG}) + \frac{3}{8}(\text{TY}) + \frac{1}{8}(\text{DG}) + \frac{1}{8}(\text{DY}) = \frac{8}{8} = 1$. ✓ All offspring accounted for.

Final Answer: (a) Tall and Green = **3/8** (37.5%). (b) Dwarf and Green = **1/8** (12.5%).

✗ Common Mistake

A common slip is to think this is a standard $9{:}3{:}3{:}1$ dihybrid. It is not – only one parent is heterozygous for seed colour. So the height cross is a Mendel monohybrid ($3{:}1$) but the seed-colour cross is a test cross ($1{:}1$). Always identify the *individual* cross at each locus before multiplying.

EXPERT'S SOLUTION *: Sneha Verma, M.Sc Biotechnology, AIIMS Delhi*

Quick reading. The trick this question wants is the "split-into-two" approach. The full 4×4 Punnett with sixteen boxes is exhausting; multiplying single-locus probabilities is faster and less error-prone.

Step 1. **Decode each parent.** Father $TtYy$ contributes one T/t allele and one Y/y allele to each gamete. Mother $Ttyy$ contributes one T/t allele and (always) one y allele. So at the height locus this is $Tt \times Tt$ (Mendel monohybrid), and at the seed-colour locus this is $Yy \times yy$ (a test cross of the Yy heterozygote).

Step 2. **Locus 1 (T/t):** $Tt \times Tt$**.** Standard monohybrid Punnett gives genotype ratio $1\,TT : 2\,Tt : 1\,tt$ and phenotype ratio Tall:Dwarf $= 3{:}1$.

$$P(\text{Tall}) = \tfrac{3}{4}, \quad P(\text{Dwarf}) = \tfrac{1}{4}.$$

Step 3. **Locus 2 (Y/y):** $Yy \times yy$**.** A test cross of the heterozygote: gametes from Yy are $\frac{1}{2}\,Y$ and $\frac{1}{2}\,y$; gametes from yy are all y. So offspring are $\frac{1}{2}\,Yy$ (yellow) and $\frac{1}{2}\,yy$ (green).

$$P(\text{Yellow}) = \tfrac{1}{2}, \quad P(\text{Green}) = \tfrac{1}{2}.$$

Step 4. **Independence rule (Mendel's second law).** The two genes (height, seed colour) assort independently, so the joint probability of a height-and-colour combination equals the product of the two single-locus probabilities:

$$P(\text{phen}_1 \cap \text{phen}_2) = P(\text{phen}_1) \cdot P(\text{phen}_2).$$

Step 5. **Apply to the two parts.**

- Tall & Green: $P = \frac{3}{4} \cdot \frac{1}{2} = \frac{3}{8}$, i.e. 37.5% of offspring.
- Dwarf & Green: $P = \frac{1}{4} \cdot \frac{1}{2} = \frac{1}{8}$, i.e. 12.5% of offspring.

Step 6. **Full distribution for reference (all four combinations).**

- Tall + Yellow: $\frac{3}{4} \cdot \frac{1}{2} = \frac{3}{8}$.
- Tall + Green: $\frac{3}{4} \cdot \frac{1}{2} = \frac{3}{8}$.
- Dwarf + Yellow: $\frac{1}{4} \cdot \frac{1}{2} = \frac{1}{8}$.
- Dwarf + Green: $\frac{1}{4} \cdot \frac{1}{2} = \frac{1}{8}$.

Ratio $3{:}3{:}1{:}1$; sum $= 1$ (every offspring is in exactly one class).

Why this matters. Multiplying single-locus probabilities is exactly what every dihybrid question (and trihybrid, and pedigree risk calculation) reduces to. Master this and the $9{:}3{:}3{:}1$ Mendelian dihybrid ratio becomes just the four products $(3/4)\cdot(3/4) : (3/4)\cdot(1/4) : (1/4)\cdot(3/4) : (1/4)\cdot(1/4)$. The same logic also explains why this question deviates from $9{:}3{:}3{:}1$: only ONE parent is heterozygous at the seed-colour locus, so the colour ratio is $1{:}1$ (test cross), not $3{:}1$ (monohybrid).

> **Final Answer:** Tall & Green $= 3/8$; Dwarf & Green $= 1/8$. Full F_1 distribution: $3{:}3{:}1{:}1$ for TY:TG:DY:DG.

Q 4.8 **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

Concept used. Linkage is the tendency of two genes located close together on the same chromosome to be inherited together because crossing-over between them is rare. Linkage breaks Mendel's law of independent assortment, so the F_2 phenotypic ratio departs from the classical $9{:}3{:}3{:}1$ expected for unlinked loci. Note: NCERT phrases the question in terms of "F_1 generation" but the segregating progeny analysed below are the offspring of two heterozygotes – what most textbooks (including NCERT itself in Morgan's experiment) draw as F_2. We work that case.

Step 1. **The cross.** Two dihybrid heterozygotes $AaBb \times AaBb$, where the loci A and B are linked (on the same chromosome). Suppose A and B entered from one parent (cis arrangement AB/ab).

Step 2. **Gametes when loci are linked.** If linkage is *complete* (no crossing over), each parent produces only two gamete types: the parental ones, AB and ab. The recombinant gametes Ab and aB do not form.

Step 3. **F_2 ratio with complete linkage.** The cross $AB/ab \times AB/ab$ behaves like a monohybrid in disguise:

	AB	ab
AB	$AABB$	$AaBb$
ab	$AaBb$	$aabb$

Phenotype counts: 3 "$A\_B\_$" : 1 "$aabb$", i.e. the *parental* combinations only. **No recombinant phenotypes appear.** The ratio is $3:1$, not $9:3:3:1$.

Step 4. **F_2 ratio with incomplete linkage.** Real linkage is rarely complete; a small fraction of gametes recombine.

- **Parental phenotypes** (AB and ab classes): frequent – more than the $10/16$

expected from independent assortment.

- **Recombinant phenotypes** (Ab and aB classes): rare – less than the $6/16$ expected from independent assortment.

So the four phenotypes appear in a ratio close to $3:1$ for the parental classes (heavily over-represented), with a small frequency of the two recombinant classes proportional to the **map distance** between the loci.

