

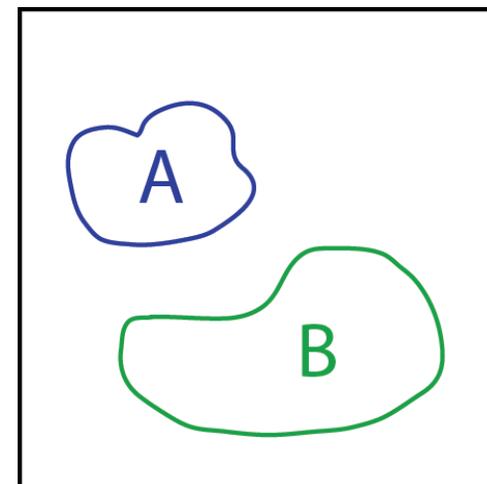
# Probability

## Chapter 5

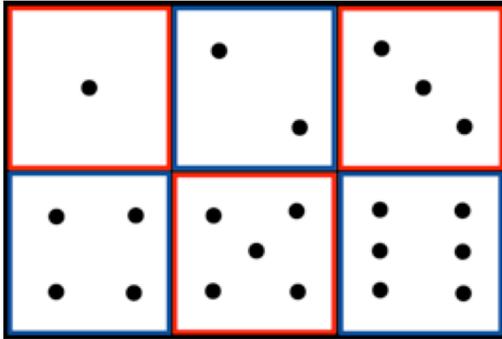
The probability of an event is its true relative frequency, the proportion of times the event would occur if we repeated the same process over and over again.

Two events are *mutually exclusive* if they cannot both be true.

A and B are mutually exclusive



Mutually exclusive



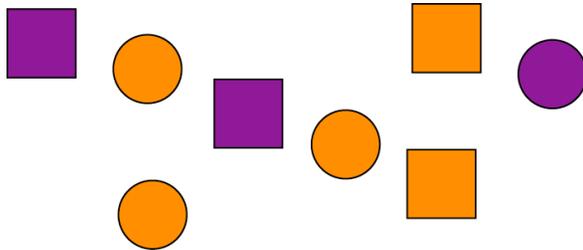
Mutually exclusive

$$\Pr(A \text{ and } B) = 0$$

Not mutually exclusive

$$\Pr(A \text{ and } B) \neq 0$$

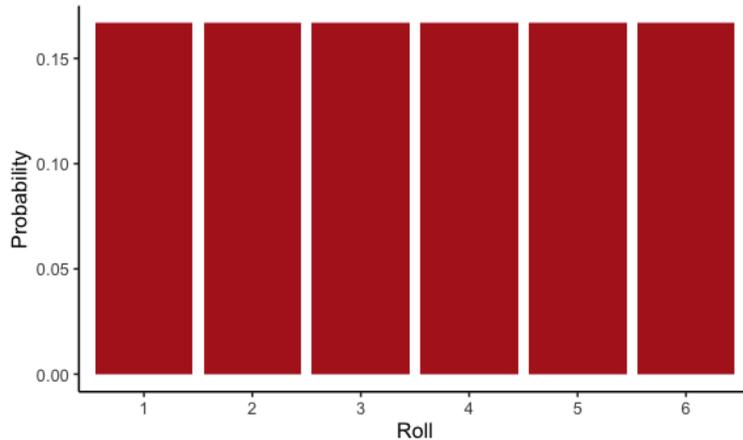
$$\Pr(\text{purple AND square}) \neq 0$$



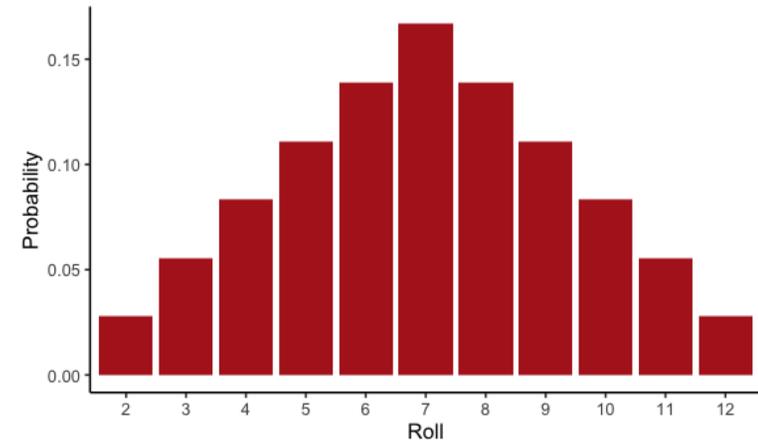
Probability distribution

A probability distribution describes the true relative frequency of all possible values of a random variable.

