# Estimation

Goal: Estimate a population parameter, $\theta$

Data: $X_1, X_2, \ldots, X_n \sim$ independent and identically distributed with distribution depending on $\theta$

If one has many estimators to choose from, pick

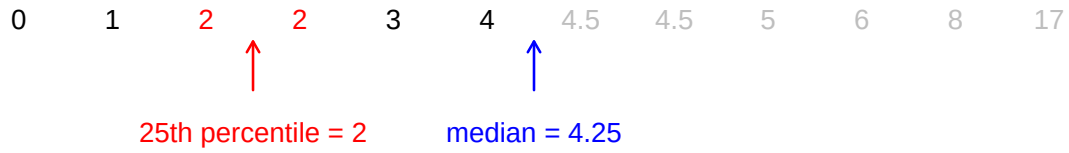
- That with the smallest SE, among all unbiased estimators
- That with the smallest RMS error, even if biased

Sometimes it's not clear how to form even one good estimator.

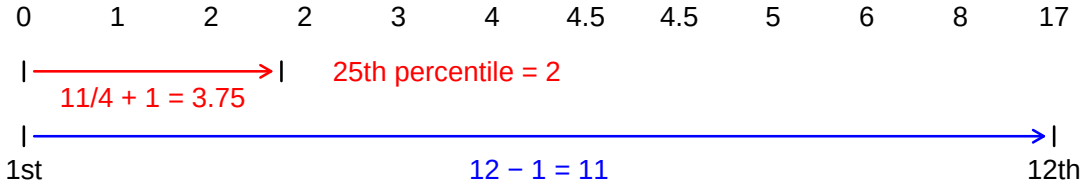


# Estimating the 25th percentile

Method in the book:




Method in R:



# Estimating the 25th percentile

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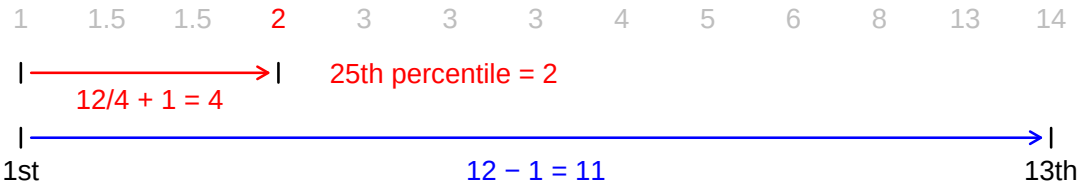
Method in R:





