3.1 Introduction

Pedigrees are used in human genetics (and in some other animals, particularly cattle, horses, dogs, and laboratory animals) to help genetic researchers organize data. They were first developed to help discover whether some physical characteristic is genetic. They are a graphical representation of the relationships among individuals and their phenotypes. Figure 3-1 is a simple pedigree that demonstrates many of the symbols used.

![Sample Pedigree](image)

Figure 3-1  Sample Pedigree
We will use pedigrees in two ways

- Pedigrees can be used to organize and follow the inheritance of genetic characteristics. In some cases, we can use the pedigree to determine the genotype of all individuals. If the characteristic is recessive, it is not possible to determine the genotype for an individual who does not have the characteristic. They will be either AA or Aa.

- Pedigrees can be used to look at possible modes of inheritance. We will use this in Chapter 5 to distinguish among various types of inheritance.

### 3.2 Determining Probabilities in Pedigrees

Imagine we are genetic counselors, and have been asked to determine the risk that a couple will have a child with a particular characteristic. The characteristic is autosomal recessive. We have tested family members and have determined the pedigree shown (Figure 3-2).

![Figure 3-2 Pedigree for Simple Mendelian Trait](image)

The couple who have come to us is II-2 and II-3. The woman has a father and a brother with the characteristic. The man has a brother with the characteristic. A little bit of thought relates the genotypes for most of the members of this family. Since the characteristic is recessive and I-3, II-1, and II-4 have the characteristic, they must have the genotype aa. Because II-1 is aa, and received his gametes from his parents, both parents must be either Aa or aa. Since neither shows the characteristic, they must both have the genotype Aa. Similarly, since II-4 is aa, then I-4 also must be Aa.
Finally, consider the couple. The female received an $a$-gamete from her father, and does not show the characteristic, so she must have genotype $Aa$. The male is the result of the cross $Aa \times Aa$. We know he cannot be $aa$, since he does not show the characteristic. However, it is possible for him to be either $AA$ or $Aa$. There is no way for us to determine which genotype he has, but we do have knowledge of the probabilities.

Consider the cross $Aa \times Aa$ (the man’s parents). The Punnett Square gives us the following frequencies:

<table>
<thead>
<tr>
<th></th>
<th>$1/4 \ AA$</th>
<th>$1/2 \ Aa$</th>
<th>$1/4 \ aa$</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>3/4 Normal</td>
<td>1/4 Effected</td>
<td></td>
</tr>
</tbody>
</table>

Since he is not effected, we know he must be either $AA$ or $Aa$, but we cannot tell which genotype he has. Notice that there is twice as much chance he is $Aa$ than $AA$. If we restrict ourselves to the $3/4$ of the offspring who are normal, we see that $2/3$ of them are $Aa$. In a similar way, $1/3$ of the normal offspring are $AA$. Arithmetically, this can be written as:

$$\frac{1}{4} AA + \frac{1}{2} \ Aa = \frac{1}{3} AA + \frac{2}{3} Aa.$$ 

It is common to write these probabilities in the form of an equation. This equation states that there is a $1/3$ chance of being $AA$ and a $2/3$ chance of being $Aa$. It is important to notice that the fractions add to 1.

**Rule 1:** When you eliminate genotypes, the probabilities of the remaining genotypes are adjusted by dividing by the total remaining.

We now have probability statements about the parents. At this stage, the pedigree looks like Figure 3-3.

We can write the cross that produces III-1 as:

$$\left( \frac{1}{3} AA + \frac{2}{3} Aa \right) \times Aa.$$ 

We are interested in their offspring (although in this case, it is their prospective offspring). For this, we will create a Punnett Square with the paternal gametes on one side and the maternal gametes on the other. The gametes from the female are straight forward, she
produces gametes of types A and a, in equal frequencies. This is
given on the left part of the Punnett Square.

**Father**

For the father, the only way he can pass on a gametes is if he is a
carrier, Aa. The chance he is a carrier is \( \frac{2}{3} \). If he is a carrier, he has a
\( \frac{1}{2} \) chance of producing the a gamete. So, overall, the chance he will
produce the a gamete is \( \frac{2}{3} \cdot \frac{1}{2} = \frac{1}{3} \). This is given on the horizontal
part of the Punnett Square. For the A gamete, there are two ways to
calculate this. The first is to notice that the father must produce a
gamete of some sort. Since the chance for the a gamete is \( \frac{1}{3} \), the
chance for the A gamete is \( 1 - \frac{1}{3} = \frac{2}{3} \). The other way is similar to the
method for calculating the a gamete, except there are two ways to
get A: From the AA genotype (with chance 1); and From the Aa

**Diagram**

![Pedigree with Genotypes for Most Individuals](image_url)
genotype (with chance $\frac{1}{2}$). Combining these two cases, the chances of producing the A gamete is
$$\frac{1}{3}(1) + \frac{2}{3}(1) = \frac{2}{3}.
$$

**Rule 2:** When computing the probability of a particular gamete, consider all the possible genotypes that can possibly pass on that allele. For each possibility, multiply the chance of that genotype by the chance of the given allele from that genotype. The second number is always either a 1 or a 1/2. The first number changes with the pedigree.

In the end, the chance that the couple will have a child with the characteristic is calculated from the Punnett Square as $\frac{1}{6}$.

### 3.3 Some Pedigree Problems

These problems will help you to practice the techniques given in this chapter. It is the nature of many of these problems to have very unusual probabilities. Don’t panic, just follow the basic procedure given in Section 3.2.

1. Consider the pedigree given in Figure 3-4 for an Autosomal Recessive trait. Assume individuals entering the pedigree are homozygous for the dominant trait, unless there is evidence to the contrary. What is the probability the indicated individual (the person with ?) will have the characteristic under study?

2. Another pedigree for an autosomal recessive trait is given in Figure 3-5. What is the probability the indicated individual will have the characteristic under study? Assume individuals entering the pedigree do not have the recessive allele unless there is evidence to the contrary.

3. A large pedigree is given in Figure 3-6. Assume individuals entering the pedigree do not have the recessive allele unless there is evidence to the contrary. Give the probability that the indicated individual in Generation V will have the characteristic.
Figure 3-4  Typical Pedigree Showing Double First Cousin Mating

Figure 3-5  Another Pedigree for Computing Probabilities
Figure 3-6  Large Pedigree for Probability Homework