

2

Mendel's Laws of Inheritance

2.1: Learning Objectives

By the end of this chapter, the reader should be able to state and apply Mendel's Law of Segregation and Law of Independent Assortment, construct and interpret Punnett squares, compute expected offspring ratios for mono- and di-hybrid crosses, and carry out a chi-square goodness-of-fit test of segregation ratios using Nigerian family data.

2.2: Historical Note



Gregor Mendel (1822–1884), whose breeding experiments on the garden pea established the quantitative laws of inheritance.

Gregor Mendel (1822–1884), an Augustinian monk working in Brno (in present-day Czech Republic), established the quantitative rules of inheritance through carefully designed breeding experiments on the garden pea (*Pisum sativum*), published in 1866. Although Mendel worked before the discovery of DNA or even chromosomes, his statistical reasoning about the segregation of discrete “factors” (what we now call genes) remains the foundation of transmission genetics and, by extension, of statistical genetics.



*The garden pea, *Pisum sativum*, Mendel's original experimental organism.*

Mendel's genius lay in his choice of experimental material and his statistical rigour. He selected seven traits in the pea plant that each showed clear, discrete alternative forms (rather than a continuous range), that bred true across generations when self-fertilised, and that turned out — fortuitously — to be controlled by seven different genes, each showing simple dominant/recessive inheritance:

Character	Traits	
	Dominant	Recessive
Flower color	 Purple	 White
Flower position	 Axial	 Terminal
Seed color	 Yellow	 Green
Seed shape	 Round	 Wrinkled
Pod shape	 Inflated	 Constricted
Pod color	 Green	 Yellow
Stem length	 Tall	 Dwarf

Mendel's seven pea traits, each with a clearly dominant and a clearly recessive form: flower colour, flower position, seed colour, seed shape, pod shape, pod colour, and stem length.

By counting, precisely, the numbers of each type of offspring across thousands of plants — a scale of quantitative record-keeping unusual for his time — Mendel was able to detect the underlying 3:1 and 9:3:3:1 ratios (Sections 2.3–2.4) that a smaller, less carefully documented study would have missed amid ordinary sampling variability, foreshadowing the statistical hypothesis-testing methods (Section 2.5) that would only be formalised decades later by Karl Pearson and R.A. Fisher.

2.3: The Law of Segregation

Mendel's First Law (Law of Segregation): Each individual carries two alleles for a given trait; these two alleles separate (segregate) during the formation of gametes, so that each gamete carries only one allele, chosen at random with probability $1/2$ each.

Consider a single autosomal locus with alleles A (dominant) and a (recessive). A heterozygous parent (Aa) produces gametes carrying A or a, each with probability $1/2$. If both parents are heterozygous (Aa $\times$ Aa), the Punnett square gives:

	A ($1/2$)	a ($1/2$)
A ($1/2$)	AA ($1/4$)	Aa ($1/4$)
a ($1/2$)	Aa ($1/4$)	aa ($1/4$)

The expected genotype ratio among offspring is 1 AA : 2 Aa : 1 aa, and if A is completely dominant, the expected phenotype ratio is 3 (dominant) : 1 (recessive).

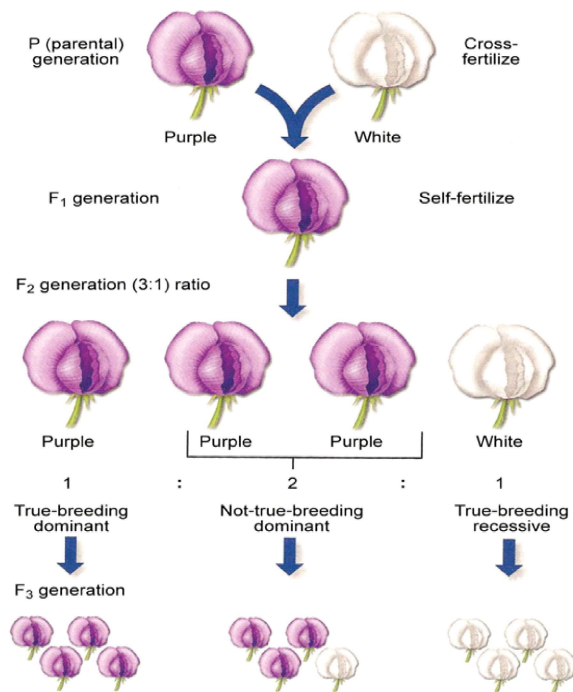
Worked Example 2.1: (Sickle-cell trait, Ibadan)

