GENETICS · STUDY GUIDE

Exam 1 Study Guide

This guide covers the first two chapters. You will follow genes from parents to offspring, turn crosses into fractions, test whether real counts fit a ratio, and then see why many real ratios bend away from 3:1. Work one chapter at a time. Read what to know, copy the ratio table out by hand, then try each worked example with the steps covered. Finish each chapter with its ten self-test items. The cram sheet at the end holds every ratio and formula in one place.

WHAT THIS GUIDE COVERS

- Chapter 1 · Mendel's design, segregation, testcrosses, independent assortment, probability rules, chi square and pedigrees.
- Chapter 2 · incomplete dominance, codominance, multiple alleles, lethal alleles, epistasis, penetrance, expressivity and complementation.

CHAPTER 1

Mendelian Transmission and Probability

What to know

Before Mendel, most people thought traits blended like paint. Mendel showed they do not. He used true breeding pea lines, traits with two clear forms, separate P, F_1 and F_2 generations, reciprocal crosses and big counts. The counting is what let him see ratios.

A **gene** comes in forms called **alleles**. The **genotype** is the pair of alleles a plant carries. The **phenotype** is what you see. Two matching alleles make a **homozygote**. Two different alleles make a **heterozygote**. A dominant allele shows in the heterozygote. Dominance says nothing about how common or how useful an allele is.

The **principle of segregation** says the two alleles of a gene separate when gametes form, so each gamete gets one, with equal odds. Its physical basis is the separation of homologous chromosomes in anaphase I of meiosis. An *Aa* × *Aa* cross gives 1 *AA* : 2 *Aa* : 1 *aa*, which shows as 3 dominant : 1 recessive.

A **testcross** mates an unknown with a homozygous recessive. The tester gives only recessive alleles, so each offspring shows which allele the unknown sent. All dominant offspring point to *AA*. A 1:1 split proves *Aa*. A single recessive offspring is enough to prove the unknown is a heterozygote.

The **principle of independent assortment** says alleles of different, unlinked genes sort into gametes on their own. It comes from the random way homologous pairs line up at metaphase I. A dihybrid makes four gamete types in equal numbers. An intercross gives 9:3:3:1 and a dihybrid testcross gives 1:1:1:1. Genes close together on one chromosome break this rule, which is the subject of Chapter 4.

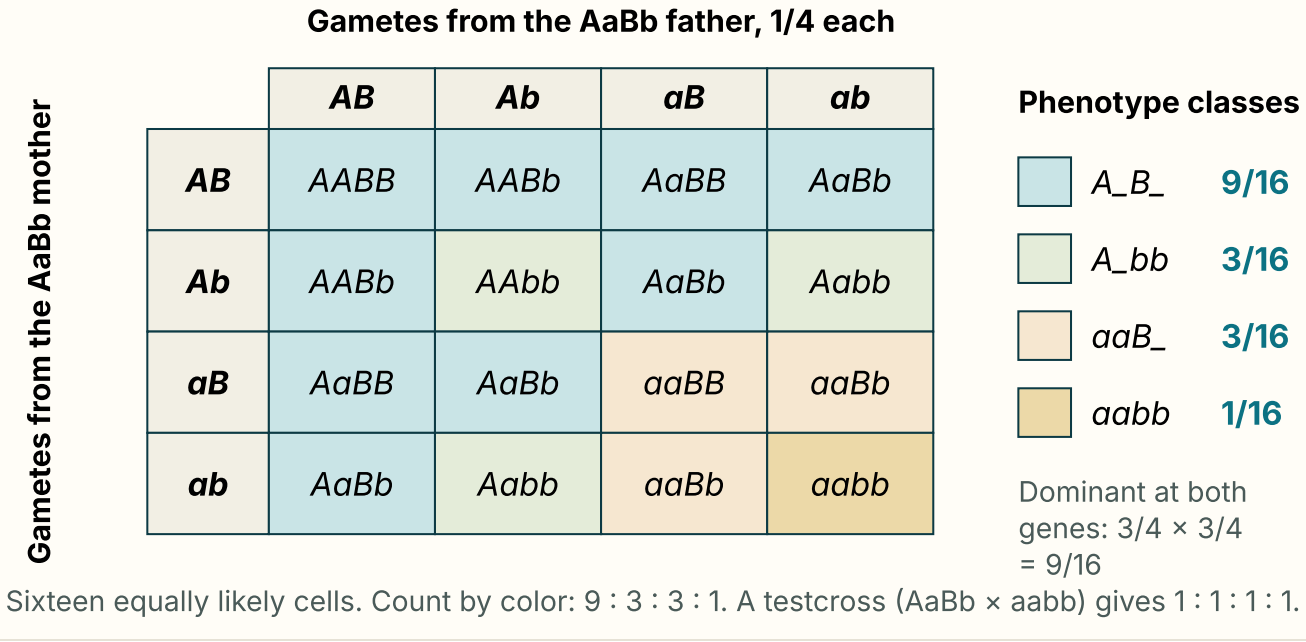
You rarely need a big Punnett square. Solve each gene alone, then multiply. Multiply for "and." Add for "or" when the outcomes cannot happen together. When you are told something, such as "this sibling is unaffected," cut away the outcomes that are now ruled out. That is **conditional probability**, and it gives the famous 2/3 carrier risk. Use the **binomial expansion** for "exactly k of n," and subtract from 1 for "at least one."

