

Lecture 4: Parameter Estimation

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Review: Probability Distributions

Discrete:

- Binomial distribution
- Hypergeometric distribution
- Poisson distribution

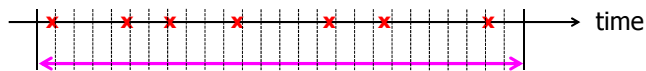
Continuous:

- Uniform distribution
- Exponential distribution
- Gamma distribution
- Normal distribution

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Relationship among Binomial, Poisson, and Exponential Distributions

- Let's revisit the e-mail example
 - Assume that e-mails come at a constant rate



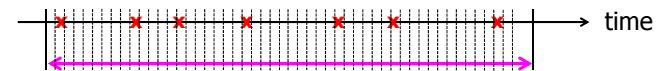
- What is the probability of getting k e-mails in a certain time interval?
 - Let's discretize the time interval into subintervals
 - In each subinterval, there are only two possible outcomes – getting an e-mail with prob p or not getting an e-mail with prob $(1-p)$
 - We can calculate the probability based on **Binomial distribution**

$$P\{X = k\} = \binom{n}{k} p^k (1-p)^{n-k}$$

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Relationship among Binomial, Poisson, and Exponential Distributions

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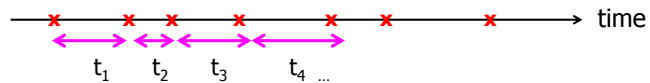
- Let's now make the sub-interval as small as possible
 - We are essentially doing $n \rightarrow \infty$ and $p \rightarrow 0$
 - Then, the Poisson distribution becomes equivalent to Binomial distribution

$$P\{X = k\} = \frac{1}{k!} \lambda^k e^{-\lambda}$$

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Relationship among Binomial, Poisson, and Exponential Distributions

- Let's revisit the e-mail example
 - Assume that e-mails come at a constant rate



- Intervals between adjacent events follow the exponential distribution

$$P(x) = \begin{cases} \lambda e^{-\lambda x}, & x \geq 0 \\ 0, & x < 0 \end{cases}$$

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Goals

- Basic concepts on parameter estimation
- Maximum likelihood estimation (MLE)
- Bayesian inference
- Confidence interval

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What Is Parameter?

- Variables vs. Parameters*
- According to Bard & Yonathan (1974)*
 - ... Usually a probabilistic model is designed to explain the relationships that exist among quantities which can be measured independently in an experiment; these are the **variables** of the model. To formulate these relationships, however, one frequently introduces "constants" which stand for inherent properties of nature. These are the **parameters**.
- We often denote by θ

* Bard, Yonathan (1974). *Nonlinear Parameter Estimation*. New York

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Examples

- Binomial distribution (coin tossing)
 - X: number of Heads after n coin tosses



$$P\{X = k\} = \binom{n}{k} p^k (1-p)^{n-k}$$

variable parameter $\theta = p$

- Poisson distribution
 - X: number of e-mails within a week

$$P\{X = k\} = e^{-\lambda} \frac{\lambda^k}{k!}$$

variable parameter $\theta = \lambda$

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Parameter Estimation

- **Estimator:** Statistic whose calculated value is used to estimate a parameter, θ
- **Estimate:** A particular realization of an estimator, θ
- Types of estimators:
 - **Point estimate:** single number that can be regarded as the most plausible value of θ
 - **Interval estimate:** a range of numbers, called a confidence interval indicating, can be regarded as likely containing the true value of θ

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Methods of Point Estimation

- Maximum likelihood estimation (MLE)
- Bayesian inference

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Properties of Good Estimators

- What are characteristics of good estimators?
- How well they *explain* the world?
- Say that you flip a coin
 - Let's say that a rv. X represents the outcome
 - p = probability of getting Head
- If you flip a coin many times, maybe we can figure out.
 - Realization of the random variable
 - Observation data $D = \{HHTHHTHTHTHTH \dots\}$