Consider two parents who are both carriers of sickle-cell trait (genotype AS). By the Law of Segregation, each parent transmits A or S to a given child with probability $1/2$. The possible offspring genotypes and their probabilities are:

- AA (normal): $P = 1/2 \times 1/2 = 1/4$
- AS (carrier/trait): $P = 2 \times (1/2 \times 1/2) = 1/2$
- SS (sickle-cell disease): $P = 1/2 \times 1/2 = 1/4$

This 1:2:1 genotypic ratio (equivalently, a 3:1 “unaffected : affected” phenotypic ratio, since AA and AS are both clinically unaffected while SS is affected) is precisely why genetic counselling clinics in Nigeria — for instance, at UCH Ibadan and LUTH Lagos — advise that when both partners in a couple carry the AS genotype, each pregnancy carries a 25% risk of a child with sickle-cell disease, a 50% chance of a carrier (AS) child, and a 25% chance of a normal (AA) child.

2.4: The Law of Independent Assortment



A monohybrid cross tracking flower colour across the P, F₁, and F₂ generations, illustrating the 3:1 segregation ratio.

Mendel's Second Law (Law of Independent Assortment):

Alleles at different loci segregate independently of one another during gamete formation, provided the loci lie on different chromosomes (or are far apart on the same chromosome).

Consider two loci: HBB (sickle-cell, alleles A/S) and the ABO locus (alleles I^A , I^B , i), which lie on different chromosomes (HBB is on chromosome 11; ABO is on chromosome 9) and therefore assort independently. If a parent is doubly heterozygous $AS I^A i$, this parent produces four types of gametes — $A I^A$, $A i$, $S I^A$, $S i$ — each with probability $1/4$, because the segregation at HBB (A vs S , each $1/2$) is statistically independent of the segregation at ABO (I^A vs i , each $1/2$), and $1/2 \times 1/2 = 1/4$.

Worked Example 2.2: (Dihybrid cross)

Suppose both parents are $AS I^A i$. Because the two loci assort independently, the joint genotype probabilities of the offspring are simply the product of the separate (Mendelian) probabilities at each locus. For example, $P(SS \text{ and } I^A I^A) = P(SS) \times P(I^A I^A) = (1/4)(1/4) = 1/16$. The complete dihybrid phenotype ratio, when both traits show complete dominance, is the classical 9:3:3:1 ratio.

Theorem 2.1: (The 9:3:3:1 Dihybrid Ratio)

Consider a cross between two individuals doubly heterozygous at two independently assorting loci, $AaBb \times AaBb$, with A dominant to a and B dominant to b . Then, among the offspring, the four phenotype classes occur in the ratio 9 ($A_B_$) : 3 (A_bb) : 3 ($aaB_$) : 1 ($aabb$), where $A_$ denotes “ A dominant phenotype, genotype AA or Aa .”

Proof. By Mendel's First Law (Theorem 1.1) applied separately to each locus, a single-locus $Aa \times Aa$ cross gives offspring genotype ratio 1 AA : 2 Aa : 1 aa , so $P(A_)=P(AA)+P(Aa)=1/4+2/4=3/4$, and $P(aa)=1/4$ — the standard monohybrid 3:1

phenotype ratio (Section 2.3). By Mendel's Second Law (independent assortment, this section), the joint probability of the two-locus phenotype is the product of the single-locus probabilities:

$$P(A_B_) = P(A_) \times P(B_) = \frac{3}{4} \times \frac{3}{4} = \frac{9}{16}, \quad P(A_bb) = \frac{3}{4} \times \frac{1}{4} = \frac{3}{16},$$

$$P(aaB_) = \frac{1}{4} \times \frac{3}{4} = \frac{3}{16}, \quad P(aabb) = \frac{1}{4} \times \frac{1}{4} = \frac{1}{16}.$$

Multiplying each probability by 16 gives the integer ratio 9:3:3:1, and the four probabilities sum to $16/16 = 1$, confirming they exhaust all possible phenotype combinations.

