## Estimation

## Session 4

Module 1 Probability \& Statistical Inference

The Summer Institutes
DEPARTMENT OF BIOSTATISTICS SCHOOL OF PUBLIC HEALTH
University of WashingTon

## Overview

Probability/statistical models depend on parameters
Binomial depends on probability of success $\pi$.
Normal depends on mean $\mu$, standard deviation $\sigma$.
Parameters are properties of the "population" and are typically unknown.

The process of taking a sample of data to make inferences about these parameters is referred to as estimation.

There are a number of different estimation methods
.. we will study two estimation methods:

## 1. Maximum likelihood (ML)

## 2. Bayes

## UNIVERSITY of WASHINGTON

Fisher (1922) invented this general method.
Problem Unknown model parameters $\theta$
Set-up Write the probability of the data $\mathbf{Y}$ in terms of the model parameter: $\mathrm{P}(\mathbf{Y} \mid \theta)$

Solution Estimate $\theta$ as the value that makes the data $\mathbf{Y}$ look most likely to occur. This estimate is denoted by $\hat{\theta}$.

## Maximum Likelihood Estimate

Suppose a man is known to have transmitted allele A1 to his child at a locus that has only two alleles: A1 and A2.
What is the maximum likelihood estimate of the man's genotype?
Solution Let X represent the data (paternal allele in the child) and let $\theta$ represent the parameter (man's genotype):

$$
\begin{aligned}
& X=A 1 \\
& \theta=\{A 1 A 1, A 1 A 2, A 2 A 2\}
\end{aligned}
$$

The probability function is based on $P(X \mid \theta)$....

$$
\begin{aligned}
& P(X=A 1 \mid \theta=A 1 A 1)=1 \\
& P(X=A 1 \mid \theta=A 1 A 2)=0.5 \\
& P(X=A 1 \mid \theta=A 2 A 2)=0
\end{aligned}
$$

Therefore, the MLE is $\hat{\theta}=\mathrm{A} 1 \mathrm{~A} 1$

## Maximum Likelihood

Suppose we have a sample of 20 gametes in which the number of recombinants (Z) and nonrecombinants ( $\mathrm{N}-\mathrm{Z}$ ) for two loci can be counted. Use these data to estimate the recombination fraction ( $\pi$ ) between the two loci.

Solution The probability of the data can be modeled using a binomial distribution. The probability distribution function is:

$$
\begin{gathered}
P(Z \mid \pi)=\left(\begin{array}{c}
20 \\
Z \\
\text { e and } \pi \text { is fixed. }
\end{array}\right) \pi^{Z}(1-\pi)^{20-Z}
\end{gathered}
$$

where $Z$ is the variable and $\pi$ is fixed.
The likelihood function is the same function:

$$
L(\pi \mid Z)=\left(\begin{array}{l}
20 \\
\end{array}\right) \pi^{Z}(1-\pi)^{20-Z}
$$

except now $\pi$ is the variable and $\mathbf{Z}$ is fixed.

## Maximum Likelihood

- We can use calculus to find the maximum of the (log) likelihood function.

$$
\begin{aligned}
\frac{d \ln L}{d \pi} & =0 \\
\frac{d}{d \pi}[Z \ln \pi+(20-Z) \ln (1-\pi)] & =0 \\
\frac{Z}{\pi}-\frac{20-Z}{1-\pi} & =0 \\
\widehat{\pi} & =\frac{Z}{20}
\end{aligned}
$$

- Not surprisingly, the likelihood in this example is maximized at the observed proportion, 3/20.
- Sometimes the MLE has a simple closed form. In more complex problems, numerical optimization is used.
- Computers can find these maximum values!