← samples (or instances) →

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Introduction to Likelihood

- **Before** an experiment is performed, the outcome is unknown. Probability function allows us to **predict the probability of any outcome based on known parameters:**

$$P(\text{Data} \mid \theta)$$

- For example, say that we know that probability of getting a Head in a coin toss is $p = 0.6$

- Then, we can calculate the probability $P(\text{Data} \mid \theta)$ for ANY data

$$D_1 = \{HTHHHTHHHT\} \quad P(D \mid \theta) = p^7(1-p)^3$$

$$D_2 = \{HTH\} \quad P(D \mid \theta) = p^2(1-p)$$

$$D_3 = \{TTTH\} \quad P(D \mid \theta) = p^3(1-p)$$

⋮

- If p were a different value, the above probabilities would have been different...

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Introduction to Likelihood

- **After** an experiment is performed, the outcome is known. Now we talk about the **likelihood that a parameter would generate the observed data**:

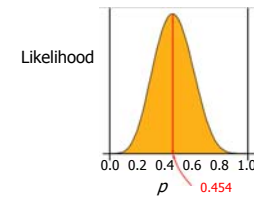
$$L(\theta: D) = P(\text{Data} \mid \theta)$$

- Estimation proceeds by finding the value of θ that makes the observed data most *likely*
 - **Maximum Likelihood Estimate (MLE)** $\hat{\theta}$

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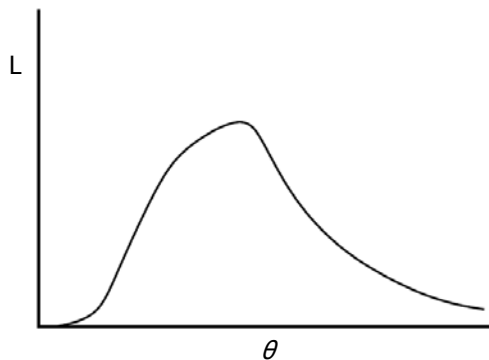
The Coin Example

- Let's toss a coin n times with probability p of heads
- Probability of outcome $D = \{\text{HHTHTTTTHTTH}\}$ is
$$pp(1-p)p(1-p)(1-p)(1-p)p(1-p)(1-p)p$$
- The likelihood is then $L = P(D \mid p) = p^5(1-p)^6$
- Plotting L against p to find its maximum



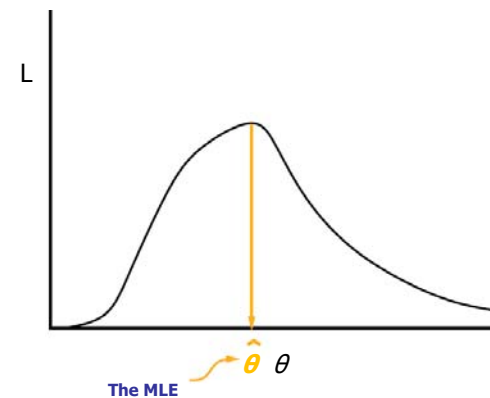
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A Likelihood Curve



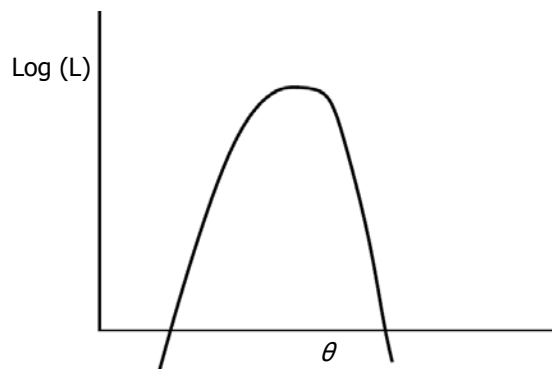
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Its Maximum Likelihood Estimate