Step 5. **Morgan's data.** In Drosophila, Morgan crossed $y^+w^+/y\,w$ (yellow body, white eye genes linked on X) and saw far more parental phenotypes than recombinants – the F_2 ratio deviated sharply from $9:3:3:1$. The deviation *is* the discovery of linkage.

Final Answer: When the two loci are linked, the F_2 phenotypic distribution departs from Mendel's $9:3:3:1$. With complete linkage only parental phenotypes appear in a $3:1$ ratio; with partial linkage the parental phenotypes still predominate and the two recombinant phenotypes are present only at low, equal frequency proportional to the genetic distance between the loci.

♥ Linkage maps

The very rarity of recombinant phenotypes is informative. By measuring the recombination frequency (RF = recombinants / total), geneticists construct **genetic maps**: 1% RF $= 1$ **centimorgan** of distance. Morgan's student Sturtevant used this principle in 1913 to draw the first linear map of genes on a chromosome.

EXPERT'S SOLUTION *: Aditya Rao, Ph.D Molecular Biology, NCBS Bangalore*

Structural angle. The key insight is that linkage cuts the number of gamete types a heterozygote can make. Independent assortment of two loci gives four gamete types in equal frequencies ($1:1:1:1$). Complete linkage gives only two gamete types (the parental ones, $1:1$). Real linkage sits between, with parental gametes over-represented.

Step 1. **Gametes from $AaBb$ with linkage.** In the configuration AB/ab, the parental gametes are AB and ab; recombinants are Ab and aB. Frequencies:

$$P(AB) = P(ab) = \tfrac{1-r}{2}, \quad P(Ab) = P(aB) = \tfrac{r}{2},$$

where r is the **recombination frequency** ($0 \le r \le 0.5$). $r = 0$: complete linkage; $r = 0.5$: independent assortment.

Step 2. **Phenotype frequencies in F_2.** The proportions of the four phenotypic classes follow from combining gametes from each parent and then summing genotypes that give the same phenotype. The closed forms (using $p = (1-r)/2$ for parental and $q = r/2$ for recombinant gametes) are:

- $A\_B\_$ (both dominant) : $1 - 2q^2$.
- A_bb (parental Ab if cis was AB/ab, otherwise recombinant) and $aaB\_$: each $\frac{1}{4} -$ adjustment.
- $aabb$ (homozygous recessive) : p^2.

The numbers are messy. The important qualitative outcome is: *parental phenotypes much* $> 9/16$ *and* $1/16$*, recombinant phenotypes much* $< 3/16$ *each*.

Step 3. **Limit cases.**

- $r = 0$ (complete linkage): F_2 phenotype ratio $3 : 0 : 0 : 1$ (parental dominant : recombinant : recombinant : parental recessive). That is the $3 : 1$ from above.
- $r = 0.5$ (independent assortment): F_2 phenotype ratio $9 : 3 : 3 : 1$ – Mendel's classical dihybrid ratio.
- $0 < r < 0.5$ (real linkage): ratio between the two, with parental classes over-represented.

Why this matters. Linkage is the first place Mendelian genetics needed an upgrade. Morgan's discovery – that loci on the same chromosome don't always assort independently – pinned genes physically to chromosomes for the first time.

> **Final Answer:** Linked loci produce mostly parental-type F_2 progeny (approaching a $3 : 1$ Mendelian monohybrid ratio in the limit of complete linkage); recombinant phenotypes are rare, with frequency determined by the map distance between the loci, so the classical $9 : 3 : 3 : 1$ dihybrid ratio fails.

Q 4.9 Briefly mention the contribution of T.H. Morgan in genetics.

SOLUTION

Concept used. Thomas Hunt Morgan (1866–1945, USA) was the geneticist who took Mendel's abstract "factors" and pinned them to physical chromosomes. He worked with the fruit fly *Drosophila melanogaster* – an organism almost as good for genetics as pea was for Mendel: short life cycle ($\sim$2 weeks), many offspring per cross, only four pairs of chromosomes, and easy identification of mutant phenotypes (eye colour, body colour, wing shape).

Step 1. **Discovery of linkage (1910–1911).** While studying white-eyed mutant flies, Morgan noticed that two genes on the same chromosome do *not* assort independently; they tend to be inherited together. He coined the term **linkage** for this physical association of genes on a chromosome.

Step 2. **Recombination and crossing-over.** Morgan also observed that linked genes can occasionally be separated: rare offspring carrying new (recombinant) gene combinations appeared. He attributed this to **crossing-over** between homologous chromosomes during meiosis, an idea confirmed by his student Stern with cytological evidence.

Step 3. **Genetic mapping.** Morgan and his student Alfred Sturtevant realised that the frequency of recombination between two loci reflects their physical distance on the chromosome: closer loci recombine less. In 1913, Sturtevant drew the first **genetic linkage map** of a chromosome (the X chromosome of *Drosophila*), ordering six genes by recombination frequency. The unit of 1% recombination was later named the **centimorgan** in Morgan's honour.

Step 4. **Sex-linked inheritance.** Morgan's white-eye mutant was inherited in a pattern that differed in males and females. He showed the gene was carried on the X chromosome and traced the first **sex-linked** inheritance pattern in animals.

Step 5. **Chromosomal theory consolidated.** Although Sutton and Boveri had proposed the chromosomal theory of inheritance in 1902, Morgan's experimental work on *Drosophila* provided the decisive evidence that *genes are located on chromosomes* and behave as physically linked units.

Step 6. **Recognition.** Morgan won the Nobel Prize in Physiology or Medicine in 1933 for his discoveries concerning the role of chromosomes in heredity.

> **Final Answer:** Morgan, working on *Drosophila melanogaster*, discovered linkage and crossing-over, identified sex-linked inheritance, and (with Sturtevant) introduced genetic mapping – collectively giving experimental backbone to the chromosomal theory of inheritance, for which he received the 1933 Nobel Prize.

☞ Exam Tip

A frequent 2- or 3-mark question. Write Morgan's contributions as a numbered list: (i) chose *Drosophila* as the model organism, (ii) discovered linkage, (iii) discovered crossing-over, (iv) demonstrated sex-linked inheritance, (v) won the 1933 Nobel Prize. Naming the organism and the Nobel year shows command of the detail examiners reward.

EXPERT'S SOLUTION *: Krishna Pillai, Ph.D Molecular Biology, NCBS Bangalore*

Strategic angle. Treat Morgan's contributions as four linked discoveries, each pushing genetics forward by one step.

