

ESAT

Biology

Inheritance

Free lesson notes

Covers	B4.1, B4.2 and B4.3
Format	Lesson, worked examples, self-check
Price	Free

About this lesson

Why do you look like your parents, but not exactly like either? Why can two parents with brown eyes have a child with blue eyes? This lesson explains how characteristics are passed on, from the genetic material in the nucleus (B4.1), through the precise vocabulary of genetics (gene, allele, dominant, recessive, homozygous, heterozygous, genotype, phenotype, autosome; B4.2), to **monohybrid crosses** and **family pedigrees** (B4.3). Genetic questions in ESAT are mostly careful bookkeeping and probability, so we practise a fixed method.

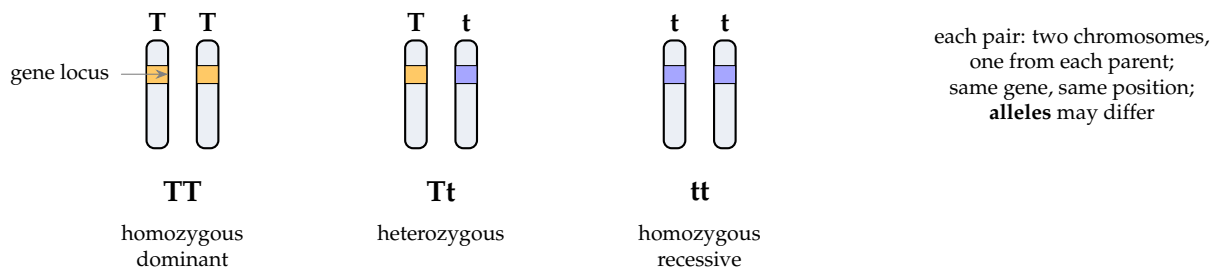
Cover each worked example and try it first. There is no calculator in the real ESAT, so the numbers are friendly. Finish with the self-check, then the practice set.

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1 Genes and alleles (B4.1, B4.2)

The **genetic material** of a cell is DNA, and in a eukaryotic cell nearly all of it is held in the **nucleus**, packaged into chromosomes (the small amount of extra DNA in mitochondria and chloroplasts aside). A **gene** is a short section of DNA that codes for a particular protein; it is the unit of inheritance. The proteins (for example enzymes) produced by genes determine the characteristics of the organism, such as flower colour or blood type.

Chromosomes come in pairs (section 2 of the cell division lesson), one from each parent, and the two chromosomes of a pair carry the same genes in the same places. But a gene can come in different versions. The different versions of a gene are called **alleles**. For example, the gene for height in pea plants has an allele for tall and an allele for dwarf. An individual has two alleles of each gene (one on each chromosome of the pair) and they can be the same or different.



In the picture the gene locus (the position of the gene) is shown as a band on each chromosome. We write alleles using a letter: a **capital** letter for the dominant allele (T for tall) and the **same letter in lower case** for the recessive allele (t for dwarf). A gamete contains only *one* allele of each gene, because it has one chromosome of each pair.

Key ideas

- DNA (the genetic material) is in the nucleus, in chromosomes. A **gene** is a section of DNA coding for a protein.
- Alleles are the different versions of one gene. A body cell has two alleles of each gene; a gamete has one.

Common mistakes

- Saying “a gene is a chromosome” or that an allele is a different gene. An allele is a version of the *same* gene.
- Saying a gamete carries two alleles of a gene. It carries one.
- Writing the two alleles with different letters (T and D). Use one letter for a gene: T and t.

2 The vocabulary of genetics (B4.2)

Every word below is worth learning exactly, as ESAT questions test them directly.

term	meaning (example: height in pea plants, T = tall, t = dwarf)
gene	a section of DNA that codes for a protein (the gene for height)
allele	a version of a gene (T or t)
dominant allele	the allele that shows in the phenotype even when only one copy is present (T)
recessive allele	shows in the phenotype only when two copies are present (t)
homozygous	the two alleles of the gene are the <i>same</i> : TT (homozygous dominant) or tt (homozygous recessive)
heterozygous	the two alleles are <i>different</i> : Tt
genotype	the combination of alleles an individual has (TT, Tt or tt)
phenotype	the observable characteristic that results (tall or dwarf)
autosome	any chromosome that is not a sex chromosome; humans have 22 pairs of autosomes

From genotype to phenotype. Because T is dominant, both TT and Tt plants are tall: only tt is dwarf. So two different genotypes can give the same phenotype, but each genotype gives only one phenotype (in the cases in this specification, where one allele is completely dominant). A tall plant could therefore be TT or Tt; you cannot tell by looking.