Worked Example 2.2a: (Dihybrid ratio in a Nigerian genetics teaching laboratory)

To teach Theorem 2.1 practically, a genetics class at a Nigerian university crosses maize plants heterozygous for two independently assorting traits: kernel colour (Purple P dominant to yellow p) and kernel texture (Smooth S dominant to wrinkled s), i.e. $PpSs \times PpSs$ — a classical dihybrid design directly analogous to Mendel's own pea-plant crosses (Section 2.2). Theorem 2.1 predicts that, of 320 offspring kernels counted on a single maize cob, the class expects $320 \times 9/16 = 180$ purple-smooth, $320 \times 3/16 = 60$ purple-wrinkled, $320 \times 3/16 = 60$ yellow-smooth, and $320 \times 1/16 = 20$ yellow-wrinkled kernels. If the class instead observes 175, 55, 68, and 22 respectively, a chi-square goodness-of-fit test (Section 2.5) can be used to assess whether this modest departure from the 9:3:3:1 theoretical ratio is within the range expected from ordinary sampling variability, or whether it suggests the two loci are not, in fact, assorting completely independently (e.g., due to linkage, Chapter 8).

2.5: Chi-Square Test of Mendelian Segregation Ratios

Because real family or breeding data are subject to sampling variation, Mendelian ratios are tested statistically rather than expected to hold exactly. The **Pearson chi-square goodness-of-fit test** compares observed counts (O_i) to those expected (E_i) under a hypothesised Mendelian ratio:

$$\chi^2 = \sum_{i=1}^k (O_i - E_i)^2 / E_i$$

which, under the null hypothesis, is approximately distributed as chi-square with $(k - 1)$ degrees of freedom, where k is the number of phenotype categories.

Worked Example 2.3:

A Nigerian secondary-school biology field study of tongue-rolling ability (a trait often taught, with caveats, as a simple dominant/recessive Mendelian trait) in the offspring of two “roller” (heterozygous, assumed Rr) parents records, among 160 children sampled across several families: 128 rollers, 32 non-rollers. Under the hypothesised 3:1 ratio, the expected counts are $E_{\text{roller}} = 120$, $E_{\text{non-roller}} = 40$.

$$\chi^2 = (128 - 120)^2 / 120 + (32 - 40)^2 / 40 = 64 / 120 + 64 / 40 = 0.53 + 1.60 = 2.13$$

With 1 degree of freedom, the critical value at $\alpha = 0.05$ is 3.84. Since $2.13 < 3.84$, we do not reject the null hypothesis: the data are consistent with the expected 3:1 Mendelian ratio.

2.6: Extensions: Incomplete Dominance and Multiple Alleles

Not every trait shows complete dominance. Under **incomplete dominance**, the heterozygote’s phenotype is intermediate between the two homozygous phenotypes (relevant to many quantitative traits treated in Chapter 9). Under **multiple allelism**, more than

two alleles segregate at a locus in the population (though any one individual still carries only two), as with the ABO blood group (I^A , I^B , i) discussed in Chapter 1 — the segregation principle still applies allele-by-allele.

Theorem 2.2: (The 9:3:3:1 Dihybrid Ratio)

Let two heterozygous parents ($AaBb \times AaBb$) be crossed at two unlinked, independently assorting autosomal loci, each showing complete dominance. Then the expected phenotypic ratio among offspring is 9 ($A_B_$) : 3 (A_bb) : 3 ($aaB_$) : 1 ($aabb$).

Proof. By the Law of Segregation (Theorem 1.1) applied separately to each locus, a single-locus $Aa \times Aa$ cross yields offspring genotype ratios 1 AA : 2 Aa : 1 aa , and under complete dominance this gives a phenotypic ratio of 3 (dominant) : 1 (recessive) at each locus independently. By Theorem 1.2, the phenotypic outcome at the A locus is independent of the phenotypic outcome at the B locus. The joint probability of any combination of phenotypes is therefore the product of the marginal probabilities: $P(A_B_) = (3/4)(3/4) = 9/16$, $P(A_bb) = (3/4)(1/4) = 3/16$, $P(aaB_) = (1/4)(3/4) = 3/16$, and $P(aabb) = (1/4)(1/4) = 1/16$, which is exactly the 9:3:3:1 ratio.

2.7: Chapter Summary

- The Law of Segregation: the two alleles at a locus separate into different gametes with equal probability.
- The Law of Independent Assortment: alleles at unlinked loci segregate independently, so joint genotype probabilities multiply.
- The chi-square goodness-of-fit test provides a formal statistical check of whether observed family data are consistent with a hypothesised Mendelian ratio.