## Probability distribution for the outcome of a roll of a die



## Probability distribution for the sum of a roll of two dice



## The addition principle

The addition principle: If two events A and B are mutually exclusive, then

$$\Pr[A \text{ OR } B] = \Pr[A] + \Pr[B]$$

## The probability of a range

$$\Pr[\text{Number of green M\&Ms} \geq 6] = \Pr[6 \text{ green}] + \Pr[7 \text{ green}] + \Pr[8 \text{ green}] + \dots$$



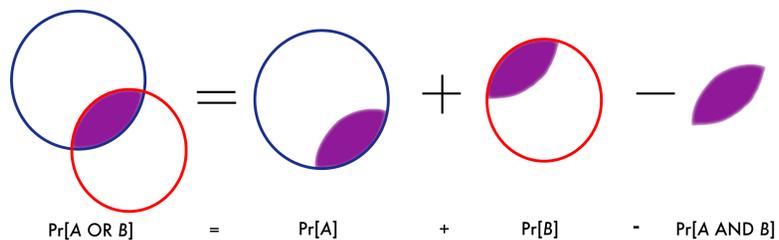
## Probability of *Not*

The probabilities of all possibilities add to 1.

$$\Pr[\text{NOT rolling a 2}] = 1 - \Pr[\text{Rolling a 2}] = 5/6$$

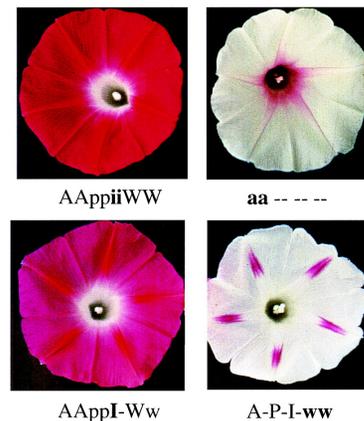


## General Addition Principle



## General addition principle

$$\Pr[A \text{ OR } B] = \Pr[A] + \Pr[B] - \Pr[A \text{ AND } B].$$



e.g., Morning glory flowers can be white because of their *ww* genotype at the *W* locus and/or because of their *aa* genotype at the *A* locus

## General addition principle

$$\Pr[A \text{ OR } B] = \Pr[A] + \Pr[B] - \Pr[A \text{ AND } B].$$

e.g., Morning glory flowers can be white because of their  $ww$  genotype at the  $W$  locus and/or because of their  $aa$  genotype at the  $A$  locus

If:

$$\begin{aligned} \Pr[ww] &= 0.1 & \Pr[aa] &= 0.2 \\ \Pr[ww \text{ AND } aa] &= 0.04 \end{aligned}$$

$$\begin{aligned} \Pr[\text{white flowers}] &= \Pr[ww] + \Pr[aa] - \Pr[ww \text{ AND } aa] \\ &= 0.1 + 0.2 - 0.04 \\ &= 0.26 \end{aligned}$$

## Multiplication principle

The multiplication principle: If two events  $A$  and  $B$  are independent, then:

$$\Pr[A \text{ AND } B] = \Pr[A] \times \Pr[B]$$

## Independence

Two events are independent if the occurrence of one gives no information about whether the second will occur.

### Short-haired, black cats

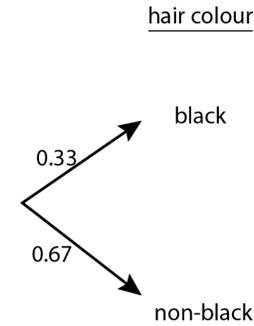
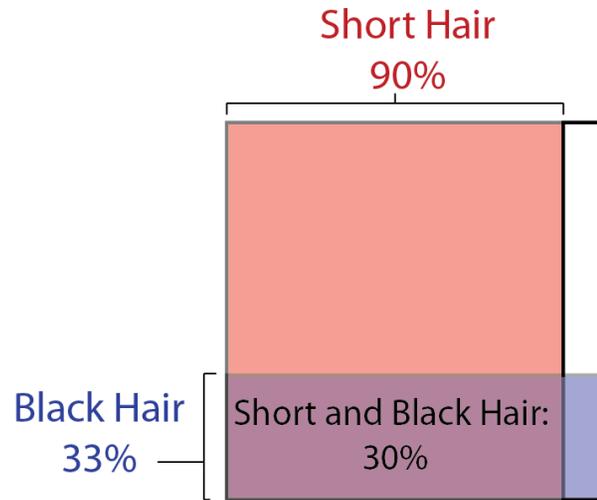
What is the probability that a cat is black- and short-haired?

