chapter 1

Mendel's Principles of Heredity

Synopsis

Chapter 1 covers the basic principles of inheritance that can be summarized as Mendel's Laws of Segregation (for one gene) and Independent Assortment (for more than one gene).

Key terms

genes and **alleles** of genes – A gene determines a trait, and different alleles or forms of a gene exist. The color gene in peas has two alleles: yellow and green.

genotype and **phenotype** – Genotype is the genetic makeup of an organism (written as alleles of specific genes), while phenotype is how the organism looks.

homozygous and **heterozygous** – When both alleles of a gene are the same, the individual is homozygous for that gene (or **pure breeding**). If the two alleles are different, the organism is heterozygous (also called a **hybrid**).

dominant and **recessive** – The dominant allele is the one that controls phenotype in the heterozygous genotype; the recessive allele controls phenotype only in a homozygote.

monohybrid or **dihybrid cross** – a cross between individuals who are both heterozygotes for one gene (monohybrid) or for two genes (dihybrid).

testcross – performed to determine if an individual with the dominant characteristic is homozygous or heterozygous: An individual with the dominant phenotype but unknown genotype is crossed with an individual with the recessive phenotype.

Key ratios

3:1 – Ratio of progeny phenotypes in a cross between monohybrids
[$Aa \times Aa \rightarrow 3\ A–$ (dominant phenotype) : 1 aa (recessive phenotype)]

1:2:1 – Ratio of progeny genotypes in a cross between monohybrids
($Aa \times Aa \rightarrow 1\ AA : 2\ Aa : 1aa$)

1:1 – Ratio of progeny genotypes in a cross between a heterozygote and a recessive homozygote
($Aa \times aa \rightarrow 1\ Aa : 1aa$)

1:0 – All progeny are the same phenotype. Can result from either of two cases:
[$AA \times – – \rightarrow A–$ (all dominant phenotype)]
[$aa \times aa \rightarrow aa$ (all recessive phenotype)]

9:3:3:1 – Ratio of progeny phenotypes in a dihybrid cross
($Aa\ Bb \times Aa\ Bb \rightarrow 9\ A–\ B– : 3\ A–\ bb : 3\ aa\ B– : 1\ aa\ bb$)

Problem Solving

The essential component of solving most genetics problems is to DIAGRAM THE CROSS in a consistent manner. Usually you will be given information about phenotypes, so the diagram would be:

Phenotype of one parent × phenotype of the other parent → phenotype(s) of progeny

The goal is to assign genotypes to the parents and then use these predicted genotypes to generate the genotypes, phenotypes, and ratios of progeny. If the predicted progeny match the observed data you were provided, then your genetic explanation is plausible.

The points listed below will be particularly helpful in guiding your problem solving:

- Remember that **two alleles of each gene exist when describing the genotypes of individuals.** But if you are describing gametes, remember that **only one allele of each gene is in a gamete.**
- **You will need to determine whether a character is dominant or recessive.** Two main clues will help you answer this question.
    - First, if the parents of a cross are true breeding for the alternative characters of the trait, look at the phenotype of the F_1 progeny. Their genotype must be heterozygous, and their phenotype is thus determined by the dominant allele of the gene.
    - Second, look at the F_2 progeny (that is, the progeny of the F_1 hybrids). The 3/4 portion of the 3:1 phenotypic ratio indicates the dominant character.
- You should **recognize the need to set up a testcross** (to establish the genotype of an individual showing the dominant character by crossing this individual to a homozygote for the recessive allele).
- You must **keep in mind the basic rules of probability:**
    - *Product rule:* If two outcomes must occur together as the result of independent events, the probability of one outcome AND the other outcome is the product of the two individual probabilities.
    - *Sum rule:* If there is more than one way in which an outcome can be produced, the probability of one OR the other occurring is the sum of the two mutually exclusive individual probabilities.
- Be aware that sometimes you need to use **conditional probability**, meaning that an event's probability is influenced by its relationship to another event that has already occurred. You were introduced to conditional probability in Solved Problem III in this chapter, and several of the problems in Section 1.3 require this kind of thinking. For example, suppose you are given a pedigree diagram for a disease caused by a recessive allele. You are asked to determine the chance that an unaffected individual is a carrier (*Dd*), when both parents are carriers. As the cross that produced the unaffected individual is *Dd* × *Dd,* you would expect the chance of a *Dd* child to be 1/2. This is true, but it was not the question you were asked! You know something about the individual in question—which is that they are unaffected—they cannot be *dd*. This means that in this case, the 1 *DD* : 2 *Dd* : 1 *dd* ratio changes to 1 *DD* : 2 *Dd*, and the chance is 2/3 that the unaffected individual is a carrier. When solving probability problems in pedigrees,

always think carefully about what you know (and what you don't know) about each individual.