- These laws, extended by co-dominance, incomplete dominance and multiple allelism, provide the probabilistic building blocks for population and quantitative genetics.

2.8: Computational Corner: Testing Segregation Ratios

R.

```
# Monohybrid cross (Section 2.3): 3:1 ratio
observed_mono <-c(dominant =732, recessive =268) # e.g.,
pigmented vs albino seedlings
expected_ratio_mono <-c(3, 1)
test_mono <-chisq.test(observed_mono, p =
expected_ratio_mono /sum(expected_ratio_mono))
print(test_mono)

# Dihybrid cross (Section 2.4): independent assortment predicts
a 9:3:3:1 ratio
# e.g., seed shape (round/wrinkled) x seed colour (yellow/green)
in an F2 generation
observed_di <-c("Round-Yellow"=315, "Round-Green"=108,
"Wrinkled-Yellow"=101, "Wrinkled-Green"=32)
expected_ratio_di <-c(9, 3, 3, 1)
test_di <-chisq.test(observed_di, p = expected_ratio_di /
sum(expected_ratio_di))
print(test_di)

# Plot observed counts against the two hypothesised Mendelian
ratios
par(mfrow=c(1, 2))
expected_mono <-sum(observed_mono) *
expected_ratio_mono /sum(expected_ratio_mono)
barplot(observed_mono, col ="steelblue",
main =sprintf("Monohybrid\nchi2=%.2f, p=%.3f",
test_mono$statistic, test_mono$p.value))
```

```

points(x = c(0.7, 1.9), y = expected_mono, pch = 18, col
      = "darkorange", cex = 2)

expected_di <- sum(observed_di) * expected_ratio_di /
sum(expected_ratio_di)
barplot(observed_di, col = "steelblue", las = 2,
main     = sprintf("Dihybrid\nchi2=%.2f,    p=%.3f",
test_di$statistic, test_di$p.value))
points(x = c(0.7, 1.9, 3.1, 4.3), y = expected_di, pch = 18, col
      = "darkorange", cex = 2)

```

Python

```

import numpy as np
from scipy.stats import chisquare
import matplotlib.pyplot as plt

# Monohybrid cross (Section 2.3): 3:1 ratio
observed_mono = np.array([732, 268])
expected_mono = observed_mono.sum() * np.array([3/4, 1/4])
stat_mono, p_mono = chisquare(observed_mono,
f_exp=expected_mono)
print(f"Monohybrid: chi2={stat_mono:.3f}, p={p_mono:.4f}")

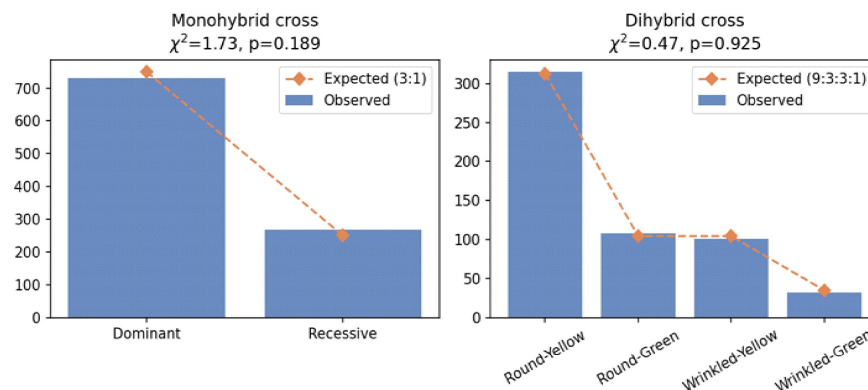
# Dihybrid cross (Section 2.4): independent assortment predicts
9:3:3:1
observed_di = np.array([315, 108, 101, 32]) # round-yellow,
round-green, wrinkled-yellow, wrinkled-green
labels_di = ["Round-Yellow", "Round-Green", "Wrinkled-
Yellow", "Wrinkled-Green"]
expected_di = observed_di.sum() * np.array([9, 3, 3, 1]) / 16
stat_di, p_di = chisquare(observed_di, f_exp=expected_di)
print(f"Dihybrid: chi2={stat_di:.3f}, p={p_di:.4f}")
print("Observed:", observed_di, "Expected:",
np.round(expected_di, 1))