Real counts never match a ratio exactly. The **chi square test** asks whether the gap could be chance. Use raw counts, keep expected values unrounded, and use df = classes minus 1. If chi square beats the critical value at P = 0.05, reject the hypothesis. A small chi square means the data fit. It does not prove the hypothesis.

A **pedigree** is a family tree drawn with set symbols. Two unaffected parents with an affected child means the trait is recessive. Two affected parents with an unaffected child means it is dominant. Recessive traits look horizontal and are more common after consanguineous matings. Dominant traits look vertical and can pass from father to son.

LOCK THESE IN

- Segregation is homologs parting in anaphase I. Independent assortment is random orientation at metaphase I.
- 3:1 hides 1:2:1. Two thirds of the dominant F_2 are heterozygous.
- For n heterozygous genes: 2^n gametes, 3^n genotypes, 2^n phenotypes, 4^n Punnett cells.
- "And" means multiply. "Or" means add. "At least one" means 1 minus none.
- The unaffected sibling of a person with a recessive condition has a 2/3 chance of being a carrier.
- Chi square uses counts, not percents. df = classes − 1. Critical values at 0.05: 3.841, 5.991, 7.815.
- Unaffected parents with an affected child: recessive. Affected parents with an unaffected child: dominant.
- Each pregnancy is independent. Chance has no memory.



Sixteen equally likely cells. Count by color: 9 : 3 : 3 : 1. A testcross (AaBb × aabb) gives 1 : 1 : 1 : 1.

A dihybrid cross, cell by cell. Each parent makes four gametes in equal numbers, so the square has sixteen equal cells. Grouping them by phenotype gives 9:3:3:1.

Ratios, rules and formulas

Item	Rule or formula	When to use it
Aa × *Aa*	1 *AA* : 2 *Aa* : 1 *aa*; 3 : 1 phenotypes	F_1 intercross or self
Aa × *aa*	1 : 1	monohybrid testcross
AaBb × *AaBb*	9 : 3 : 3 : 1	dihybrid intercross, unlinked genes
AaBb × *aabb*	1 : 1 : 1 : 1	dihybrid testcross
Trihybrid intercross	27 : 9 : 9 : 9 : 3 : 3 : 3 : 1	three unlinked genes
Product rule	P(A and B) = P(A) × P(B)	independent events that all happen
Sum rule	P(A or B) = P(A) + P(B)	mutually exclusive outcomes
Conditional probability	$P(A \mid B) = P(A \text{ and } B) \div P(B)$	some outcomes are ruled out by what you know
Binomial expansion	$P = [n! \div (s!\, t!)]\, p^s\, q^t$	"exactly s of n," any order
Complement	P(at least one) = 1 − P(none)	"at least one," "one or more"
Chi square	$\chi^2 = \Sigma\, (O - E)^2 \div E$	do counts fit a ratio?
Degrees of freedom	df = number of classes − 1	every genetic ratio test
Critical values, P = 0.05	df 1: 3.841; df 2: 5.991; df 3: 7.815; df 4: 9.488	reject H_0 if χ^2 is larger

Worked examples

WORKED EXAMPLE 1 · CHI SQUARE ON A TESTCROSS

A plant thought to be *Aa* is testcrossed. It gives 88 dominant and 112 recessive offspring. Do the data fit 1:1?