Step 1. **Choice of model organism.** Morgan picked *Drosophila* for the same reasons Mendel picked pea: short generation time, large progeny, easy to score

phenotypes. This choice itself opened up high-throughput genetics.

Step 2. **Linkage.** Genes on the same chromosome tend to travel together; Mendel's second law fails for them. Morgan demonstrated this with cross data showing departures from $9{:}3{:}3{:}1$.

Step 3. **Crossing-over and mapping.** The exceptions to linkage (rare recombinants) are themselves systematic: their frequency measures distance. Sturtevant turned this into the first linear gene map.

Step 4. **Sex linkage.** Morgan's white-eye fruit-fly mutant was inherited differently in males and females because the gene sits on the X chromosome. This was the first time a specific gene was localised to a specific chromosome – the moment "factor on a chromosome" stopped being a metaphor and became a measurable physical fact.

Step 5. **Confirmation of the chromosomal theory.** Sutton and Boveri had *proposed* the chromosomal theory of inheritance in 1902, but it was an inference, not a proof. Morgan's experimental data on linkage, crossing-over and sex-linked white eye were the decisive evidence that Mendelian factors live on chromosomes and behave according to chromosome behaviour during meiosis.

Step 6. **Lab legacy.** The "fly room" at Columbia that Morgan ran trained a generation of geneticists (Sturtevant, Bridges, Muller – the last won his own Nobel for showing X-rays cause mutations), and the cM unit of genetic distance is named the centiMorgan after him. Modern *Drosophila* biology is the direct descendant of his programme.

Why this matters. Morgan turned genetics from a body of abstract laws into a chromosome-anchored experimental science. The work also legitimised *Drosophila* as the central "workhorse" organism that still drives modern developmental genetics.

> **Final Answer:** Discovery of linkage, recognition of crossing-over, the first genetic linkage map, the first sex-linked gene – Morgan's *Drosophila* programme physically pinned Mendelian factors to chromosomes (Nobel Prize 1933).

Q 4.10 What is pedigree analysis? Suggest how such an analysis can be useful.

SOLUTION

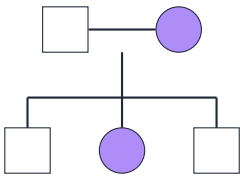
Concept used. You can't run breeding experiments on humans; generation time is too long, family size is small and ethics forbids it. **Pedigree analysis** is the workaround: you draw a family tree showing which members across several generations expressed a

given trait, then deduce the inheritance pattern (dominant/recessive, autosomal/sex-linked) from how the trait travels through the tree.

Step 1. **Definition.** A pedigree is a chart that records the history of a heritable character through generations of a family, using standard symbols: square for male, circle for female, filled symbol for affected individuals, horizontal line for mating, vertical line for descent, Roman numerals for generations, Arabic numerals for individuals within a generation.

Step 2. **Inference rules.**

- *Autosomal dominant* traits appear in every generation, with affected fathers passing the trait to about half their children, sons and daughters equally (e.g. Huntington's chorea, Myotonic dystrophy).
- *Autosomal recessive* traits often skip generations, requiring two carrier parents to produce an affected child; sons and daughters equally affected (e.g. sickle-cell anaemia, phenylketonuria).
- *X-linked recessive* traits show many more affected males than females; an affected mother's sons are affected; no male-to-male transmission (e.g. haemophilia, colour blindness).
- *X-linked dominant* traits: affected fathers transmit to all daughters but no sons.



Affected female (carrier of dominant trait)
Square = male, Circle = female
Filled = trait expressed
Horizontal = mating; Vertical = descent

Step 1. **Uses of pedigree analysis.**

1. **Tracing the inheritance pattern** of a particular trait (dominant vs. recessive, autosomal vs. sex-linked).
2. **Predicting probabilities** that future children will be affected or carriers – used in **genetic counselling** for couples with a family history of disease.
3. **Identifying carriers** of recessive diseases in unaffected family members.
4. **Estimating risk** for late-onset disorders (Huntington's) before symptoms appear.
5. **Tracking source of mutation** when a new trait suddenly appears in a family.

Final Answer: Pedigree analysis = study of a heritable character across generations of a family using standard symbols. It identifies the mode of inheritance, predicts the probability of affected offspring, identifies carriers and underpins genetic counselling.

✗ Common Mistake

A frequent slip is to treat "pedigree analysis" as just a family tree drawing. The chart is only half the work. The other half is *inference*: deducing whether the trait is dominant or recessive, autosomal or sex-linked, by reading the pattern of affected and unaffected individuals across generations.

EXPERT'S SOLUTION *: Diya Kapoor, M.Sc Biotechnology, AIIMS Delhi*

Strategic angle. A pedigree is genetics done backwards – instead of crossing to predict outcomes, you read outcomes to deduce the cross.

Step 1. **Read the symbols.** Squares males, circles females, filled = affected; horizontal line = couple, vertical drop = offspring, Roman numerals for generations.

Step 2. **Spot the signatures.** The trait's distribution in the tree betrays its mode of inheritance:

- Every generation, no skipping $\Rightarrow$ likely dominant.
- Generations are skipped $\Rightarrow$ likely recessive.
- Males disproportionately affected $\Rightarrow$ likely X-linked recessive.
- Father-to-son transmission seen $\Rightarrow$ rules out X-linked (rules *in* autosomal or Y-linked).

Step 3. **Apply to counselling.** Once the pattern is known, probabilities for future children can be computed. For example, two unaffected carrier parents of an autosomal recessive disease ($Aa \times Aa$) have a $1/4$ chance per pregnancy of an affected child.

Step 4. **Practical impact.** Pedigrees are the entry point to clinical genetics – they guide which gene to test, which family members to screen, and which pregnancies merit prenatal testing.

Step 5. **Worked counselling example.** Consider a couple in which both partners are unaffected but each had a sibling with sickle-cell anaemia. The pedigree implies both partners are carriers ($HbA\,HbS$) with prior probability $2/3$ each. For a child to be affected, both parents must transmit HbS – probability $(2/3)(2/3)(1/4) = 4/36 \approx 11\%$ per pregnancy. A pedigree converts a vague worry into a concrete number that doctors and families can act on.