Worked example: naming

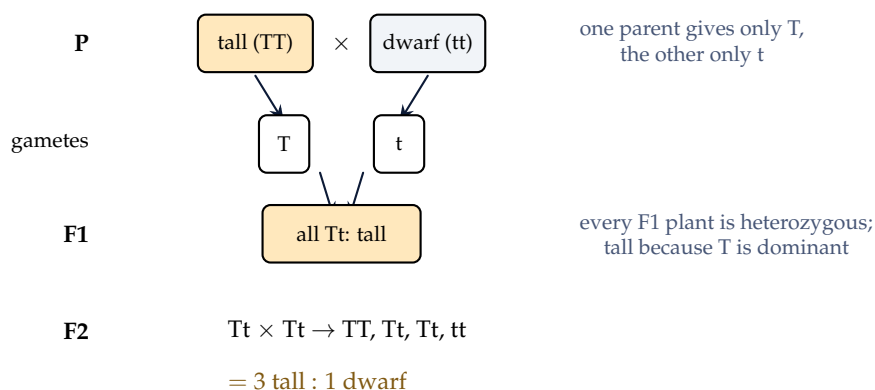
In rabbits, black fur (B) is dominant to brown fur (b). State the phenotype, and whether each is homozygous or heterozygous: (a) BB, (b) Bb, (c) bb.
(a) black, homozygous (dominant). (b) black, heterozygous. (c) brown, homozygous (recessive). A brown rabbit must be bb; a black rabbit is BB or Bb.

Common mistakes

- Mixing up genotype (the alleles, e.g. Bb) and phenotype (what you see, e.g. black).
- Thinking “dominant” means “more common” or “stronger”. It only describes which allele shows when the two alleles differ.
- Saying a recessive allele “skips” or is “lost”. It is still there in heterozygotes; it can be passed on.

3 Monohybrid crosses (B4.3)

A **monohybrid cross** follows the inheritance of *one* gene (a single pair of contrasting characteristics). Here is the classic example. A pure-breeding tall plant (TT) is crossed with a pure-breeding dwarf plant (tt). Each parent makes gametes with only one kind of allele, so all the offspring (the first generation, F1) are Tt: tall, because T is dominant. If the F1 plants are crossed together (or self-pollinated), the F2 generation shows 3 tall : 1 dwarf: the recessive dwarf characteristic reappears.



The F2 result comes from a **genetic diagram** (Punnett square). The method is the same every time:

- Write the genotypes of the two parents.

- Write the gametes each can make (one allele from each parent's pair).
- Draw a 2×2 grid with one parent's gametes along the top and the other's down the side.
- Fill in each box with the two alleles that meet at fertilisation.
- Read off the offspring genotypes, then phenotypes, and the ratio.

		parent 2 gametes		
		T	t	
parent 1 gametes	T	TT	Tt	
	t	Tt	tt	

Tt × Tt
 genotypes: 1 TT : 2 Tt : 1 tt
 phenotypes: 3 tall : 1 dwarf
 probability of dwarf = $\frac{1}{4}$

The four boxes are equally likely (each gamete type is equally likely, and fertilisation is random). So the genotype ratio is 1 TT : 2 Tt : 1 tt and, since TT and Tt are both tall, the phenotype ratio is 3 tall : 1 dwarf: a probability of $\frac{3}{4}$ of being tall and $\frac{1}{4}$ of being dwarf.

Worked example: a monohybrid cross

In guinea pigs, rough coat (R) is dominant to smooth coat (r). Two heterozygous rough-coated guinea pigs are crossed. What proportion of the offspring are expected to be smooth?

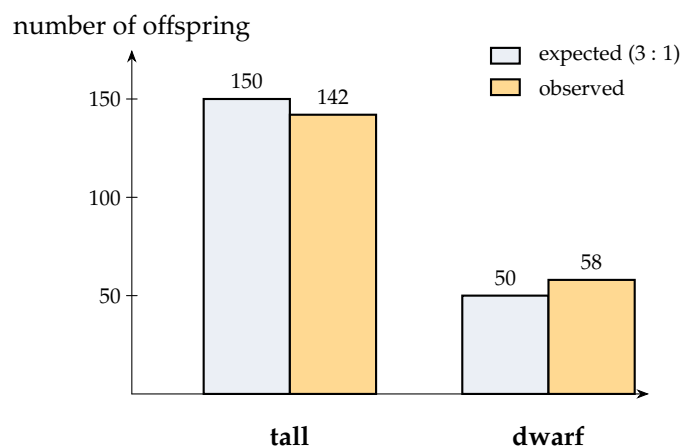
Parents Rr × Rr; gametes R, r and R, r; offspring RR, Rr, Rr, rr. Only rr is smooth, which is $\frac{1}{4}$ of the offspring.