## Maximum Likelihood <br> General Notation

$L(\theta)=$ likelihood as a function of the parameter $\theta$
$\ell(\theta)=\ln (L(\theta))$, the log likelihood
> Usually more convenient to work with analytically and numerically
$S(\theta)=d e(\theta) / d \theta$, the score
> Set to zero and solve for $\theta$ to calculate the MLE
$I(\theta)=-d^{2} \ell(\theta) / d \theta^{2}$, the information
$>$ Inverse gives variance of $\hat{\theta}$
$\operatorname{Var}(\hat{\theta})=\mathrm{E}[\mathrm{I}(\theta)]^{-1}$ (in most cases)

## Bayes Estimation

Recall Bayes theorem:
(written in terms of data X and parameter $\theta$ )

$$
P(\theta \mid X)=\frac{P(X \mid \theta) P(\theta)}{\int_{\theta} P(X \mid \theta) P(\theta)}
$$

Notice the change in perspective - $\boldsymbol{\theta}$ is now treated as a random variable instead of a fixed number.
$>P(X \mid \theta)$ is the likelihood function, as before.
$>P(\theta)$ is called the prior distribution of $\theta$.
> $P(\theta \mid X)$ is called the posterior distribution of $\theta$ and is used for estimation
Based on $P(\theta \mid X)$ we can define a number of possible estimators of $\theta$. $A$ commonly used estimate is the maximum a posteriori (MAP) estimate:

$$
\hat{\theta}_{M A P}=\max _{\theta} P(\theta \mid X)
$$

We can also use $P(\theta \mid X)$ to define "credible" intervals for $\theta$.

## Bayes Estimation

## Comments

> The Bayesian procedure provides a convenient way of combining external information or previous data (through the prior distribution) with the current data (through the likelihood) to create a new estimate.
> As N increases, the data (through the likelihood) overwhelms the prior and the Bayes estimator typically converges to the MLE.
> Controversy arises when $\mathrm{P}(\theta)$ is used to incorporate subjective beliefs or opinions.
> If the prior distribution $P(\theta)$ is simply that $\theta$ is uniformly distributed over all possible values, this is called an uninformative prior, and the MAP is the same as the MLE.

## Bayes Estimation

Suppose a man is known to have transmitted allele A1 to his child at a locus that has only two alleles: A1 and A2.
What is the maximum likelihood estimate of the man's genotype?

Solution Let $X$ represent the data (paternal allele in the child) and let $\theta$ represent the parameter (man's genotype):

$$
\begin{aligned}
& X=A 1 \\
& \theta=\{A 1 A 1, A 1 A 2, A 2 A 2\}
\end{aligned}
$$

The probability function is based on $P(X \mid \theta)$....

$$
\begin{aligned}
& P(X=A 1 \mid \theta=A 1 A 1)=1 \\
& P(X=A 1 \mid \theta=A 1 A 2)=0.5 \\
& P(X=A 1 \mid \theta=A 2 A 2)=0
\end{aligned}
$$

Therefore, the MLE is $\hat{\theta}=\mathrm{A} 1 \mathrm{~A} 1$

## 

Suppose that we know that the frequency of the A1 allele in the general population is only 1\%. Assuming Hardy-Weinberg Equilibrium we have

$$
\begin{aligned}
& P(\theta=A 1 A 1)=0.01 * 0.01=0.0001 \\
& P(\theta=A 1 A 2 \text { or } A 2 A 1)=2 * 0.01 *(1-0.01)=0.0198 \\
& P(\theta=A 2 A 2)=(1-0.01) *(1-0.01)=0.9801
\end{aligned}
$$

Also

$$
P(X)=\sum_{\theta} P(X=A 1 \mid \theta) P(\theta)=0.01
$$

This leads to the posterior distribution

$$
\begin{aligned}
& P(\theta=A 1 A 1 \mid X=A 1)=0.01 \\
& P(\theta=A 1 A 2 \mid X=A 1)=0.99 \\
& P(\theta=A 2 A 2 \mid X=A 1)=0
\end{aligned}
$$

Therefore the Bayesian MAP estimator is

$$
\theta=\mathrm{A} 1 \mathrm{~A} 2
$$

## Summary

Maximum likelihood is a method of estimating parameters from data
> ML requires you to write a probability model for the data
> MLEs may be found analytically or numerically
> (Inverse of the negative of the) second derivative of the log-likelihood gives variance of estimates

Bayesian procedures allow us to incorporate additional information about the parameters in the form of prior data, external information, or personal beliefs.

## Break \#1

Pause the video, take a break, stretch, then review relevant exercises from worksheet.

Afterwards, continue on!