$$\Pr[\text{black hair}] = 0.33 \quad \Pr[\text{short-haired}] = 0.90$$

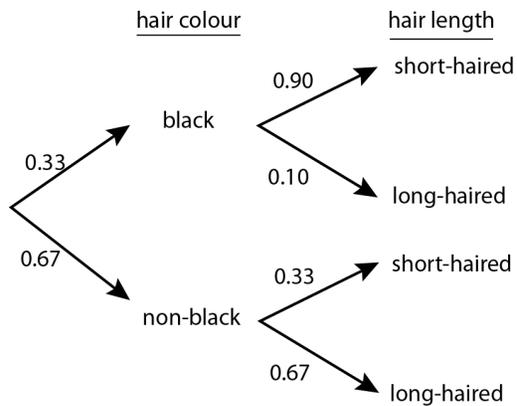
$$\begin{aligned} \Pr[\text{black hair AND short hair}] &= 0.33 \times 0.90 \\ &= 0.30. \end{aligned}$$



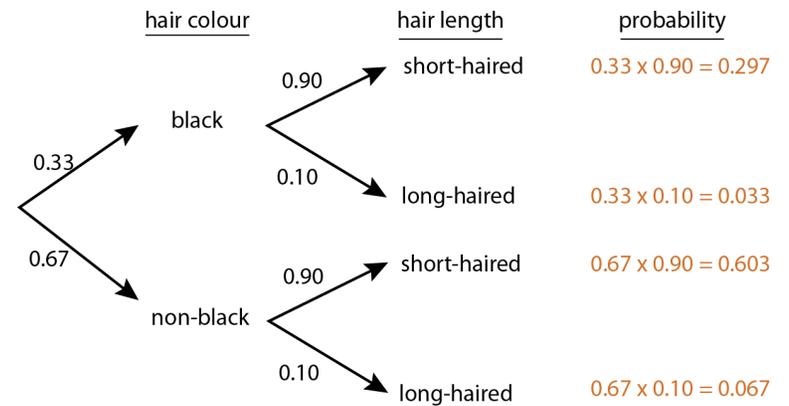
## Probability trees



## Phenotypes in two-child family



## Phenotypes in two-child family



## Short summary

The probability of A OR B involves addition.

$\Pr(A \text{ or } B) = \Pr(A) + \Pr(B)$  if the two are mutually exclusive.

The probability of A AND B involves multiplication.

$\Pr(A \text{ and } B) = \Pr(A) \Pr(B)$  if the two are independent

## Triple test and detection of trisomy 23

The triple test detects Down syndrome when it is present 60% of the time.

This means it has a 40% false negative rate of 40%.

The triple test gives a false positive 5% of the time (when the fetus does not have trisomy 23).

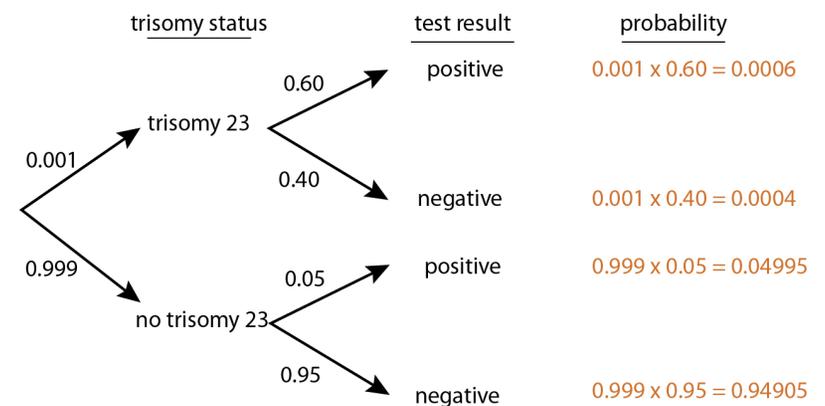
The probability that a fetus has trisomy 23 is 0.001.