- Remember that **Punnett squares are not the only means of analyzing a cross**; **branched-line diagrams and calculations of probabilities using the product and sum rules are more efficient ways of looking at complicated crosses** involving more than one or two genes.
- **You should be able to draw and interpret pedigrees**. When the trait is rare, look for vertical patterns of inheritance characteristic of dominant traits, and horizontal patterns that typify recessive traits. Check your work by assigning genotypes to all individuals in the pedigree and verifying that these make sense.
- The vocabulary problem (the first problem in the set) is a useful gauge of how well you know the terms most crucial for your understanding of the chapter.

Vocabulary

1.

a.	phenotype	4.	observable characteristic
b.	alleles	3.	alternate forms of a gene
c.	independent assortment	6.	alleles of one gene separate into gametes randomly with respect to alleles of other genes
d.	gametes	7.	reproductive cells containing only one copy of each gene
e.	gene	11.	the heritable entity that determines a characteristic
f.	segregation	13.	the separation of the two alleles of a gene into different gametes
g.	heterozygote	10.	an individual with two different alleles of a gene
h.	dominant	2.	the allele expressed in the phenotype of the heterozygote
i.	F_1	14.	offspring of the P generation
j.	testcross	9.	the cross of an individual of ambiguous genotype with a homozygous recessive individual
k.	genotype	12.	the alleles an individual has
l.	recessive	8.	the allele that does not contribute to the phenotype of the heterozygote
m.	dihybrid cross	5.	a cross between individuals both heterozygous for two genes
n.	homozygote	1.	having two identical alleles of a given gene

Section 1.1

2. Prior to Mendel, **people held two basic misconceptions about inheritance.** First was the common idea of **blended inheritance:** that the parental characteristics become mixed in the offspring and forever changed. Second, many people thought that **one parent contributes the most to an offspring's inherited features.** (For example, some people thought they saw a fully formed child in a human sperm.)

In addition, **people who studied inheritance did not approach the problem in an organized way.** They did not always control their crosses. They did not look at traits with clear-cut alternative characteristics. They did not start with pure-breeding lines. They did not count the progeny types in their crosses. For these reasons, they could not develop the same insights as did Mendel.

3. **Several advantages exist for using peas for the study of inheritance:**

- Peas have a fairly rapid generation time (at least two generations per year if grown in the field, three or four generations per year if grown in greenhouses).
- Peas can either self-fertilize or be crossed artificially by an experimenter.
- Peas produce large numbers of offspring (hundreds per parent).
- Peas can be maintained as pure-breeding lines, simplifying the ability to perform subsequent crosses.
- Because peas have been maintained as inbred stocks, two easily distinguished and discrete forms of many traits are known.
- Peas are easy and inexpensive to grow.

In contrast, **studying genetics in humans has several disadvantages:**

- The generation time of humans is very long (roughly 20 years).
- No self-fertilization occurs in humans, and it is not ethical to manipulate crosses.
- Humans produce only a small number of offspring per mating (usually only one) or per parent (almost always fewer than 20).
- Although people who are homozygous for a trait do exist (analogous to pure-breeding stocks), homozygosity cannot be maintained because mating with another individual is needed to produce the next generation.
- Because human populations are not inbred, most human traits show a continuum of phenotypes; only a few traits have two distinct forms.
- People require a lot of expensive care to "grow".

One major advantage exists nonetheless for the study of genetics in humans: Because many inherited traits result in disease syndromes, and because the world's population now exceeds 7 billion people, a very large number of people with diverse, variant phenotypes can be recognized. These variations are the raw material of genetic analysis.

Section 1.2

4. a. **Two phenotypes are seen in the second generation of this cross: normal and albino**. Thus, only one gene with two alleles is needed to control the phenotypes observed. **The 3:1 ratio of these phenotypes in the F_2 generation** will be seen only if a single gene is involved.

b. Note that the phenotype of the first generation progeny is normal color, and that in the second generation, there is a ratio of 3 normal : 1 albino. Both observations show that **the allele controlling the normal phenotype (*A*) is dominant to the allele controlling the albino phenotype (*a*).**

c. In a testcross, an individual showing the dominant phenotype but that has an unknown genotype (the normal-colored female parent in this problem) is mated with an individual that shows the recessive phenotype and is therefore homozygous for the recessive allele. In this case, the individual with the recessive phenotype must be both male and an albino, so **the male parent's genotype is *aa*.** The normally colored offspring must receive an *A* allele from the mother, so **the genotype of the normal offspring of the testcross is *Aa*.** The albino offspring must receive an *a* allele from the mother, so **the genotype of the albino offspring of the testcross is *aa*.** Thus, **the female parent must be heterozygous *Aa*.**