Step 6. **Limits of the method.** Pedigrees fail when families are small (no statistics), when penetrance is incomplete (gene present but trait absent), and when the trait is sporadic or polygenic. In those cases molecular tests must complement the family tree.

Why this matters. Before genome sequencing was cheap, the pedigree was the only diagnostic tool a clinical geneticist had. Even now, a clean pedigree narrows the genetic

test down from ∼20,000 genes to a handful, which is the difference between ordering one focused test and sequencing the whole exome.

> **Final Answer:** Pedigree analysis charts a heritable trait through a family and from that infers its inheritance mode (dominant vs. recessive, autosomal vs. X-linked), predicts risk for future generations, identifies silent carriers and informs genetic counselling.

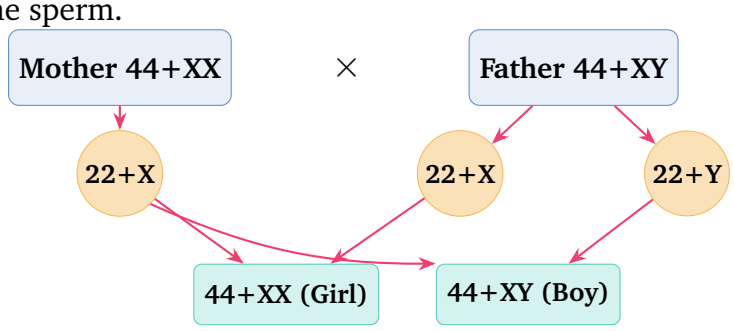
Q 4.11 How is sex determined in human beings?

SOLUTION

Concept used. Human sex determination follows the **XX-XY system**. Of the 46 chromosomes in a human somatic cell, 22 pairs are **autosomes** (the same in males and females) and one pair is **sex chromosomes**.

- Female karyotype: $44\,(autosomes) + XX = 46$.
- Male karyotype: $44\,(autosomes) + XY = 46$.

Because the male carries two different sex chromosomes (X and Y), he is the **heterogametic** sex; the female (XX) is **homogametic**. Sex of the child is therefore determined by the sperm.



Step 1. **Gamete formation.**

- In the female (XX), meiosis produces *one* kind of egg: each ovum carries 22 autosomes plus one X chromosome.
- In the male (XY), meiosis produces *two* kinds of sperm in equal numbers: half carry 22 autosomes + X, half carry 22 autosomes + Y.

Step 2. **Fertilisation.**

- Egg $(22 + X)$ + X-bearing sperm → zygote $44 + XX$ → daughter.
- Egg $(22 + X)$ + Y-bearing sperm → zygote $44 + XY$ → son.

Step 3. **The sex of the baby is determined by the father's sperm.** Because 50% of sperm carry X and 50% carry Y, the chance of a son at each conception is $1/2$

and of a daughter is $1/2$. (Cultural blame on mothers for having only daughters is biologically wrong.)

Step 4. **Role of the Y chromosome.** The Y chromosome carries the **SRY gene** (Sex-Determining Region Y), which triggers male development by directing the indifferent gonad to become a testis. Without SRY (and hence the Y), the embryo develops as female.

> **Final Answer:** Human sex follows the XX–XY mechanism: females are $44 + XX$, males are $44 + XY$. The father is heterogametic, so the sperm determines the child's sex – X-bearing sperm gives a girl, Y-bearing sperm gives a boy, each with probability $1/2$.

♥ Genotype ratios are 1:1

Because half of all sperm are X-bearing and half are Y-bearing, the expected sex ratio at conception is $1{:}1$. Slight deviations observed at birth (~ 1.05 boys per girl in many populations) arise from differences in survival of XY vs. XX embryos in the womb, not from biased sperm production.

EXPERT'S SOLUTION *: Yash Chatterjee, M.Sc Microbiology, JNU*

Quick reading. The cleanest way to write this answer is: identify the heterogametic sex, list the gametes each parent makes, combine them, and note the role of SRY.

Step 1. **Karyotypes.** Humans have 46 chromosomes $= 22$ pairs of autosomes $+1$ pair of sex chromosomes. In females the sex pair is XX; in males it is XY.

Step 2. **Gametes.**

$$\text{Mother (XX)} \longrightarrow \text{all eggs: } 22\,\text{aut} + X.$$

$$\text{Father (XY)} \longrightarrow \tfrac{1}{2}\,(22 + X) \text{ and } \tfrac{1}{2}\,(22 + Y) \text{ sperm.}$$

Step 3. **Fertilisation outcomes.**

- $X_{\text{egg}} \times X_{\text{sperm}} \to 44 + XX \to$ daughter (probability $1/2$).
- $X_{\text{egg}} \times Y_{\text{sperm}} \to 44 + XY \to$ son (probability $1/2$).

Step 4. **Genetic switch.** The Y chromosome carries the **SRY gene**; its product triggers testis development in the embryo. Absent SRY, the default developmental path is female.

Step 5. **Sex ratio.** Expected $1{:}1$ male:female because the two sperm classes are equally frequent. Actual birth ratios run slightly male-skewed (~ 1.05 boys per girl in most populations) due to subtle differences in fetal survival, not biased sperm production.

Step 6. **Contrast with other systems.** The XX/XY pattern is not universal: birds use ZW (females ZW, males ZZ – female heterogametic), grasshoppers use XO (females XX, males X–), and honeybees use haplodiploidy (females diploid, males haploid). NCERT mentions these alongside XX/XY to show that sex determination is mechanism-dependent.

Step 7. **Cultural footnote.** Because the deciding chromosome sits in the sperm, blaming a mother for the sex of her children is biologically wrong; every conception has a $1/2$ chance of either outcome regardless of the mother's karyotype.

Why this matters. Because the father's sperm carries the deciding chromosome, biology unambiguously places sex determination on the male side. The misconception that "the mother is to blame" for the sex of the child is not just culturally wrong – it is biologically wrong.

> **Final Answer:** Sex in humans = XX/XY; female $44 + XX$, male $44 + XY$; father's sperm (X or Y) decides; SRY on Y triggers maleness; expected $1 : 1$ ratio.

Q 4.12 **A child has blood group O. If the father has blood group A and mother blood group B, work out the genotypes of the parents and the possible genotypes of the other offsprings.**

SOLUTION

Concept used. The ABO blood-group system in humans is controlled by a single autosomal gene with three alleles:

- I^A – produces antigen A (dominant over i).
- I^B – produces antigen B (dominant over i).
- i – produces no antigen (recessive to both I^A and I^B).