STEP 1 · STATE THE HYPOTHESIS

H_0: the offspring segregate 1:1. Total = 88 + 112 = 200, so E = 100 in each class.

STEP 2 · COMPUTE EACH TERM

$(88 - 100)^2 \div 100 = 144 \div 100 = 1.44$. $(112 - 100)^2 \div 100 = 1.44$.

STEP 3 · ADD AND FIND DF

$\chi^2 = 1.44 + 1.44 = 2.88$. Two classes, so df = 1.

STEP 4 · COMPARE

The critical value at P = 0.05 with df 1 is 3.841. Since 2.88 < 3.841, **fail to reject 1:1**. P lies between 0.05 and 0.10.

WHAT THIS DOES NOT SAY

Failing to reject does not prove the plant is *Aa*. It says chance alone could easily give a gap this size.

WORKED EXAMPLE 2 · EXACTLY TWO OF SIX

Two carriers of albinism (*Aa* × *Aa*) plan six children. What is the chance that exactly two have albinism?

STEP 1 · SET P AND Q

p = P(*aa*) = 1/4. q = P(unaffected) = 3/4. n = 6, s = 2, t = 4.

STEP 2 · COUNT THE ORDERS

$6! \div (2!\ 4!) = 720 \div (2 \times 24) = 15$.

STEP 3 · ONE ORDER

$(1/4)^2 \times (3/4)^4 = 1/16 \times 81/256 = 81/4{,}096$.

STEP 4 · MULTIPLY

$15 \times 81/4{,}096 =$ **1,215/4,096, about 0.297**.

SANITY CHECK

The expected number affected is 6 × 1/4 = 1.5, so two affected children should be one of the likelier outcomes. About 30 percent fits that.

WORKED EXAMPLE 3 · THE TWO THIRDS RULE IN A FAMILY

A man has cystic fibrosis. His parents and his sister are unaffected. The sister plans a child with an unrelated partner from a group where the carrier frequency is 1/30. What is the chance their first child is affected?

STEP 1 · THE PARENTS

Unaffected parents with an affected son must both be *Aa*.

STEP 2 · THE SISTER

She is unaffected, so she is not *aa*. P(she is *Aa*) = (1/2) ÷ (3/4) = 2/3.

STEP 3 · THE PARTNER

P(partner is *Aa*) = 1/30.

STEP 4 · THE CHILD

If both are carriers, P(*aa*) = 1/4.

STEP 5 · MULTIPLY

2/3 × 1/30 × 1/4 = 2/360 = **1/180, about 0.56 percent**.

COMPARE

Two people with no family history face 1/30 × 1/30 × 1/4 = 1/3,600. The family history raises the risk 20 fold. Whether to seek carrier testing is the family's choice to make.

WORKED EXAMPLE 4 · A MIXED CROSS BY BRANCHES

In *AaBbCc* × *AabbCc*, with complete dominance and unlinked genes, what fraction of offspring are *aabbcc*, and what fraction show all three dominant traits?

STEP 1 · SPLIT BY GENE

Aa × *Aa*: 1/4 *aa*, 3/4 *A_*. *Bb* × *bb*: 1/2 *bb*, 1/2 *Bb*. *Cc* × *Cc*: 1/4 *cc*, 3/4 *C_*.

STEP 2 · FULLY RECESSIVE

1/4 × 1/2 × 1/4 = **1/32**.

STEP 3 · FULLY DOMINANT

3/4 × 1/2 × 3/4 = **9/32**.

WHY IT WORKS

The genes assort independently, so each gene is its own small cross. Stay with phenotype fractions for every gene once you choose phenotype.