5. Because two different phenotypes result from the mating of two cats of the same phenotype, and because the ratio of the short-haired to long-haired progeny is 3:1, only a single gene is involved, and the short-haired parent cats must have been heterozygous. The phenotype expressed in the heterozygotes (the parent cats) is the dominant phenotype. Therefore, **short hair is dominant to long hair.**

6. a. Two affected individuals have an affected child and a normal child. This outcome is not possible if the affected individuals were homozygous for a recessive allele conferring piebald spotting, and if the trait is controlled by a single gene. Therefore, **piebald must be the dominant characteristic.**

b. If the trait is dominant, the piebald parents could be either homozygous (*PP*) or heterozygous (*Pp*). However, because the two affected individuals have an unaffected child (*pp*), **they both must be heterozygous (*Pp*).** A diagram of the cross follows:

piebald × piebald → 1 piebald : 1 normal
Pp *Pp* *Pp* *pp*

Note that although the apparent ratio is 1:1, this is not a testcross but is instead a cross between two monohybrids. The reason for this discrepancy is that only two progeny were obtained, so this number is insufficient to establish what the true ratio would be (it should be 3:1) if many progeny resulted from the mating.

7. **You would conduct a testcross between your normal-winged fly (*W*–) and a short-winged fly that must be homozygous recessive (*ww*).** The possible results are diagrammed here; the first genotype in each cross is that of the normal-winged fly whose genotype was originally unknown. Note that **if the normal-winged fly is a homozygote,**

all the progeny should have normal wings, but if the normal-winged fly is a heterozygote, half the progeny should have normal wings and the other half should have short wings.

$$WW \times ww \rightarrow \text{all } Ww \text{ (normal wings)}$$

$$Ww \times ww \rightarrow 1/2\ Ww \text{ (normal wings)} : 1/2\ ww \text{ (short wings)}$$

8. First diagram the crosses:

closed × open → F_1 all open → F_2 145 open : 59 closed

F_1 open × closed → 81 open : 77 closed

The results of the crosses fit the pattern of inheritance of a single gene, with open being dominant to closed. The first cross is similar to those Mendel did with pure-breeding parents, although you were not provided with the information that the starting plants were true-breeding. **The F_1 plants are open, indicating that open is dominant. The closed parent must be homozygous for the recessive allele. Because only one phenotype is seen among the F_1 plants, the open parent must be homozygous for the dominant allele.** Thus, the parental cucumber plants were indeed true-breeding homozygotes.

Self-fertilization of the F_1 plants results in a 3:1 ratio of open : closed among the F_2 progeny. **The 3:1 ratio in the F_2 shows that a single gene controls the trait and that the F_1 plants are all monohybrids (that is, they are heterozygotes).**

The final cross verifies that the F_1 plants from the first cross are heterozygotes because **this testcross yields a 1:1 ratio of open: closed progeny.** In summary, all the data are consistent with the trait being determined by one gene with two alleles, and open being the dominant characteristic. Rewritten as genotypes for a gene with alleles *O* and *o*, the crosses were:

oo (closed) × *OO* (open) → F_1 *Oo* (open) → F_2 145 *O*– (open): 59 *oo* (closed)

F_1 *Oo* (open) × *oo* (closed) → 81 *Oo* (open) : 77 *oo* (closed)

9. **The dominant characteristic (short tail) is easier to eliminate from the population by selective breeding. The reason is you can recognize every animal that has inherited the *short tail* allele, because only one such dominant allele is needed to see the characteristic. If you prevent all the short-tailed animals from mating, then the allele would become extinct.**

On the other hand, the recessive *dilute* allele can be passed unrecognized from generation to generation in heterozygous mice (who are *carriers*). The heterozygous mice are not dilute, so they cannot be distinguished from homozygous dominant mice with normal coat color. You could prevent the homozygous recessive mice with the dilute characteristic from mating, but the *dilute* allele would remain among the carriers, which you could not recognize.

10. The problem states that only one gene is involved in this trait, and that the dominant allele is dimpled (*D*) while the recessive allele is nondimpled (*d*). (We are using Mendelian symbols here instead of human symbols for genotypes to emphasize which allele is dominant and which is recessive.)

a. Diagram the cross described in this part of the problem:

nondimpled ♂ × dimpled ♀ → proportion of dimpled F_1?