Common mistakes

- Putting two alleles from the same parent along the top. Each box needs one allele from *each* parent.
- Reading the ratio from the *genotype* instead of the phenotype: 1 : 2 : 1 is the genotype ratio; 3 : 1 is the phenotype ratio.
- Writing a probability as a ratio (3 : 1) or the other way round. $\frac{3}{4}$ tall means a 3 : 1 ratio of tall to dwarf.
- Forgetting that a homozygous parent produces only one type of gamete (TT gives only T).

4 Probabilities, expected numbers and the test cross (B4.3)

A Punnett square gives *probabilities*, and expected numbers follow from them. If 240 offspring are produced from Tt × Tt, we expect $\frac{3}{4} \times 240 = 180$ tall and $\frac{1}{4} \times 240 = 60$ dwarf. Real results vary by chance, particularly with small numbers: ten offspring might be 9 tall : 1 dwarf, and that does not disprove 3 : 1. With large numbers the results are close to the expected ratio.



In the graph, 142 tall : 58 dwarf is close to the expected 150 : 50, so the data are consistent with Tt × Tt.

Combining probabilities. Each child is a separate fertilisation, so the events are independent and probabilities multiply: if the chance of a dwarf is $\frac{1}{4}$, the chance that two children in a row are both dwarf is $\frac{1}{4} \times \frac{1}{4} = \frac{1}{16}$. Chance of "at least one" is easiest as $1 - P(\text{none})$.

The test cross. A tall plant could be TT or Tt. To find out, cross it with a dwarf plant (tt, whose gametes all carry t). If the tall plant is TT, all offspring are Tt (tall). If it is Tt, about half the offspring will be dwarf.

	T	t			T	T	
t	Tt	tt	Tt × tt 1 tall : 1 dwarf	t	Tt	Tt	TT × tt all tall (all Tt)
t	Tt	tt		t	Tt	Tt	

Worked example: using a test cross

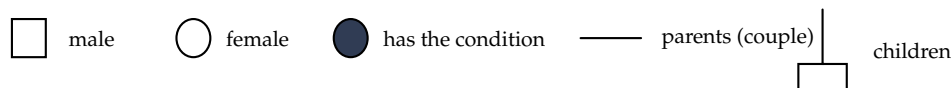
A tall pea plant of unknown genotype is crossed with a dwarf plant. The offspring are 48 tall and 52 dwarf. What was the genotype of the tall plant?
Any dwarf offspring (tt) must have received a t from the tall parent, so it was heterozygous, Tt. The 1 : 1 ratio is the expected result for $Tt \times tt$.

Common mistakes

- Expecting results to match the ratio exactly in a small sample.
- Adding the probabilities of independent events instead of multiplying them.
- Thinking a test cross works with a heterozygous partner: it must be with a homozygous *recessive*.

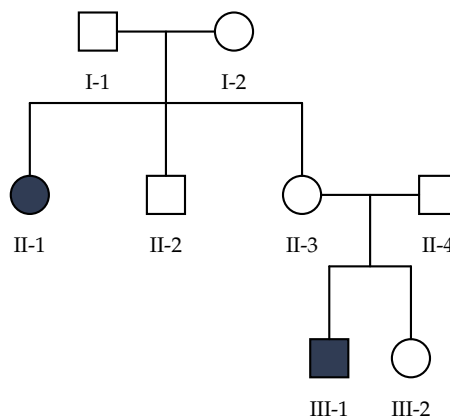
5 Pedigree diagrams (B4.3)

A **pedigree** (family tree) shows how a characteristic or disorder passes through the generations of a family. The symbols:



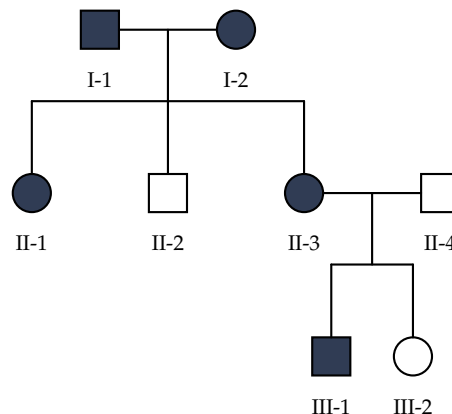
Generations are numbered I, II, III, and individuals within a generation 1, 2, 3..., so "II-3" is the third person of the second generation. We read the pedigree to work out whether the allele is dominant or recessive, and the genotypes of the people.