TRAPS PEOPLE FALL INTO

- **Answering 1/2 for the unaffected sibling.** Once you know the sibling is unaffected, *aa* is gone. The carrier chance is 2/3.
- **Running chi square on percents.** Always use raw counts. The same percents give a bigger chi square in a bigger sample.
- **Rounding expected values.** Expected numbers like 312.75 stay as decimals.
- **Setting df from the number of offspring.** df comes from classes. A 9:3:3:1 test has df 3 at any sample size.
- **Mixing genotype and phenotype fractions.** Pick one. Use 1/4, 1/2, 1/4 for genotype or 3/4, 1/4 for phenotype for every gene.
- **Thinking dominant means common.** Dominance is about the heterozygote's phenotype only.
- **Treating a small chi square as proof.** You fail to reject. Other hypotheses might fit just as well.

- **Expecting a run of unaffected children after one affected child.** Each child has the same 1/4 risk.

Chapter 1 self-test · ten items

1. Mendel's reciprocal crosses for seed shape gave the same results both ways. What did that show?

A. Both parents contribute equally to these traits
B. The trait is carried on the male parent alone
C. The dominant allele travels only in the pollen
D. Seed shape is passed through the cytoplasm

A. Both parents contribute equally to these traits. Matching results in both directions mean the sex of the parent does not matter. Different results would point to sex linkage or cytoplasmic inheritance, which Chapter 3 covers.

2. Which event in meiosis is the physical basis of segregation?

A. Egg and sperm nuclei fuse at fertilization
B. Chromosomes replicate before meiosis starts
C. Sister chromatids separate in anaphase II
D. Homologous chromosomes separate in anaphase I

D. Homologous chromosomes separate in anaphase I. Sutton and Boveri saw that homologs come in pairs and part into different cells in anaphase I, just as Mendel's paired factors do.

3. A black guinea pig is mated with a white one (*bb*) and has nine black pups. What is the chance of nine black pups if she were *Bb*?

A. $(3/4)^9$, about 0.075

B. 9 × 1/2, which is 4.5

C. $(1/2)^9$, about 0.002

D. $(1/4)^9$, nearly zero

C. $(1/2)^9$, about 0.002. In a testcross each pup from a *Bb* mother has a 1/2 chance of being black. Nine in a row is $(1/2)^9$ = 1/512. She is very likely *BB*, but only a white pup would settle it.

4. In *AaBbCcDd* × *AaBbCcDd*, how many different genotypes are possible?

A. 16

B. 81

C. 64

D. 256

B. 81. Each heterozygous gene gives three genotypes, so the count is 3^4 = 81. The 256 is the number of Punnett cells, 4^4.

5. Two carriers have a child. What is the chance it is an affected girl?

A. 3/8

B. 1/8

C. 1/2

D. 1/4

B. 1/8. Affected (1/4) and a girl (1/2) are independent, so multiply: 1/4 × 1/2 = 1/8.

6. Two carriers have three children. What is the chance that at least one is affected?

A 3/4

B 27/64

C 37/64

D 1/64

C. 37/64. Subtract the chance that none is affected: $1 - (3/4)^3 = 1 - 27/64 = 37/64$.

7. A dihybrid F_2 chi square comes to 6.2. What should you conclude at P = 0.05?

A Fail to reject, since 6.2 is below 7.815

B Reject, since 6.2 is above 3.841 for df 1

C Reject, since 6.2 is above 5.991 for df 2

D Accept 9:3:3:1 as proven by the data

A. Fail to reject, since 6.2 is below 7.815. Four classes give df 3, and the critical value is 7.815. The fit is acceptable. Using df 1 or 2 is the usual slip, and no chi square ever proves a hypothesis.

8. Why is 60 : 40 out of 100 a worse fit to 1:1 than 6 : 4 out of 10?

A The larger sample has more degrees of freedom

B Percents must be squared before running the test

C Small samples always fit any genetic ratio perfectly

D Chi square grows with sample size at equal percents

D. Chi square grows with sample size at equal percents. 60:40 gives $\chi^2 = 4.0$, but 6:4 gives only 0.4. Bigger samples give the test more power to detect a real departure. df stays at 1 in both.

9. In a pedigree, two affected parents have an unaffected daughter. What does this tell you?

A. The trait shows complete penetrance

B. The trait must be X linked

C. The trait must be recessive

D. The trait must be dominant

D. The trait must be dominant. If affected parents can have an unaffected child, they must each carry a hidden normal allele. That only works if the affected phenotype is dominant.