Note that the dimpled woman in this cross had a *dd* (nondimpled) mother, so the dimpled woman MUST be heterozygous. We can thus rediagram this cross with genotypes:

dd (nondimpled) ♂ × *Dd* (dimpled) ♀ → 1/2 *Dd* (dimpled) : 1/2 *dd* (nondimpled)

One half of the children produced by this couple would be dimpled.

b. Diagram the cross:

dimpled (*D?*) ♂ × nondimpled (*dd*) ♀ → nondimpled F_1 (*dd*)

Because they have a nondimpled child (*dd*), the man must have a *d* allele to contribute to the offspring. **The man is thus of genotype *Dd*.**

c. Diagram the cross:

dimpled (*D?*) ♂ × nondimpled (*dd*) ♀ → 8 F_1, all dimpled (*D*–)

The *D* allele in the children must come from their father. The father could be either *DD* or *Dd*, but **it is most probable that the father's genotype is *DD*.** We cannot rule out completely that the father is a *Dd* heterozygote. However, if this was the case, the probability that all 8 children would inherit the *D* allele from a *Dd* parent is only $(1/2)^8 = 1/256$.

11. a. The 3:1 ratio in the progeny of cross 1 × 4 suggests that a single gene controls flowering time, and that late (*F*) is dominant to early (*f*).

b. **Plants 1 and 4 are *Ff*** because their progeny exhibit a 3 late (*F*–) :1 early (*ff*) ratio. **Plant 2 is *ff*** because when crossed with plant 1 (*Ff* × *ff*), the progeny are in a 1:1 ratio of late (*Ff*) to early (*ff*). **Plant 3 is *FF*** because when crossed with either 1, 3, or 4, all the progeny are late (*F*–).

c. **Selfing of Plant 1 or Plant 4 (*Ff*) would produce progeny in a phenotypic ratio of 3 late : 1 early (3 *F*– : 1 *ff*). Selfing of Plant 2 (*ff*) would produce all early (*ff*) progeny, and selfing of Plant 3 would result in progeny that are all late (*FF*).**

12. a. You need to realize that the results tabulated are unlike those of the plants in the previous problem in an important way: The parents are not two individuals who had 30–50 progeny. Instead, the entries in the table show the progeny of many pairs of parents with the same characteristics, but who could have different genotypes. This means that if sticky and dry are controlled by alternate alleles of a single gene, in any row of the table involving parents with the dominant phenotype, those parents could actually be a mixture of homozygous dominant and heterozygous individuals.

Thus, in a row where both parents have the dominant phenotype, some of the crosses could be *Ss* × *Ss,* while in a row where one parent has the dominant phenotype and the other parent has the recessive phenotype, some of the crosses could be *Ss* × *ss*. Both cases could produce some progeny who will be homozygous recessive (*ss*). Only crosses between two homozygous recessive individuals will result in progeny that all have the recessive phenotype. The only crosses that fit this criterion are in the row: dry × dry → all dry. Therefore, **dry is the recessive phenotype (*ss*) and sticky is the dominant phenotype (*S*–).**

b. **3:1 or 1:1 ratios are not observed in the table because some sticky individuals in each of the first two rows are *SS* while others are *Ss*.**

In the first row of the table, the sticky × sticky matings in this human population are not simply crosses between heterozygotes, but instead a mix of three different kinds of matings: between two heterozygotes (*Ss* × *Ss*), between two homozygotes (*SS* × *SS*), and between a homozygote and heterozygote (*SS* × *Ss*). The progeny from the two latter crosses obscure the 3:1 ratio that would result from crosses between heterozygotes.

In the second row of the table, the sticky × dry matings include both *Ss* × *ss* and *SS* × *ss*. The progeny of the latter crosses obscure the 1:1 ratio that would result from any *Ss* × *ss* testcrosses in the same row.