Is the allele recessive? A pedigree shows a recessive condition if *two unaffected parents have an affected child*. Both parents must carry the recessive allele without showing it (Rr), and the child inherited a recessive allele from each (rr).



Here I-1 and I-2 are unaffected but their daughter II-1 is affected, so the condition is recessive. We can then name genotypes: II-1 is rr; her parents must each be Rr (each gave her an r). II-3 is unaffected but had an affected son, so she is Rr, and so is her partner II-4.

Is the allele dominant? The condition is dominant if *two affected parents have an unaffected child*: each parent is Dd, and the child inherited d from both (dd).



Here I-1 and I-2 are both affected, but II-2 is unaffected, so the condition must be dominant; the unaffected people are dd, and I-1 and I-2 are both Dd.

Worked example: reading a pedigree

In the first (recessive) pedigree, which people must be heterozygous (carriers)?

An affected person is rr. Anyone with an rr child must carry at least one r; being unaffected, they must be Rr. So I-1 and I-2 (parents of II-1), and II-3 and II-4 (parents of III-1). II-2 could be RR or Rr: we cannot tell from the diagram.

Exam tip

First decide dominant or recessive using the “two unaffected parents, affected child” rule (recessive) or “two affected parents, unaffected child” rule (dominant). Then write letters in the diagram, starting with the people whose genotype is certain: all those showing the recessive phenotype are homozygous recessive.

Common mistakes

- Deciding a condition is dominant just because every generation has affected people. Use the two-parents rule.
- Assuming that an unaffected person in a recessive pedigree is RR. They could be a carrier (Rr).
- Mixing up the symbols: a square is male, a circle is female.

6 Inherited disorders, carriers and probability (B4.3)

Some inherited disorders are caused by alleles of a single gene. **Cystic fibrosis** is caused by a *recessive* allele: sufferers have two copies (rr) and have thick sticky mucus in the lungs and gut. **Huntington’s disease** and **polydactyly** (extra fingers or toes) are caused by *dominant* alleles: a single copy is enough to show the condition.

A **carrier** is a person who is heterozygous for a recessive disorder allele: they do not have the disorder, but can pass the allele on. Two carriers have a one in four chance of an affected child with each pregnancy:

	R	r	
R	RR	Rr	Rr × Rr (both carriers) RR: unaffected Rr, Rr: unaffected carriers rr: affected $P(\text{affected}) = \frac{1}{4}, P(\text{carrier}) = \frac{1}{2}$
r	Rr	rr	

A common trap: unaffected child of two carriers. What is the probability that an *unaffected* child of two carriers is itself a carrier? The unaffected outcomes are RR, Rr and Rr: three of the four boxes. Two of these three are carriers, so the probability is $\frac{2}{3}$ (not $\frac{1}{2}$). Whenever you are told the child is unaffected, remove the affected box from the grid before counting.

Worked example: carrier probability

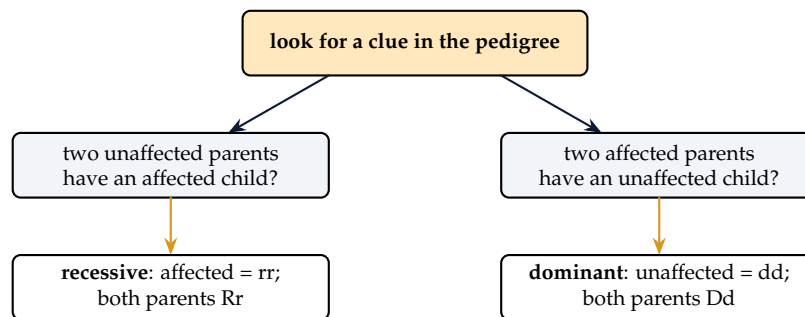
Two unaffected parents have a child with cystic fibrosis. What is the probability that their next child is (a) affected, (b) an affected boy, (c) unaffected and a carrier? The parents must both be Rr. (a) $\frac{1}{4}$. (b) $\frac{1}{4} \times \frac{1}{2} = \frac{1}{8}$ (the sex of the child is independent of the cystic fibrosis genotype). (c) Rr is $\frac{2}{4} = \frac{1}{2}$.

Common mistakes

- Giving $\frac{1}{2}$ as the probability that an unaffected child of two carriers is a carrier. It is $\frac{2}{3}$.
- Believing carriers have a mild form of the disorder: for a recessive disorder, carriers are unaffected.
- Saying a person with a dominant disorder must have two affected parents: they could have one affected parent, with genotype Dd, and one unaffected (dd).
- Forgetting that genetic probabilities for each child are independent of the previous children.