10. Which feature points toward autosomal recessive rather than dominant inheritance?

A. Each affected person has an affected parent

B. The trait shows in every single generation

C. The trait is more common after first cousin matings

D. Affected fathers pass it to about half their sons

C. The trait is more common after first cousin matings. Relatives are more likely to carry the same rare allele from a shared ancestor, so consanguinity raises recessive conditions. The other three describe a dominant, vertical pattern.

Read the full chapter: Chapter 1 · Mendelian Transmission and Probability

CHAPTER 2

Extensions of Mendel

What to know

Dominance describes the heterozygote, not the allele. In **complete dominance** the heterozygote looks like one homozygote, usually because one working copy is enough (the gene is

haplosufficient). In **incomplete dominance** the heterozygote is in between, as in pink snapdragons from red and white parents. The F_2 is 1 red : 2 pink : 1 white, so the phenotypes match the genotypes. In **codominance** both products show, as in the MN blood group.

The level of analysis can change the answer. The sickle cell allele is recessive for disease, incompletely dominant for cell shape at low oxygen, and codominant at the protein level, where both hemoglobin A and hemoglobin S show on a gel.

A population can hold many alleles of one gene, even though one person carries only two. With n alleles there are n(n + 1)/2 genotypes. ABO has three main alleles. I^A and I^B are each dominant to i and codominant with each other. Rabbit coat color shows a **dominance hierarchy**: $C > c^{ch} > c^h > c$.

A **lethal allele** removes a genotype class. The yellow mouse allele A^Y is dominant for coat color but recessive for lethality, so yellow by yellow gives 2 yellow : 1 agouti among live pups. Manx cats behave the same way. A late acting dominant lethal, such as the allele for Huntington disease, persists because it acts after people may have had children. A **conditional lethal**, such as a temperature sensitive allele, kills only under certain conditions.

Two genes can shape one trait. In chicken combs, *R_P_* is walnut, *R_pp* rose, *rrP_* pea and *rrpp* single. The ratio stays 9:3:3:1, but walnut and single are new phenotypes.

Epistasis is masking of one gene by another. The masking gene is epistatic and the hidden one is hypostatic. The 16 Punnett cells keep their usual frequencies, but some classes share a phenotype, so the ratio collapses. Learn 9:3:4, 12:3:1, 9:7, 15:1, 13:3 and 9:6:1, each with its pathway logic. To spot one, divide each count by one sixteenth of the total.

Penetrance is the percent of people with a genotype who show the trait at all. **Expressivity** is how strongly it shows in those who do. NF1 has penetrance near 100 percent but very variable expressivity. Penetrance can depend on age, as in Huntington disease. Modifier genes, the environment and chance all shape both. A **phenocopy** is an environmental mimic of a genetic phenotype, and a **norm of reaction** charts one genotype across many environments.

The **complementation test** crosses two recessive mutants with the same phenotype. A wild type F_1 means the mutations complement and sit in different genes. A mutant F_1 means they fail to complement and sit in the same gene. Mutants that fail to complement form one **complementation group**, which is one gene. The test needs recessive mutations.

LOCK THESE IN

- Incomplete dominance and codominance both give 1:2:1 phenotypes in the F_2.
- n alleles give n(n + 1)/2 genotypes: 3 alleles give 6, 4 give 10.
- A recessive lethal with a visible heterozygote gives 2:1. A twelve in the denominator hints at a lethal.
- Epistatic ratios still add to 16. Only the grouping changes.
- 9:7 means two steps in series. 15:1 means either of two genes is enough. 9:3:4 means a recessive block masks the second gene.
- Penetrance asks "does it show?" Expressivity asks "how much?"
- In a risk problem with reduced penetrance, multiply the transmission chance by the penetrance.
- Complementation (wild type F_1) means different genes. No complementation means the same gene.

Ratio	*A_B_* 9/16	*A_bb* 3/16	*aaB_* 3/16	*aabb* 1/16	Name
9:3:3:1	9	3	3	1	no merging (baseline)
9:3:4	9	3	3	1	recessive epistasis
12:3:1	9	3	3	1	dominant epistasis
9:7	9	3	3	1	complementary genes
15:1	9	3	3	1	duplicate dominant
13:3	9	3	3	1	dominant and recessive
9:6:1	9	3	3	1	cumulative effect

Same color means the classes look alike, and dashed lines mark classes that merged.
Every row still adds to 16. Divide each count by one sixteenth of the total to spot the ratio.