```

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fig, axes = plt.subplots(1, 2, figsize=(9, 4))
axes[0].bar(["Dominant", "Recessive"], observed_mono,
color="#4C72B0", alpha=0.85, label="Observed")
axes[0].plot(["Dominant", "Recessive"], expected_mono,
"D—", color="#DD8452", label="Expected (3:1)")
axes[0].set_title(f"Monohybrid cross\n $\chi^2$ = $\{stat\_mono:.2f\}$ ,  $p$ = $\{p\_mono:.3f\}$ ")
axes[0].legend()
axes[1].bar(labels_di, observed_di, color="#4C72B0",
alpha=0.85, label="Observed")
axes[1].plot(labels_di, expected_di, "D—", color="#DD8452",
label="Expected (9:3:3:1)")
axes[1].set_title(f"Dihybrid cross\n $\chi^2$ = $\{stat\_di:.2f\}$ ,
 $p$ = $\{p\_di:.3f\}$ ")
axes[1].tick_params(axis='x', rotation=30)
axes[1].legend()
plt.tight_layout()
plt.savefig("ch02_segregation_tests.png", dpi=150)

```



Observed F_2 counts against the two classical Mendelian ratios: 3:1 for a monohybrid cross (left) and 9:3:3:1 for a dihybrid cross testing independent assortment (right). Both fits are statistically consistent with Mendel's laws ($p > 0.05$).

Worked Example 2.4: (dihybrid cross, Section 2.4 revisited computationally)

The seed-shape/seed-colour F2 data above (round-yellow 315, round-green 108, wrinkled-yellow 101, wrinkled-green 32; $n = 556$) give $\chi^2 = 0.47$ on 3 degrees of freedom (4 categories “1”), far below the critical value of 7.81 at $\alpha = 0.05$, so the data are entirely consistent with the 9:3:3:1 ratio predicted by independent assortment of the two seed-trait loci — the same F2 pattern, and essentially the same numbers, that Mendel himself reported for this cross in 1866, illustrating why his original data are still used to teach the chi-square goodness-of-fit test more than 150 years later.

Both give the same chi-square statistic and p-value for testing Mendel’s 3:1 segregation ratio; a non-significant p-value (as here) supports the single-gene, complete-dominance model of Section 2.3, while a significant departure would prompt the extensions of Section 2.6 (incomplete dominance, multiple alleles) or the ascertainment-aware methods of Chapter 8.

Mini-Project

Mini-Project 2: Testing a Real or Simulated Dihybrid Cross

Design and analyse a simulated dihybrid cross to test both Mendelian laws simultaneously.

Step 1. Simulate an F2 generation of at least 400 offspring from a dihybrid cross at two unlinked loci.

Step 2. Tabulate the observed four-phenotype counts and compare them to the theoretical 9:3:3:1 ratio.

Step 3. Perform a chi-square test of goodness-of-fit (Section 2.5) and report the test statistic, degrees of freedom, and p-value.

Step 4. Repeat the simulation assuming the two loci are tightly linked (recombination fraction 0.05) and observe how the ratio distorts.

Step 5. Write a short paragraph explaining why the linked-loci simulation no longer fits a 9:3:3:1 ratio, foreshadowing Chapter 8.

Deliverable: *Simulation code, a results table for both the unlinked and linked scenarios, and a chi-square test summary for each.*

2.9: Practice Questions

Q1. Two AS (sickle-cell trait) parents in Kano plan to have four children. What is the probability that exactly one of the four children has sickle-cell disease (SS)? (Assume independence across children and apply the binomial distribution built on the Mendelian 1/4 probability of SS per child.)

Q2. A man of genotype $AS\ I^A B\ i$ marries a woman of genotype $AA\ I^A i$. Assuming the HBB and ABO loci assort independently, find the probability that a child of this couple is AS and blood group O.

Q3. In a genetics practical at a Nigerian university, 200 offspring of a monohybrid cross are classified as 160 dominant-phenotype and 40 recessive-phenotype. Test, at the 5% level, whether this is consistent with the expected 3:1 ratio.

Q4. Explain why sickle-cell trait (AS) and sickle-cell disease (SS) illustrate, respectively, co-dominance (at the molecular/haemoglobin level) and recessiveness (at the clinical/phenotype level).