A key point: I^A and I^B are **co-dominant**; when both are present in a heterozygote (I^AI^B), both antigens A and B are produced. The mapping of genotype to phenotype is therefore:

Genotype	Blood group (phenotype)
I^AI^A, I^Ai	A
I^BI^B, I^Bi	B
I^AI^B	AB
$i\,i$	O

Step 1. **Use the child's phenotype to constrain the parents.** The child has blood group O, so the child's genotype must be ii. That means the child received an i

allele from each parent.

Step 2. **Father (blood group A) gave an i.** Father's possible genotypes for blood group A are I^AI^A or I^Ai. To pass an i to the child, father must be I^Ai (heterozygous). I^AI^A is ruled out.

Step 3. **Mother (blood group B) gave an i.** Mother's possible genotypes for blood group B are I^BI^B or I^Bi. Similarly she must be I^Bi. I^BI^B is ruled out.

Step 4. **Set up the cross.**

$$I^Ai \times I^Bi.$$

Step 5. **Punnett square.**

	I^B	i
I^A	I^AI^B	I^Ai
i	I^Bi	ii

Step 6. **Read off offspring possibilities (each with probability $1/4$).**

- I^AI^B : blood group **AB**.
- I^Ai : blood group **A**.
- I^Bi : blood group **B**.
- ii : blood group **O**.

Final Answer: Father is $\mathbf{I^Ai}$ (heterozygous A) and mother is $\mathbf{I^Bi}$ (heterozygous B). Each future child has a $1/4$ chance of being A (I^Ai), $1/4$ of being B (I^Bi), $1/4$ of being AB (I^AI^B), and $1/4$ of being O (ii). All four ABO blood groups can appear among the offspring of these parents.

☞ Exam Tip

This is a standard 3-mark NCERT question. The marker checks three things: (i) you correctly deduced both parents are heterozygous, (ii) the Punnett square is drawn, and (iii) you listed all four possible offspring groups (AB, A, B, O), not just three. Forgetting the AB group is the single most common slip.

EXPERT'S SOLUTION *: Ishita Singh, M.Sc Biotechnology, AIIMS Delhi*

Strategic angle. The question is really a small puzzle. Step backwards from the child's genotype (ii) to the alleles each parent contributed, then deduce each parent's full genotype, then cross.

Step 1. **Decode the child.** Blood O $\Rightarrow$ child is ii. So father donated an i allele and mother donated an i allele.

Step 2. **Decode the father.** Father is blood A, so his genotype has at least one I^A. He also donated an $i \Rightarrow$ his genotype is $I^A i$.

Step 3. **Decode the mother.** Mother is blood B, so her genotype has at least one I^B. She also donated an $i \Rightarrow$ her genotype is $I^B i$.

Step 4. **Cross.** $I^A i \times I^B i$ produces four equally likely genotypes:

- $I^A I^B$: phenotype AB (probability $1/4$).
- $I^A i$: phenotype A (probability $1/4$).
- $I^B i$: phenotype B (probability $1/4$).
- $i\,i$: phenotype O (probability $1/4$).

Step 5. **Surprising consequence.** A child can have a blood group (AB) that neither parent has. This is a textbook consequence of co-dominance between I^A and I^B, and it is also the reason the ABO system features so often in disputed-paternity scenarios.

Why this matters. The ABO system is the simplest worked example of **multiple alleles** and **co-dominance** operating at one locus. It's also clinically essential: ABO incompatibility is the most common cause of transfusion reactions.

> **Final Answer:** Parents: father $I^A i$, mother $I^B i$. Offspring possibilities (each $1/4$): A ($I^A i$), B ($I^B i$), AB ($I^A I^B$), O (ii).

Q 4.13 **Explain the following terms with example:**
(a) Co-dominance
(b) Incomplete dominance

SOLUTION

Concept used. In Mendel's monohybrid cross, the heterozygote shows the dominant phenotype *exactly* – the recessive allele is masked. But not all alleles behave this neatly. Two important departures are co-dominance and incomplete dominance. Both lead to F_1 phenotypes that differ from either parent.

Step 1. **(a) Co-dominance.**

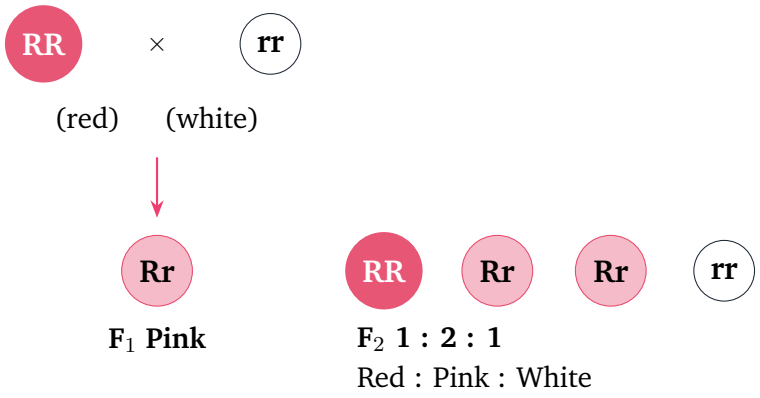
- **Definition.** When two alleles in the heterozygote both express their phenotype independently and simultaneously, with neither allele masking the other, the alleles are **co-dominant**. The heterozygote shows *both*

parental phenotypes side by side.

- **Example: ABO blood groups in humans.** The I^A and I^B alleles are co-dominant. A heterozygous I^AI^B individual produces *both* the A antigen and the B antigen on red blood cells – and so has blood group AB, which is a phenotype distinct from either A or B alone.
- **Example: roan coat in cattle.** A red bull (RR, red hairs only) crossed with a white cow (WW, white hairs only) gives F_1 calves with a roan coat – a mixture of red *and* white hairs visible together on the same animal.