Epistasis only regroups. The sixteen Punnett cells keep their usual 9, 3, 3 and 1. Masking makes some classes look alike, so the ratio collapses.

Modified ratios and the logic behind them

Ratio	Name	Classes merged	Example
1 : 2 : 1	incomplete dominance or codominance	none; each genotype has its own phenotype	pink snapdragons; MN blood type
2 : 1	recessive lethal with visible heterozygote	homozygote dies	yellow mice, Manx cats
9 : 3 : 3 : 1	novel phenotypes from two genes	none	chicken comb shape
9 : 3 : 4	recessive epistasis	*aaB_* + *aabb*	Labrador coat; Bombay phenotype
12 : 3 : 1	dominant epistasis	*A_B_* + *A_bb*	summer squash color
9 : 7	duplicate recessive, complementary genes	all but *A_B_*	sweet pea flower color
15 : 1	duplicate dominant	all but *aabb*	shepherd's purse capsules
13 : 3	dominant and recessive	*A_B_* + *A_bb* + *aabb*	white Leghorn × white Wyandotte
9 : 6 : 1	duplicate genes, cumulative effect	*A_bb* + *aaB_*	summer squash shape

Formulas for this chapter

Quantity	Formula	Note
Genotypes from n alleles	n(n + 1) ÷ 2	n homozygotes plus n(n − 1)/2 heterozygotes
Penetrance	carriers who show the trait ÷ all carriers × 100%	a yes or no question across a group
Risk with reduced penetrance	P(inherit) × penetrance	for example 1/2 × 0.8 = 0.40
Carrier, given unaffected	P(carrier and unaffected) ÷ P(unaffected)	conditional probability again
One sixteenth of the F_2	total ÷ 16	divide each class by it to read the ratio

Worked examples

WORKED EXAMPLE 1 · ABO IN A FAMILY

A man with type AB blood has children with a woman of type A whose father was type O. What blood types can the children have, and in what ratio?

STEP 1 · GENOTYPES

The man is I^AI^B. The woman got i from her type O father, so she is I^Ai.

STEP 2 · GAMETES

Man: 1/2 I^A, 1/2 I^B. Woman: 1/2 I^A, 1/2 i.

STEP 3 · COMBINE

I^AI^A 1/4 (A), I^Ai 1/4 (A), I^AI^B 1/4 (AB), I^Bi 1/4 (B).

STEP 4 · READ IT

2 A : 1 AB : 1 B. No child can be type O, because the man has no i to give.

WORKED EXAMPLE 2 · NAMING AN EPISTATIC RATIO

Two dogs heterozygous at both coat genes have 320 pups in total over many litters: 180 black, 62 chocolate and 78 yellow. Which ratio fits?

STEP 1 · FIND ONE SIXTEENTH

320 ÷ 16 = 20.

STEP 2 · DIVIDE EACH CLASS

180 ÷ 20 = 9.0. 62 ÷ 20 = 3.1. 78 ÷ 20 = 3.9. That reads as 9 : 3 : 4.

STEP 3 · CONFIRM WITH CHI SQUARE

Expected: 180, 60 and 80. $\chi^2 = 0 + 4/60 + 4/80 = 0 + 0.067 + 0.050 = 0.117$.

STEP 4 · DECIDE

Three classes give df 2, with a critical value of 5.991. Since 0.117 is far below it, **fail to reject 9:3:4**. This is recessive epistasis: *ee* dogs are yellow whatever their *B* genotype.

WORKED EXAMPLE 3 · PENETRANCE AND AN UNAFFECTED CHILD

A dominant condition has 70 percent penetrance. A woman known to be heterozygous has a child with a man who does not carry the allele. (a) What is the chance the child is affected? (b) The child is unaffected. What is the chance the child carries the allele?

STEP 1 · AFFECTED

P(inherits) × penetrance = 1/2 × 0.7 = **0.35**.

STEP 2 · THE TWO WAYS TO BE UNAFFECTED

Carrier and unaffected: 1/2 × 0.3 = 0.15. Noncarrier: 1/2 × 1 = 0.50. Total unaffected = 0.65.