7 Three full walkthroughs (B4.3)

Here is the pedigree decision in one picture, then three complete problems that combine the ideas in this lesson.



Worked example: from data to genotypes

Brown eyes (B) are dominant to blue eyes (b). A brown-eyed father and a brown-eyed mother have a blue-eyed child. (a) State all the genotypes you can be sure of. (b) What is the probability that their next child has blue eyes? (c) That it has brown eyes and is a carrier of blue? (a) The child is bb, so each parent passed on b. As both parents have brown eyes (a dominant phenotype) both must be Bb. (b) $Bb \times Bb: P(bb) = \frac{1}{4}$. (c) Brown and a carrier means Bb: $\frac{2}{4} = \frac{1}{2}$.

Worked example: expected numbers and a comparison

A gardener crosses two heterozygous red-flowered plants (R = red, dominant to r = white) and grows 80 seeds. How many of each colour would you expect? She actually gets 56 red and 24 white. Is that consistent with the prediction? Expected: $\frac{3}{4} \times 80 = 60$ red and $\frac{1}{4} \times 80 = 20$ white. The observed 56 : 24 is close to 60 : 20 (a difference of only 4 plants each way), which is the sort of variation chance produces in a sample this size, so the result is consistent with a cross of two heterozygotes.

Worked example: a pedigree with probabilities

Two carriers of a recessive disorder have a child. (a) What is the probability that the child is a carrier? (b) The child is unaffected: now what is the probability that it is a carrier? (c) That child later has a child with a carrier of the same disorder, who is also unaffected. What is the probability that their first child is affected? (a) $\frac{1}{2}$. (b) Given unaffected, $\frac{2}{3}$. (c) The child is Rr with probability $\frac{2}{3}$ (and RR otherwise, in which case no child can be affected); for an affected child both need to pass r. $P = \frac{2}{3} \times \frac{1}{4} = \frac{1}{6}$.

Common mistakes

- Not reading the question for what is *given*: conditional information ("the child is unaffected") changes the probability.

- Writing a genotype for a person who shows the dominant phenotype without checking the evidence: it may be homozygous or heterozygous.
- Rushing the arithmetic of probabilities: write each fraction, then multiply.

8 Putting it together: exam technique

1. Define the letters first: "Let T be the dominant allele (tall) and t the recessive allele (dwarf)".
2. Work out the parents' genotypes. Recessive phenotype $\Rightarrow$ homozygous recessive. Dominant phenotype $\Rightarrow$ at least one dominant allele (use the offspring or the pedigree to decide).
3. Write the gametes (one allele each) and draw the Punnett square.
4. Read off the genotype ratio, then convert to the phenotype ratio and probability.
5. For several children, multiply independent probabilities. For "at least one", use $1 - P(\text{none})$.
6. For a pedigree, apply the two-parents rules, then label the genotypes of certain people first.

Spec coverage (B4.1 to B4.3). The nucleus as the home of the genetic material; the terms gene, allele, dominant, recessive, homozygous, heterozygous, genotype, phenotype and autosome; monohybrid crosses and pedigrees.

Check yourself

Try these without looking back, then compare with the answers on the next page.

1. Where in a cell is most of the genetic material found?
2. A pea plant has genotype Tt. State its genotype type (homozygous or heterozygous) and its phenotype.
3. What is the difference between an allele and a gene?
4. Cross $Rr \times rr$. What proportion of the offspring are rr?
5. From $Tt \times Tt$, how many dwarf plants do you expect among 400 offspring?
6. Two unaffected parents have an affected son. Is the condition dominant or recessive? Give the parents' genotypes.
7. Two carriers of cystic fibrosis have two children. What is the probability that both are affected?
8. Why is a test cross done with a homozygous recessive individual?

Answers to the self-check

1. In the nucleus, as chromosomes made of DNA.
2. Heterozygous; tall (T is dominant).
3. A gene is a section of DNA coding for a protein; alleles are different versions of the same gene.
4. Gametes R, r and r, r: offspring Rr, Rr, rr, rr, so $\frac{1}{2}$ are rr.
5. $\frac{1}{4} \times 400 = 100$.
6. Recessive; both parents are carriers (Rr).
7. $\frac{1}{4} \times \frac{1}{4} = \frac{1}{16}$.
8. Its gametes all carry the recessive allele, so the phenotypes of the offspring reveal which alleles the other parent contributed.

Next step

Heurisco's *ESAT Biology: Inheritance Practice Set* has twenty original exam-style questions on this lesson, with Punnett squares and pedigrees, in order of difficulty, with a full worked solution and a note on the trap in each.