Step 2. **(b) Incomplete dominance.**

- **Definition.** When the heterozygote shows an *intermediate* phenotype – a blend between the two homozygous parental phenotypes – neither allele is fully dominant; this is **incomplete dominance**.
- **Example: snapdragon flower colour (*Antirrhinum majus*).** A red-flowered homozygote (RR) crossed with a white-flowered homozygote (rr) gives F_1 heterozygotes (Rr) with *pink* flowers – an intermediate colour. On selfing, the F_2 ratio is 1 Red $(RR) : 2$ Pink $(Rr) : 1$ White (rr), i.e. the phenotypic ratio matches the genotypic ratio (because each genotype now has its own distinguishable phenotype).
- **Mechanism.** A single dose of the R allele makes only enough red pigment to produce pink, not fully red, so the heterozygote is intermediate.



Final Answer: Co-dominance: both alleles express simultaneously and distinctly in the heterozygote (ABO blood-group $I^AI^B \rightarrow$ AB; roan cattle). Incomplete dominance: heterozygote shows an *intermediate* phenotype (snapdragon $Rr \rightarrow$ pink; F_2 ratio 1 red : 2 pink : 1 white).

✗ Common Mistake

Don't swap the two definitions: in *co-dominance* the heterozygote shows BOTH parental phenotypes side by side (AB blood group, roan coat), whereas in *incomplete dominance* it

shows a BLENDED intermediate (pink snapdragon). Also note: the F_2 ratio for BOTH is 1:2:1 (each genotype is now distinguishable), not Mendel's 3:1.

EXPERT'S SOLUTION *: Tara Bhat, M.Sc Botany, Delhi University*

Structural angle. Co-dominance and incomplete dominance both break the heterozygote-equals-dominant rule, but they do it in different ways. Hold one image in your head for each:

Step 1. **Image for co-dominance – roan cattle.** A red bull $\times$ white cow gives a roan calf: *red hairs and white hairs both visible, mixed on the same coat*. Neither allele dominates the other; both express in their own cells. Similarly, an $I^A I^B$ person has *both* antigens on red cells – that's AB blood group.

Step 2. **Image for incomplete dominance – snapdragon.** A red flower $\times$ white flower gives a pink F_1. The heterozygote is *intermediate*, not a mosaic. One dose of the red allele makes half as much pigment, so the flower looks pink, not red.

Step 3. **Tell them apart in a cross.**

- Co-dominance: F_1 shows BOTH parental phenotypes simultaneously (e.g. blood group AB).
- Incomplete dominance: F_1 shows a BLENDED phenotype between the two parents (e.g. pink).

Step 4. **F_2 ratios.** For BOTH co-dominance and incomplete dominance, the F_2 phenotypic ratio is $1:2:1$ (same as the genotypic ratio), because every genotype now has its own distinguishable phenotype. Contrast this with the Mendelian $3:1$ where heterozygotes look like dominant homozygotes.

Step 5. **Biochemical reason for each.** In incomplete dominance, the dominant allele's protein is a rate-limiting enzyme; one functional copy makes only half the product, so the phenotype is intermediate (half-as-much red pigment = pink). In co-dominance, the two alleles encode *different* products that are BOTH made and BOTH visible (different antigens on the same RBC, or different pigment in different hair cells). The distinction sits in molecular biology, not in Mendelian counting.

Step 6. **Spotting them on a Punnett square.** Mendelian dominance: F_2 shows three phenotype classes with ratio 3:1. Incomplete dominance: F_2 shows three classes with ratio 1:2:1 – intermediate visible. Co-dominance: F_2 shows three classes with ratio 1:2:1 – both parental phenotypes plus their hybrid visible. The 1:2:1 phenotype ratio is the give-away that one of the two non-Mendelian rules is at work.

Why this matters. Both phenomena show that "dominance" is a feature of how alleles' *products* interact in a cell, not a fundamental property of the gene itself. Whether a

heterozygote looks dominant, blended or co-dominant depends on the biochemistry downstream of the gene.

Final Answer: Co-dominance: heterozygote expresses BOTH alleles distinctly (ABO blood group $I^A I^B \rightarrow$ AB; roan cattle). Incomplete dominance: heterozygote shows an intermediate phenotype (snapdragon $Rr \rightarrow$ pink; F_2 ratio $1:2:1$).

Q 4.14 What is point mutation? Give one example.

SOLUTION

Concept used. A **mutation** is any heritable change in the DNA sequence of an organism. Mutations come in many flavours – chromosomal (large rearrangements), insertions/deletions, etc. A **point mutation** is the smallest kind: a change in a *single base pair* of the DNA, which usually translates into the substitution of *one nucleotide* for another (and hence potentially one amino acid for another in the protein).

Step 1. **Definition.** A point mutation is the alteration of a single nucleotide base in DNA. There are three kinds:

- **Substitution** – one base replaces another (A $\rightarrow$ G, T $\rightarrow$ C, etc.). When the new codon still codes for the same amino acid, the substitution is **silent**; when it codes for a different amino acid the substitution is **missense**; when it creates a stop codon the substitution is **nonsense**.
- **Insertion** – one extra base is added.
- **Deletion** – one base is lost.

Insertions and deletions shift the reading frame downstream – a **frameshift** mutation – usually with severe consequences for the protein.

Step 2. **Example: Sickle-cell anaemia.** A single point substitution in the gene for the β-globin chain of haemoglobin: the codon GAG (which codes for glutamic acid) becomes GTG (which codes for valine) at the sixth codon of the chain.

$$\text{GAG (Glu)} \longrightarrow \text{GTG (Val)}$$

The mutant haemoglobin (HbS) molecules polymerise into long fibres when oxygen tension is low, deforming the red blood cell from a round biconcave disc into a rigid sickle shape. This sickling causes the symptoms of sickle-cell disease: anaemia, vaso-occlusive pain, organ damage.

Step 3. **Why this is "one base, big effect".** Replacing one amino acid (out of $\sim$146 in β-globin) is enough because the substituted residue sits on the surface of the

protein and creates a sticky hydrophobic patch that promotes haemoglobin polymerisation. One nucleotide → one amino acid → a multi-organ disease.

Normal (HbA): G A G → Glu (Glu, OK)

Sickle (HbS): G T G → Val (sickling)

Final Answer: A point mutation is a change in a single nucleotide base in DNA (substitution, insertion or deletion of one base). Example: sickle-cell anaemia – the substitution GAG → GTG at codon 6 of the β-globin gene replaces glutamic acid with valine, making haemoglobin polymerise and red cells sickle.

☞ Exam Tip

For full marks, your definition must mention "single base pair" or "single nucleotide" – a generic "small change in DNA" loses the point. For the example, give the codon-level detail (GAG → GTG, position 6, β-globin) rather than just naming the disease.