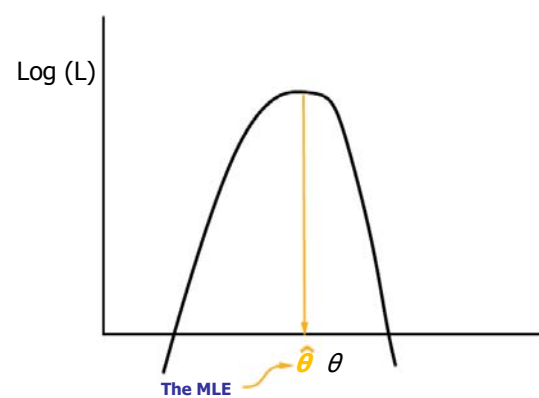
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Better to Plot log (L) than L



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Better to Plot log (L) than L



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Differentiating to Find the Maximum

- Differentiate the expression for log (L) with respect to p

$$\log L = \log[p^5(1-p)^6] = \frac{5}{p} + \frac{6}{1-p}$$

- Equate the derivative to 0

$$\frac{\partial \log L}{\partial p} = \left(\frac{5}{p} - \frac{6}{1-p} \right) = 0$$

$$5 - 11p = 0 \quad \Rightarrow \quad \hat{p} = \frac{5}{11}$$

- The value of p that is at the peak can be found to be $p = 5/11$

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Formal Statement of MLE

- Let $x[1], x[2], \dots, x[M]$ be a sequence of M observed values
 - e.g. $x[m] = H$ or $x[m] = T$ in coin tossing

- **Joint probability:**

$$\begin{aligned} P(D | \theta) &= P(X = x[1])P(X = x[2]) \cdots P(X = x[M]) \\ &= \prod_{m=1}^M P(X = x[m]) \end{aligned}$$

- **Likelihood** is then:

$$L(\theta : D) = \prod_{m=1}^M P(X = x[m])$$

$$\log L(\theta : D) = \sum_{m=1}^M \log P(X = x[m])$$

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More MLE Example

- Say that we want to estimate the recombination fraction (θ) between locus A and B from 5 heterozygous (AaBb) people. We examined 30 gametes for each and observed 4,3,5,6 and 7 recombinant gametes in the five parents. What is the MLE of the recombination fraction θ ?
- Define X = number of recombinant gametes for a single person. Then, the probability that $X = r$ is the following:

$$P\{X = r\} = \binom{n}{r} \theta^r (1-\theta)^{n-r}$$

- where n represents the total number gametes ($n = 30$)

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MLE Example: Specifying Likelihood

- How about all 5 subjects?

- **Probability:**

$$\begin{aligned} P(X_1 = r_1, X_2 = r_2, \dots, X_5 = r_5 | \theta, n) &= P(X_1 = r_1 | \theta, n) \dots P(X_5 = r_5 | \theta, n) \\ &= \binom{n}{r_1} \theta^{r_1} (1-\theta)^{n-r_1} \dots \binom{n}{r_5} \theta^{r_5} (1-\theta)^{n-r_5} \end{aligned}$$

- **Likelihood:**

$$L(\theta : r_1, \dots, r_5, n) = \prod_{i=1}^5 \binom{n}{r_i} \theta^{r_i} (1-\theta)^{n-r_i}$$

$$\log L(\theta : r_1, \dots, r_5, n) = \sum_{i=1}^5 \left[\log \binom{n}{r_i} + r_i \log \theta + (n - r_i) \log(1 - \theta) \right]$$

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MLE Example: Maximizing the Likelihood

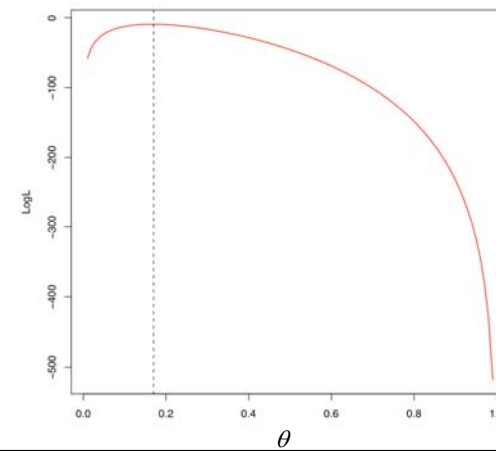
- Again, we want to find p such that $\log(L)$ is maximized:

$$\log L(\theta : r_1, \dots, r_5, n) = \sum_{i=1}^5 \left[\log \binom{n}{r_i} + r_i \log \theta + (n - r_i) \log(1 - \theta) \right]$$