## Dependent events

Variables are not always independent.

The probability of one event may depend on the outcome of another event.

## Triple test outcomes



Are trisomy status and test results independent?

$$\Pr(\text{trisomy}) = 0.001$$

$$\Pr(\text{positive test result}) = 0.006 + 0.04995 = 0.05595$$

$$\begin{aligned} \Pr(\text{trisomy AND positive result}) &= 0.006 \neq \\ &\Pr(\text{trisomy}) \times \Pr(\text{positive result}) = \\ &0.001 \times 0.05595 = 0.000056 \end{aligned}$$

So these two events are NOT independent.

$\Pr(X | Y)$  means the probability of  $X$  if  $Y$  is true.

It is read as "the probability of  $X$  given  $Y$ ."

$$\Pr(\text{positive test} | \text{trisomy}) = 0.60.$$

## Conditional probability

The conditional probability of an event is the probability of that event occurring *given that* a condition is met.

$$\Pr[X | Y]$$

Law of total probability:

$$\Pr[X] = \sum_{\text{All values of } Y} \Pr[X|Y] \Pr[Y]$$

## The general multiplication rule

The probability of a positive test result is

$$\begin{aligned}\Pr[\text{positive result}] &= \\ &\Pr(\text{positive result} \mid \text{trisomy}) \Pr(\text{trisomy}) + \\ &\Pr(\text{positive result} \mid \text{no trisomy}) \Pr(\text{no trisomy}) \\ &= 0.60 (0.001) + 0.05 (0.999) = 0.05055\end{aligned}$$

$$\Pr[A \text{ AND } B] = \Pr[A] \Pr[B \mid A].$$

## The general multiplication rule

$$\Pr[A \text{ AND } B] = \Pr[A] \Pr[B \mid A].$$

$$\Pr[A \text{ AND } B] = \Pr[B] \Pr[A \mid B].$$

Therefore

$$\Pr[A] \Pr[B \mid A] = \Pr[B] \Pr[A \mid B].$$

## Bayes' theorem

$$\Pr[A \mid B] = \frac{\Pr[B \mid A] \Pr[A]}{\Pr[B]}$$

# Applying Bayes' theorem

For the triple test, what is the probability that a pregnancy with a positive result is affected by trisomy 23?

In other words:

$$\Pr[\text{trisomy 23} \mid \text{positive result}] = ?$$

$$\Pr[\text{trisomy 23} \mid \text{positive result}] =$$

$$\frac{\Pr[\text{positive result} \mid \text{trisomy 23}] \Pr[\text{trisomy 23}]}{\Pr[\text{positive result}]}$$

We already know:

$$\Pr[\text{positive result} \mid \text{trisomy 23}] = 0.60$$

$$\Pr[\text{trisomy 23}] = 0.0001$$

$$\Pr[\text{trisomy 23} \mid \text{positive result}] =$$

$$\frac{\Pr[\text{positive result} \mid \text{trisomy 23}] \Pr[\text{trisomy 23}]}{\Pr[\text{positive result}]}$$

$$\Pr[\text{trisomy 23} \mid \text{positive result}] =$$

$$\frac{\Pr[\text{positive result} \mid \text{trisomy 23}] \Pr[\text{trisomy 23}]}{\Pr[\text{positive result}]}$$

We need to know:

$$\Pr[\text{positive result}] = ?$$

$$\Pr[\textit{trisomy 23} \mid \textit{positive result}] =$$

$$\frac{\Pr[\textit{positive result} \mid \textit{trisomy 23}] \Pr[\textit{trisomy23}]}{\Pr[\textit{positive result}]}$$

We need to know: (Law of total probability)

$$\begin{aligned} \Pr[\textit{positive result}] &= \\ \Pr[\textit{positive result} \mid \textit{trisomy 23}] \Pr[\textit{trisomy23}] &+ \Pr[\textit{positive result} \mid \textit{no trisomy}] \Pr[\textit{no trisomy}] \\ &= 0.60(0.001) + 0.05(0.999) \\ &= 0.05595 \end{aligned}$$

$$\Pr[\textit{trisomy 23} \mid \textit{positive result}] =$$

$$\frac{\Pr[\textit{positive result} \mid \textit{trisomy 23}] \Pr[\textit{trisomy23}]}{\Pr[\textit{positive result}]}$$

$$= \frac{0.60 \times 0.001}{0.05595}$$

$$= 0.011$$