EXPERT'S SOLUTION *: Meera Desai, Ph.D Molecular Biology, NCBS Bangalore*

Quick reading. Two halves: define "point mutation" cleanly, then give one fully worked example.

Step 1. **Definition.** Point mutation = a change affecting a single base pair in DNA. The three flavours are substitution (most common), insertion of one base, and deletion of one base. Substitutions are further classified as silent, missense or nonsense, depending on the effect on the protein.

Step 2. **Pick sickle-cell anaemia as the example.** It's the classic NCERT example and is genuinely a point mutation (single base substitution).

Step 3. **State the molecular detail.** On the β-globin gene of chromosome 11, codon 6 changes from GAG (Glu) to GTG (Val). On the protein, the sixth amino acid of the β chain changes from glutamic acid to valine.

Step 4. **Trace the phenotypic consequence.** The valine creates a hydrophobic patch on the surface of haemoglobin. Under low oxygen, deoxygenated HbS polymerises into fibres that bend the red cell into a sickle shape; the rigid cells block capillaries, hemolyse, and cause vaso-occlusive crises and chronic anaemia.

Step 5. **Genotype to phenotype.** $HbA\,HbA$ = normal; $HbS\,HbS$ = sickle-cell anaemia (severe); $HbA\,HbS$ = sickle-cell trait (carriers, mostly asymptomatic; also resistant to falciparum malaria).

Why this matters. Sickle-cell was the first molecular disease – the first time a single-amino-acid change was traced from DNA all the way to clinical pathology. It set

the template for "one gene, one mutation, one disease" thinking.

> **Final Answer:** Point mutation = single-base-pair change in DNA. Example: sickle-cell anaemia, GAG → GTG (Glu → Val) at codon 6 of β-globin, causing HbS polymerisation and red-cell sickling.

Q 4.15 Who had proposed the chromosomal theory of inheritance?

SOLUTION

Concept used. By the early 1900s, two strands of biology needed to be unified: Mendel's abstract "factors" of inheritance (rediscovered in 1900) and the visible behaviour of chromosomes during meiosis (described in detail by cytologists in the 1880s–1890s). The **chromosomal theory of inheritance** proposed that Mendel's factors are physically carried on the chromosomes – and so the behaviour of chromosomes during meiosis *is* the cellular basis of Mendel's laws.

Step 1. **The proposal (1902–1903).** The chromosomal theory of inheritance was independently proposed in 1902 by **Walter Sutton** (American, working on grasshopper chromosomes) and in 1902–1903 by **Theodor Boveri** (German, working on sea-urchin embryos and the consequences of chromosomal imbalance).

Step 2. **What the theory states.**

- Mendelian factors (genes) are located on chromosomes.
- Chromosomes occur in pairs in diploid cells, just as Mendel's factors do.
- During meiosis homologous chromosomes pair and segregate to opposite poles – the cellular basis of Mendel's *law of segregation*.
- Different pairs of homologous chromosomes assort independently of each other on the meiotic plate – the cellular basis of Mendel's *law of independent assortment*.

Step 3. **Experimental confirmation by Morgan.** Sutton and Boveri's proposal was a strong inference but not yet experimentally proven. **Thomas Hunt Morgan**, working on *Drosophila* (1910 onwards), provided the experimental confirmation by demonstrating sex-linked inheritance of the white-eye mutation (showing the gene was on the X chromosome) and by discovering linkage and recombination – which showed that genes are physically located on chromosomes and inherited together when close.

> **Final Answer:** The **chromosomal theory of inheritance** was proposed by **Walter Sutton** and **Theodor Boveri** in 1902, and experimentally confirmed and extended by **Thomas Hunt Morgan** using *Drosophila*.

☞ Exam Tip

Single-mark questions like this reward exactness. Write BOTH names ("Sutton and Boveri"), the year (1902), and credit Morgan for the experimental confirmation. Naming just one of the two proposers is the single most common slip.

EXPERT'S SOLUTION *: Ananya Sharma, M.Sc Botany, Delhi University*

Strategic angle. A one-name answer is half a mark short. Two names + the year + Morgan's confirmation gives full marks.

Step 1. **Proposers.** Walter Sutton and Theodor Boveri, independently, in 1902.

Step 2. **Insight.** Mendel's "factors" must reside on chromosomes because chromosomes behave during meiosis exactly like Mendel's factors behave during gamete formation: paired in diploids, segregating to gametes, and assorting independently across pairs.

Step 3. **Experimental backing.** Morgan's *Drosophila* work (linkage, recombination, sex linkage) supplied the experimental confirmation a decade later.

Step 4. **Why both names matter.** Sutton emphasised the parallel between meiosis and Mendelian segregation; Boveri emphasised the necessity of a complete chromosome set for normal development (sea-urchin work). Together they made the case airtight; the proposal is fairly called the **Sutton–Boveri chromosomal theory of inheritance**.

Why this matters. This theory bridged the gap between classical genetics (counting offspring ratios) and cytology (looking at chromosomes under a microscope). Once chromosomes carried genes, genetics had a physical substrate – and the road to DNA, half a century later, was open.

> **Final Answer:** Walter Sutton and Theodor Boveri (1902); experimentally confirmed by T.H. Morgan via his *Drosophila* studies on linkage and sex-linked inheritance.

Q 4.16 Mention any two autosomal genetic disorders with their symptoms.

SOLUTION

Concept used. An **autosomal genetic disorder** is one whose causative gene sits on one of the 22 autosomes (not on the X or Y chromosome). Autosomal disorders affect males and females equally. They are further classified as *autosomal recessive* (require both copies mutant, aa) or *autosomal dominant* (a single mutant copy, Aa, is enough).
We pick the two NCERT examples discussed in the chapter: **Sickle-cell anaemia** (autosomal recessive) and **Phenylketonuria, PKU** (autosomal recessive). Down's syndrome is excluded here because it is chromosomal (trisomy 21), not a single-gene disorder.