- How?
 - Graphically
 - Draw a plot of $\log(L)$ (y-axis) against p (x-axis)
 - Calculus
 - Differentiate $\log(L)$ with respect to p and equating the derivative to 0
 - Numerically

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MLE Example: Finding the MLE of p



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Goals

- Basic concepts on parameter estimation
- Maximum likelihood estimation (MLE)
- Bayesian inference ←

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World View According to Bayesian's

- The classic philosophy (frequentist) assumes that parameters are **fixed** quantities that we want to estimate as precisely as possible
- Bayesian perspective is different: parameters are **random variables** with probabilities assigned to particular values of parameters to reflect the degree of evidence for that value

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Bayes' Rule

$$P(A|B) = \frac{P(B|A)P(A)}{P(B)}$$

- **Discrete** $P(B) = \sum_{i=1}^n P(B|A=a_i)P(A=a_i)$
- **Continuous** $P(B) = \int P(B|A)P(A)dA$

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Bayesian Estimation

- In order to make probability statements about θ given some observed data, D , we make use of Bayes' rule

$$P(\theta|D) = \frac{P(\theta)P(D|\theta)}{P(D)} = \frac{P(\theta)P(D|\theta)}{\int P(\theta)P(D|\theta)d\theta}$$

← Not a function of θ !

$$P(\theta|D) \propto P(\theta)P(D|\theta)$$

Posterior \propto Prior \times Likelihood

- The **prior** is the probability of the parameter and represents what was thought **before** observing the data
- The **likelihood** is the probability of the data given the parameter and represents the data now available
- The **posterior** represents what is thought given both prior information and the data just **observed**

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Bayesian Estimation

- Find θ such that the posterior $P(\theta|D)$ is maximized

$$P(\theta|D) = \frac{P(\theta)P(D|\theta)}{P(D)} = \frac{P(\theta)P(D|\theta)}{\int P(\theta)P(D|\theta)d\theta}$$

Not a function of θ !

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Simple Example

- Say that we want to estimate the recombination fraction (θ) between locus A and B from 5 heterozygous (AaBb) people. We examined 30 gametes for each and observed 4,3,5,6 and 7 recombinants gametes in the five parents. What is the MLE of the recombination fraction θ ?
- Let's simplify and ask what the recombination fraction (θ) is for subject # 3, who had 5 observed recombinant gametes.

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Specifying The Posterior Density

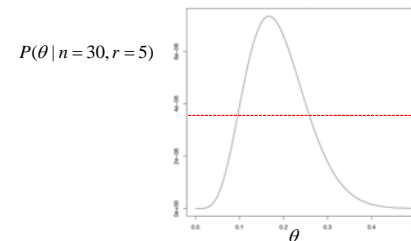
$$P(\theta|D) = P(\theta|n=30, r=5) = \frac{P(\theta)P(r=5|\theta, n=30)}{\int_0^{0.5} P(r=5|\theta, n=30)P(\theta)d\theta}$$

- Prior** $P(\theta) = \text{uniform}[0,0.5] = 0.5$
- Likelihood** $P(r=5|\theta, n=30) = \binom{30}{5} \theta^5 (1-\theta)^{25}$
- Normalizing constant** $\int_0^{0.5} P(r=5|\theta, n=30)P(\theta)d\theta = 0.5 \cdot \binom{30}{5} \int_0^{0.5} \theta^5 (1-\theta)^{25} d\theta \approx 6531$

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Specifying The Posterior Density

$$P(\theta|D) = P(\theta|n=30, r=5) = \frac{P(\theta)P(r=5|\theta, n=30)}{\int_0^{0.5} P(r=5|\theta, n=30)P(\theta)d\theta} = \frac{0.5 \cdot \binom{30}{5} \theta^5 (1-\theta)^{25}}{6531}$$



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