From alleles to offspring

A genetic diagram follows parental genotypes, the alleles in gametes and their combinations at fertilisation. Predicted ratios describe probabilities, not guaranteed family sizes.

1. THE LANGUAGE OF INHERITANCE

A gene is a DNA sequence with a functional product; its locus is its chromosome position. Alleles are versions of a gene. Genotype is the allele combination; phenotype is the expressed characteristic, influenced by genotype and environment.
AA / aa: homozygous. Aa: heterozygous. With complete dominance, A determines the heterozygote phenotype; a is recessive. Dominant does not mean common or better.

2. MONOHYBRID CROSS: Aa × Aa

Gametes	A	a
A	AA	Aa
a	Aa	aa

Each parent produces A and a gametes, each with probability 1/2.

Genotypes: 1 AA : 2 Aa : 1 aa
Phenotypes: 3 dominant : 1 recessive

Starting with AA × aa gives all Aa in F_1 (first filial generation). Crossing F_1 individuals gives F_2, the second filial generation shown above.

3. TEST CROSS: REVEAL A HIDDEN RECESSIVE ALLELE

Cross the dominant-phenotype individual with aa.
AA × aa: A gametes + a gametes give all Aa.
Aa × aa: A or a gametes + a gametes give 1 Aa : 1 aa.
Recessive offspring reveal that the tested parent carries a. A small sample with only dominant offspring does not prove AA: an Aa parent could produce that sample by chance.

Definitions and required genetic diagrams: Cambridge 9700, section 16.2.
Monohybrid and test-cross principles: OpenStax, Biology 2e 12.2.

Scope: Cambridge 9700, 16.2.1-5 (epistasis in next lesson), p. 41.

Two genes: separate or linked?

A dihybrid cross follows two loci. First establish which gametes can form and their probabilities; do not assume every cross produces a 9 : 3 : 3 : 1 ratio.

1. INDEPENDENT ASSORTMENT: AaBb × AaBb

Assume unlinked genes, complete dominance at both loci, no epistasis and equal offspring survival. Each parent makes AB, Ab, aB and ab gametes, each with probability 1/4.

Gametes	AB	Ab	aB	ab
AB	AABB	AABb	AaBB	AaBb
Ab	AABb	AAbb	AaBb	Aabb
aB	AaBB	AaBb	aaBB	aaBb
ab	AaBb	Aabb	aaBb	aabb

Phenotypes: 9 A_B_ : 3 A_bb : 3 aaB_ : 1 aabb
An underscore means either allele can occupy that position. Each of the 16 equally likely boxes represents probability 1/16.

2. AUTOSOMAL LINKAGE CHANGES GAMETE FREQUENCIES

Genes on the same autosome are physically linked. In AB / ab, AB is on one homologue and ab on the other. Without crossing over between the loci, gametes are only AB and ab (1/2 each). Crossing over can produce Ab and aB recombinants; detectable linkage gives more parental than recombinant combinations.

Test-cross comparison with aabb: unlinked AaBb gives four classes in 1 : 1 : 1 : 1; complete linkage in AB / ab gives AaBb and aabb in 1 : 1.

Independent assortment and dihybrid ratios: OpenStax, Biology 2e 12.3.
Linked genes and recombination: OpenStax, Biology 2e 13.1.

Scope: Cambridge 9700, 16.2.1-5 (epistasis in next lesson), p. 41.

Codominance and sex linkage

Define allele symbols before drawing a cross. Superscripts distinguish alleles without implying that a codominant allele is recessive.

1. ABO: MULTIPLE ALLELES AND CODOMINANCE

The standard ABO model has three alleles in the population: I^A, I^B and i. An individual has two. I^A and I^B are codominant; both dominate i. An AB individual expresses both A and B antigens, not a blend.

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

Cross: $I^Ai \times I^Bi$

Offspring:
1/4 AB, 1/4 B,
1/4 A, 1/4 O.

2. AN X-LINKED RECESSIVE EXAMPLE

Let X^N carry a dominant allele and X^n a recessive allele. Assume the locus is absent from Y. Cross a heterozygous female (X^NX^n) with an unaffected male (X^NY).

Gametes	X^N	Y
X^N	X^NX^N	X^NY
X^n	X^NX^n	X^nY

All daughters unaffected;
half are carriers.

Half the sons affected:
1/4 of all offspring if
sexes are equally likely.

A male has one allele at this X-linked locus (hemizygous), so a recessive allele can be expressed alone. Fathers pass X to daughters, not sons.

ABO alleles and antigens: NCBI, The ABO blood group.
Codominance: NHGRI genetics glossary.
Sex-linked inheritance: OpenStax, Biology 2e 12.2.

Scope: Cambridge 9700, 16.2.1-5 (epistasis in next lesson), p. 41.

Do the results fit the prediction?

Observed offspring counts rarely match an expected ratio exactly. A chi-squared goodness-of-fit test assesses the discrepancy against a stated inheritance model.

1. STATE THE MODEL BEFORE CALCULATING

Null hypothesis (H_0): offspring frequencies fit the predicted 3 : 1 ratio; deviations reflect sampling variation. Use independent observations in mutually exclusive categories. Work with counts, not percentages; for this course-level application, each expected count should be at least 5.

2. WORKED EXAMPLE: 80 OFFSPRING

Observed: 62 dominant and 18 recessive.
Expected: 80 × 3/4 = 60 dominant; 80 × 1/4 = 20 recessive.

$$\chi^2 = \Sigma [(O - E)^2 / E]$$

Category	O	E	$(O - E)^2 / E$
Dominant	62	60	4 / 60 = 0.067
Recessive	18	20	4 / 20 = 0.200

Total chi-squared = 0.267
Degrees of freedom = categories - 1 = 2 - 1 = 1.

3. COMPARE AND CONCLUDE

At the 5% significance level and 1 degree of freedom, the critical value is 3.841. Since 0.267 is smaller, do not reject H_0: these data do not show a significant departure from 3 : 1. This does not prove the model is true. A value above the critical value would lead to rejecting H_0 and investigating why the model does not fit.

Goodness-of-fit procedure: NIST, Chi-Square Goodness-of-Fit Test.
Course requirements and probability table: Cambridge 9700, Mathematics.

Scope: Cambridge 9700, 16.2.1-5 (epistasis in next lesson), p. 41.