STEP 3 · CONDITION ON IT

P(carrier | unaffected) = 0.15 ÷ 0.65 = **3/13, about 0.23**.

WHY IT MATTERS

An unaffected child of an affected parent can still pass on an incompletely penetrant allele. The risk is lower than 1/2, but not zero.

WORKED EXAMPLE 4 · A LETHAL PLUS A SECOND GENE

Manx (*M*) is lethal when homozygous. A second, unlinked gene has *B* dominant to *b*. What ratio do you expect among living kittens of *MmBb* × *Mmbb*?

STEP 1 · THE MANX GENE AMONG SURVIVORS

Mm × *Mm* loses the *MM* quarter, so survivors are 2/3 Manx and 1/3 tailed.

STEP 2 · THE SECOND GENE

Bb × *bb* gives 1/2 *B_* and 1/2 *bb*.

STEP 3 · MULTIPLY

Manx *B_* = 2/3 × 1/2 = 2/6. Manx *bb* = 2/6. Tailed *B_* = 1/6. Tailed *bb* = 1/6.

STEP 4 · READ THE RATIO

2 : 2 : 1 : 1. The fractions add to 6/6, so nothing is missing.

TRAPS PEOPLE FALL INTO

- **Calling incomplete dominance blending.** Red and white come back unchanged in the F_2, so the alleles never mixed.
- **Mixing up epistasis and dominance.** Dominance is between alleles of one gene. Epistasis is between different genes.
- **Forgetting that dominance is set trait by trait.** The yellow mouse allele is dominant for color and recessive for lethality.
- **Reading penetrance as severity.** Penetrance is whether it shows. Expressivity is how severe it is.
- **Reading 9:7 as two separate traits.** It is one trait with two required steps in one pathway.
- **Doing a complementation test with dominant mutants.** A dominant allele makes the F_1 mutant either way, so the test tells you nothing.
- **Confusing complementation with recombination.** Complementation is scored in the F_1 and asks about function. Recombination asks about position.
- **Thinking a type O parent rules out an AB child.** Usually true, but the Bombay phenotype (*hh*) can hide I^A or I^B.

Chapter 2 self-test · ten items

1. Two pink four o'clock plants are crossed. What do you expect in the offspring?

A. 1 red : 2 pink : 1 white

B. 3 pink : 1 white

C. All pink, like the parents

D. 9 red : 3 pink : 4 white

A. 1 red : 2 pink : 1 white. Pink is the heterozygote in incomplete dominance. $C^RC^W \times C^RC^W$ gives 1:2:1, and each genotype has its own color.

2. A gene has five alleles in a population. How many genotypes are possible?

A 20

B 15

C 25

D 10

B. 15. n(n + 1)/2 = 5 × 6 ÷ 2 = 15: five homozygotes and ten heterozygotes.

3. At which level is the sickle cell allele codominant with the normal allele?

A Red cell shape in low oxygen

B Overall body size and growth

C Hemoglobin proteins on a gel

D Health under ordinary conditions

C. Hemoglobin proteins on a gel. Heterozygotes make both hemoglobin A and hemoglobin S, and both show. For health it acts recessive, and for cell shape it acts incompletely dominant.

4. Yellow mice crossed with yellow mice give about 2 yellow : 1 agouti. Why?

A Embryos with two copies of A^Y die early

B Agouti pups die soon after their birth

C Yellow mice are all homozygous for A^Y

D Yellow is dominant to agouti only in males

A. Embryos with two copies of A^Y die early. The A^Y allele is recessive lethal. Losing the homozygous quarter leaves 1/2 yellow and 1/4 agouti, which is 2:1 among survivors.

5. Why can the allele for Huntington disease persist even though the condition is fatal?

A. The allele is lethal only at high temperature

B. Symptoms usually begin after people may have had children

C. The allele causes death only in homozygotes

D. The allele is recessive, so carriers stay healthy

B. Symptoms usually begin after people may have had children. It is a late acting dominant lethal. Onset is often between 30 and 50, so the allele can pass on before it acts.