Step 1. **(i) Sickle-cell anaemia.**

- **Inheritance:** autosomal recessive. Causative gene on chromosome 11 (β-globin gene). Affected genotype $HbS\,HbS$; carriers ($HbA\,HbS$) have sickle-cell trait, are usually asymptomatic and are resistant to falciparum malaria.
- **Molecular cause:** point mutation GAG $\rightarrow$ GTG at codon 6 of β-globin, changing glutamic acid to valine. Mutant haemoglobin (HbS) polymerises under low oxygen.
- **Symptoms:**
  - Chronic haemolytic anaemia (fatigue, pallor, breathlessness on exertion).
  - Sickling of red blood cells into rigid crescent shapes, especially when oxygen tension is low.
  - Vaso-occlusive crises: blockage of small blood vessels causing severe pain in the chest, abdomen, joints and bones.
  - Spleen damage, recurrent infections, delayed growth in children, stroke risk and organ damage (kidneys, lungs).

Step 2. **(ii) Phenylketonuria (PKU).**

- **Inheritance:** autosomal recessive. Causative gene on chromosome 12 (gene for the enzyme phenylalanine hydroxylase).
- **Molecular cause:** mutation in the phenylalanine hydroxylase gene; the enzyme is absent or inactive, so dietary phenylalanine cannot be converted to tyrosine. Phenylalanine accumulates in body fluids and is converted to phenylpyruvic acid and other toxic derivatives.
- **Symptoms:**
  - Severe, progressive intellectual disability (mental retardation) developing in untreated infants.
  - Accumulation of phenylalanine and its derivatives in the blood, causing brain damage.
  - Excretion of phenylpyruvic acid and other phenyl derivatives in urine

(gives a characteristic musty odour).
- Reduced melanin pigmentation: paler skin, lighter hair, blue eyes.
- Seizures and behavioural problems if untreated.

- **Management:** early diagnosis (neonatal screening) followed by a strict low-phenylalanine diet prevents most symptoms – a striking case where a genetic disease is managed by diet.

Final Answer: Two autosomal genetic disorders: (1) **Sickle-cell anaemia** (autosomal recessive; single base substitution Glu $\rightarrow$ Val at codon 6 of β-globin): chronic haemolytic anaemia, sickling of RBCs, vaso-occlusive pain crises, splenic dysfunction. (2) **Phenylketonuria** (autosomal recessive; deficiency of phenylalanine hydroxylase): build-up of phenylalanine, severe intellectual disability, light pigmentation, musty-smelling urine.

✗ Common Mistake

Don't list Down's syndrome, Klinefelter's syndrome or Turner's syndrome here. Those are chromosomal disorders (whole-chromosome imbalance), not autosomal single-gene disorders. Likewise, haemophilia and colour blindness are X-linked, not autosomal.

EXPERT'S SOLUTION *: Ishaan Gupta, M.Sc Biotechnology, AIIMS Delhi*

Strategic angle. Two clean autosomal examples, each with a crisp molecular cause and a clinical phenotype paragraph. Don't pad with extras – quality of two beats a poorly-described five.

Step 1. **Example 1 – Sickle-cell anaemia.**
- Inheritance: autosomal recessive (chromosome 11).
- Molecular lesion: GAG $\rightarrow$ GTG point mutation at codon 6 of β-globin; Glu $\rightarrow$ Val.
- Pathophysiology: HbS polymerises under low O_2; red cells deform into rigid sickles; cells lyse and block capillaries.
- Symptoms: chronic anaemia, vaso-occlusive pain crises, infections, organ damage, stunted growth.

Step 2. **Example 2 – Phenylketonuria.**
- Inheritance: autosomal recessive (chromosome 12).
- Molecular lesion: deficient/absent phenylalanine hydroxylase; phenylalanine cannot be converted to tyrosine.
- Pathophysiology: phenylalanine and its toxic derivatives (phenylpyruvate)

accumulate in blood and brain.

- Symptoms: progressive intellectual disability in untreated infants, reduced pigmentation, musty urine odour, seizures.
- Management: low-phenylalanine diet from birth prevents symptoms – a rare example of a managed inborn error of metabolism.

Why this matters. Both examples illustrate how a single gene defect on an autosome produces a definite disease pattern that is independent of the patient's sex, and both have well-defined molecular mechanisms – a far cry from the symptomatic guesswork of pre-genetic medicine.

> **Final Answer:** Sickle-cell anaemia (autosomal recessive: HbS polymerisation, sickled red cells, anaemia, pain crises); phenylketonuria (autosomal recessive: phenylalanine hydroxylase deficiency, intellectual disability, musty urine, light pigmentation, treatable by diet).

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Key Takeaways

- Mendel's three laws – Dominance, Segregation, Independent Assortment – were inferred from sharply contrasting traits in pea plants and quantitative counts on large F_2 populations. The signature ratios are $3:1$ (monohybrid) and $9:3:3:1$ (dihybrid).
- Punnett squares are tabular tools for combining gametes; for n heterozygous independently-assorting loci, an individual produces 2^n gamete types.
- Test crosses (unknown $\times$ homozygous recessive) distinguish homozygous from heterozygous dominant individuals: all-dominant offspring $\Rightarrow TT$; 1:1 split $\Rightarrow Tt$.
- Mendel's laws have exceptions: **incomplete dominance** (snapdragon Rr pink, F_2 ratio $1:2:1$), **co-dominance** (ABO blood-group I^AI^B = AB; roan cattle), **multiple alleles** (three ABO alleles), **pleiotropy**, and **linkage** (Morgan's flies).
- The Sutton–Boveri chromosomal theory (1902) localised Mendelian factors on chromosomes; Morgan's *Drosophila* work confirmed it and added the concepts of linkage, recombination and genetic mapping.

- In humans, sex is determined by the XX/XY system; the father's sperm (X-bearing $\rightarrow$ daughter, Y-bearing $\rightarrow$ son) determines the sex of the child.
- Pedigree analysis is the human-genetics workaround for the impossibility of controlled human crosses; it traces the mode of inheritance from family-tree symbols and is the foundation of genetic counselling.
- Point mutations are single-base-pair changes in DNA (substitutions, insertions, deletions); sickle-cell anaemia (GAG $\rightarrow$ GTG at codon 6 of β-globin) is the textbook example.
- Genetic disorders are either chromosomal (Down's, Klinefelter's, Turner's – whole-chromosome imbalances) or Mendelian/single-gene (autosomal: sickle-cell, PKU, thalassemia; sex-linked recessive: haemophilia, colour blindness, Duchenne muscular dystrophy).

End of Exercises