13. Diagram the cross:

black × red → 1 black : 1 red

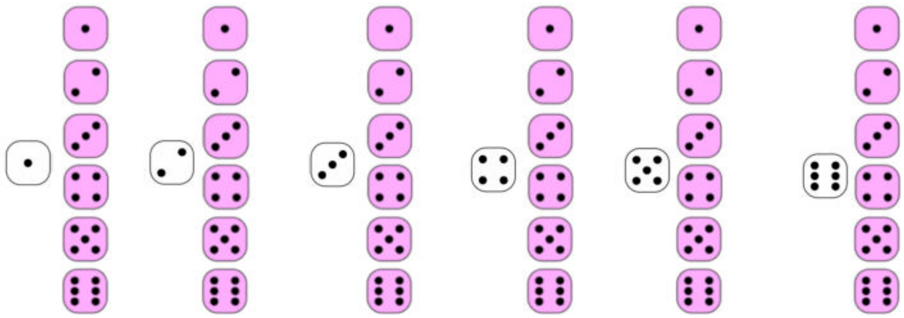
No, you cannot tell how coat color is inherited from the results of this one mating. The 1:1 ratio indicates that this was a testcross—a cross between a heterozygote and a homozygous recessive. However, we cannot know from the testcross whether red or black is the dominant phenotype. To determine which phenotype is dominant, remember that an animal with a recessive phenotype must be homozygous. Thus, **if you mate several red horses to each other and also mate several black horses to each other, the crosses that always yield only offspring with the parental phenotype must have been between homozygous recessives.** For example, if all the black × black matings result in only black offspring, black is recessive. Some of the red × red crosses (that is, crosses between heterozygotes) would then result in both red and black offspring in a ratio of 3:1. To establish this point, you might have to do several red × red crosses, because some of these crosses could be between red horses homozygous for the dominant allele. You could of course ensure that you were sampling heterozygotes by using the progeny of black × red crosses (such as that described in the problem) for subsequent [black × black] or [red × red] crosses.

14. **a.** **1/6** because a die has 6 different sides.

b. Three even numbers exist (2, 4, and 6). The probability of obtaining any one of these is 1/6. Because the 3 events are mutually exclusive, use the sum rule: 1/6 + 1/6 + 1/6 = 3/6 = **1/2.**

c. You must roll either a 3 or a 6, so 1/6 + 1/6 = 2/6 = **1/3.**

When thinking about probabilities involving 2 dice (in the diagram that follows, a *white* one and a *pink* one), it helps to realize that: (1) The probabilities calculated for rolling both dice simultaneously will be the same as those calculated for rolling them in succession (first one, then the other), and (2) as shown in the diagram that follows, 36 different outcomes are possible:



d. Each die is independent of the other, thus the product rule is used: 1/6 × 1/6 = **1/36.** (Two 6s is one of the 36 possible outcomes.)

e. The probability of getting an even number on one die is 3/6 = 1/2 [see part (b)]. This is also the probability of getting an odd number on the second die. This result could happen either of 2 ways—you could get the odd number first and the even number second, or vice versa, and these are mutually exclusive possibilities. Thus, the probability of both occurring is (1/2 × 1/2) + (1/2 × 1/2) = 1/4 + 1/4 = **1/2.**

Another way of thinking about this problem is that the first die can land any way (probability = 1), but the second die must show an odd number (probability = 1/2) if the first die was even, and an even number (probability = 1/2) if the first die was odd. Therefore, no matter how the first die lands, the second die has a 1/2 chance of landing on a number that satisfies the stated criterion, and so the probability that the criterion is satisfied is 1 × 1/2 = 1/2. (You can see in the diagram above that 18 of the 36 possible outcomes satisfy the criterion that one die has an odd number and the other die has an even number.)

f. The probability of any specific number on a die = 1/6. The probability of the same number on the other die = 1/6. The probability of both occurring at the same time is 1/6 × 1/6 = 1/36. The same probability is true for the other 5 possible numbers on the dice. Thus, the probability of any of these mutually exclusive situations occurring is 1/36 + 1/36 + 1/36 + 1/36 + 1/36 + 1/36 = 6/36 = **1/6.**

Another way of thinking of about this problem is that the first die can land any way at all (probability = 1), but the second die must match the first one (probability = 1/6). The chance of both events happening is 1 × 1/6 = 1/6. (In other words, 6 of the 36 possible outcomes satisfy the criterion that both numbers are the same.)

g. The probability of getting two numbers both over four is the probability of getting a 5 or 6 on one die (1/6 + 1/6 = 1/3) and a 5 or 6 on the other die (1/3). The results for the two dice are independent events, so 1/3 × 1/3 = **1/9.** (In other words, 4 of the 36 possible outcomes satisfy the criterion both numbers are over 4.)

15. a. The probability of drawing a face card = 0.23 (= 12 face cards / 52 cards). **The probability of drawing a red card = 0.5** (= 26 red cards / 52 cards). **The probability of drawing a red face card = probability of a red card × probability of a face card = 0.23 × 0.5 = 0.12.** (Alternatively, 6 red face cards / 52 cards = 0.12.)