6. An F_2 of 400 flowers has 228 purple and 172 white. Which ratio fits best?

A. 3 purple : 1 white

B. 1 purple : 1 white

C. 15 purple : 1 white

D. 9 purple : 7 white

D. 9 purple : 7 white. One sixteenth is 25. Then 228 ÷ 25 = 9.1 and 172 ÷ 25 = 6.9. That reads as 9:7, two complementary genes acting in series.

7. In summer squash, a dominant *W* gives white fruit no matter what. What F_2 ratio follows?

A. 13 white : 2 yellow : 1 green

B. 12 white : 3 yellow : 1 green

C. 9 white : 6 yellow : 1 green

D. 9 white : 3 green : 4 yellow

B. 12 white : 3 yellow : 1 green. *W_Y_* and *W_yy* are both white, which is 9 + 3 = 12. That is dominant epistasis.

8. Of 200 people with a dominant allele, 150 show the trait. What is the penetrance?

- A 75 percent
- B 150 percent
- C 25 percent
- D 50 percent

A. 75 percent. Penetrance is carriers who show it over all carriers: 150 ÷ 200 = 0.75.

9. Thalidomide caused limb changes that look like those of a rare inherited syndrome. What is this called?

- A Reduced penetrance
- B A norm of reaction
- C Epistasis
- D A phenocopy

D. A phenocopy. A phenocopy is an environmentally caused mimic of a genetic phenotype. It is not inherited.

10. Two true breeding white eyed fly lines are crossed and every F_1 fly has red eyes. What can you conclude?

- A The two genes must be tightly linked
- B At least one mutation is dominant
- C The mutations lie in two different genes
- D The mutations lie in the same gene

C. The mutations lie in two different genes. A wild type F_1 means complementation. Each parent supplied a working copy of the gene the other lacked.

Read the full chapter: Chapter 2 · Extensions of Mendel

ONE PAGE

Cram sheet

Everything from chapters 1 and 2 in one place. Cover the right column and see if you can rebuild each row.

Ratios

Cross or situation	Ratio	Meaning
Monohybrid intercross	3 : 1 (1 : 2 : 1 genotypes)	complete dominance
Monohybrid testcross	1 : 1	unknown was a heterozygote
Incomplete dominance or codominance F_2	1 : 2 : 1	phenotypes match genotypes
Recessive lethal, visible heterozygote	2 : 1	homozygous class lost
Dihybrid intercross	9 : 3 : 3 : 1	unlinked genes
Dihybrid testcross	1 : 1 : 1 : 1	unlinked genes; excess parentals mean linkage
Epistasis	9:3:4, 12:3:1, 9:7, 15:1, 13:3, 9:6:1	still sixteenths, regrouped
Lethal plus second gene	twelfths, such as 6:3:2:1	four of sixteen cells lost

Formulas and numbers

Name	Formula or value	Use
Product rule	P(A) × P(B)	"and," independent events
Sum rule	P(A) + P(B)	"or," mutually exclusive
Conditional	P(A and B) ÷ P(B)	the 2/3 carrier rule
Binomial	$[n! \div (s!\ t!)]\ p^s\ q^t$	exactly s of n
Complement	1 − P(none)	at least one
Gene counts	2^n gametes, 3^n genotypes, 4^n cells	n heterozygous genes
Multiple alleles	n(n + 1) ÷ 2 genotypes	allelic series
Chi square	$\Sigma\ (O - E)^2 \div E$, df = classes − 1	use counts
Critical values at 0.05	3.841 (df 1), 5.991 (df 2), 7.815 (df 3)	reject if larger
Penetrance in risk	P(inherit) × penetrance	reduced penetrance

Rules that are not formulas

Topic	The rule
Recessive pedigree	Unaffected parents have an affected child. Horizontal pattern. Consanguinity raises it.
Dominant pedigree	Affected parents have an unaffected child. Vertical pattern. Male to male transmission possible.
Testcross	One recessive offspring proves the unknown is heterozygous.
Dominance	Describes the heterozygote phenotype only, and can change with the level of analysis.
ABO	I^A and I^B are codominant, and both are dominant to *i*. Bombay (*hh*) types as O.
Epistasis	An early block in a pathway masks later steps. Divide counts by total ÷ 16 to read the ratio.
Penetrance vs expressivity	Does it show at all, versus how strongly it shows.
Complementation	Wild type F_1: different genes. Mutant F_1: same gene. Needs recessive mutations.

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