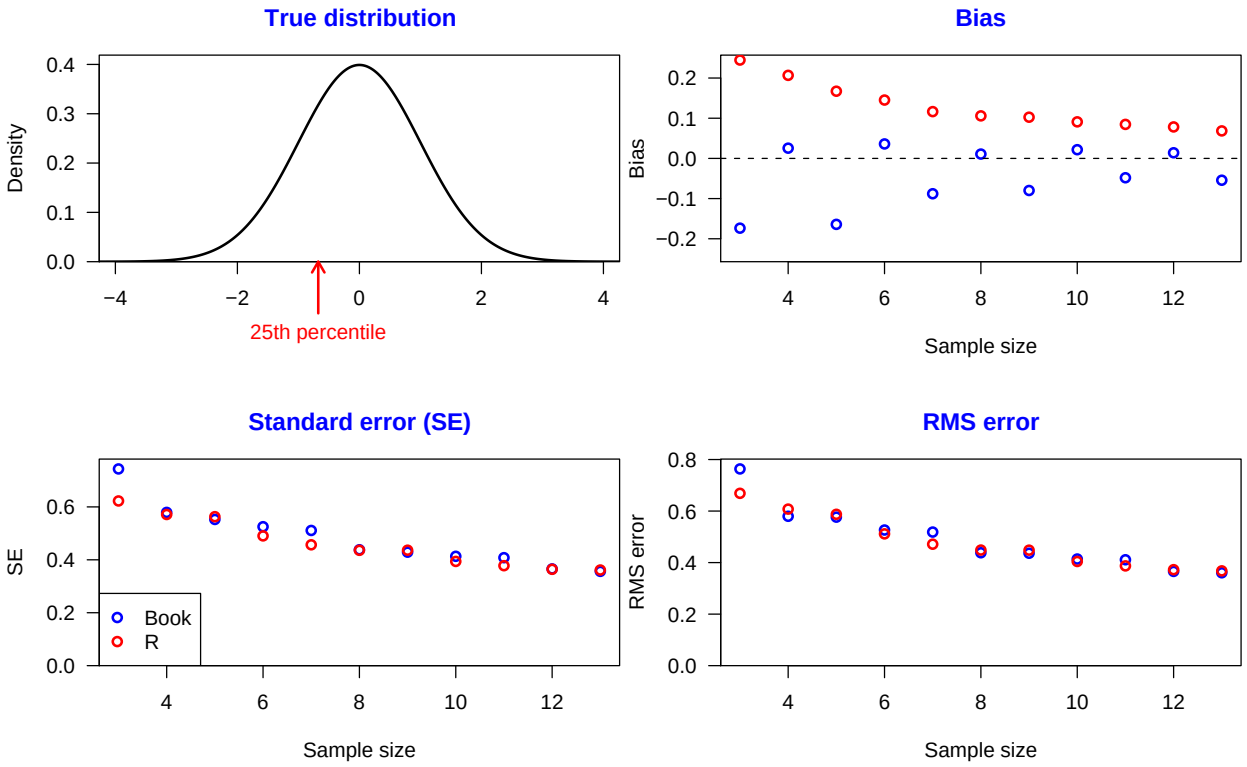

# Estimating the 25th percentile

How to compare these two methods?

- View the data as a random sample from some population.
- How good are the two estimates?
  - Bias
  - Standard error (SE)
  - RMS error (RMSE)
- Computer simulation

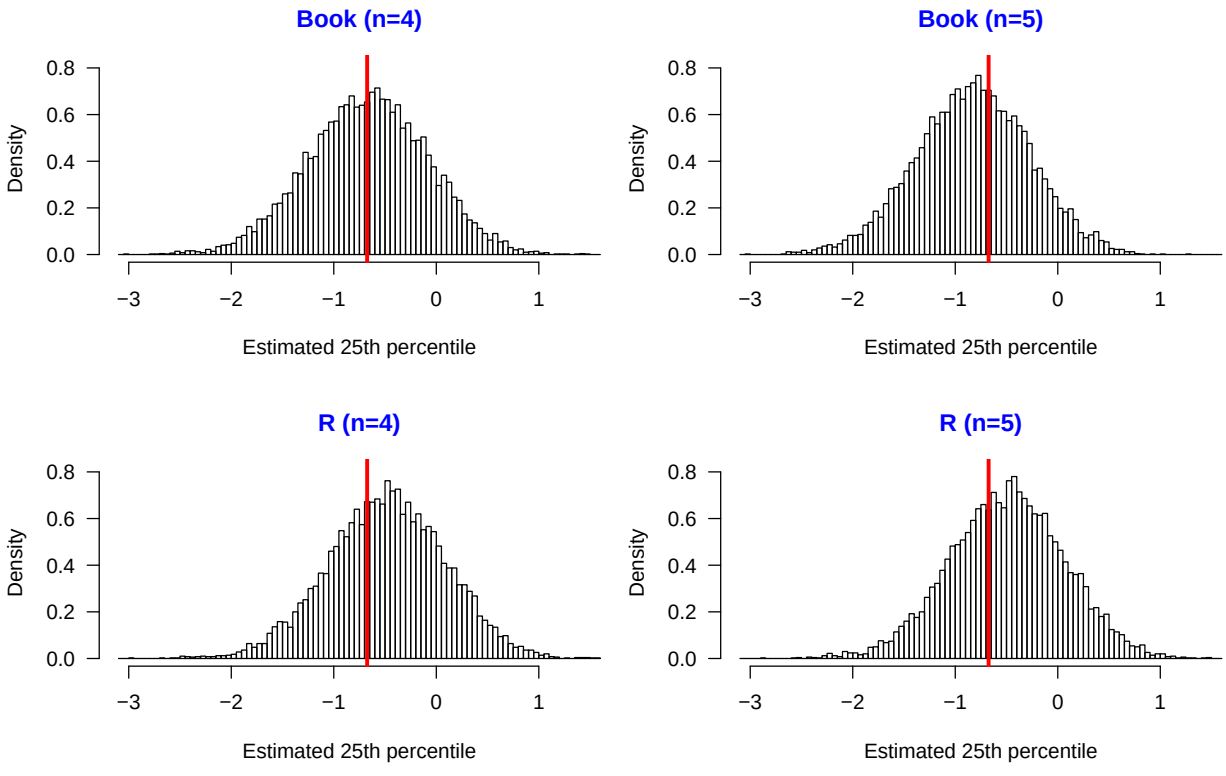
  (using different models for the population distribution)

# Truth = normal distribution

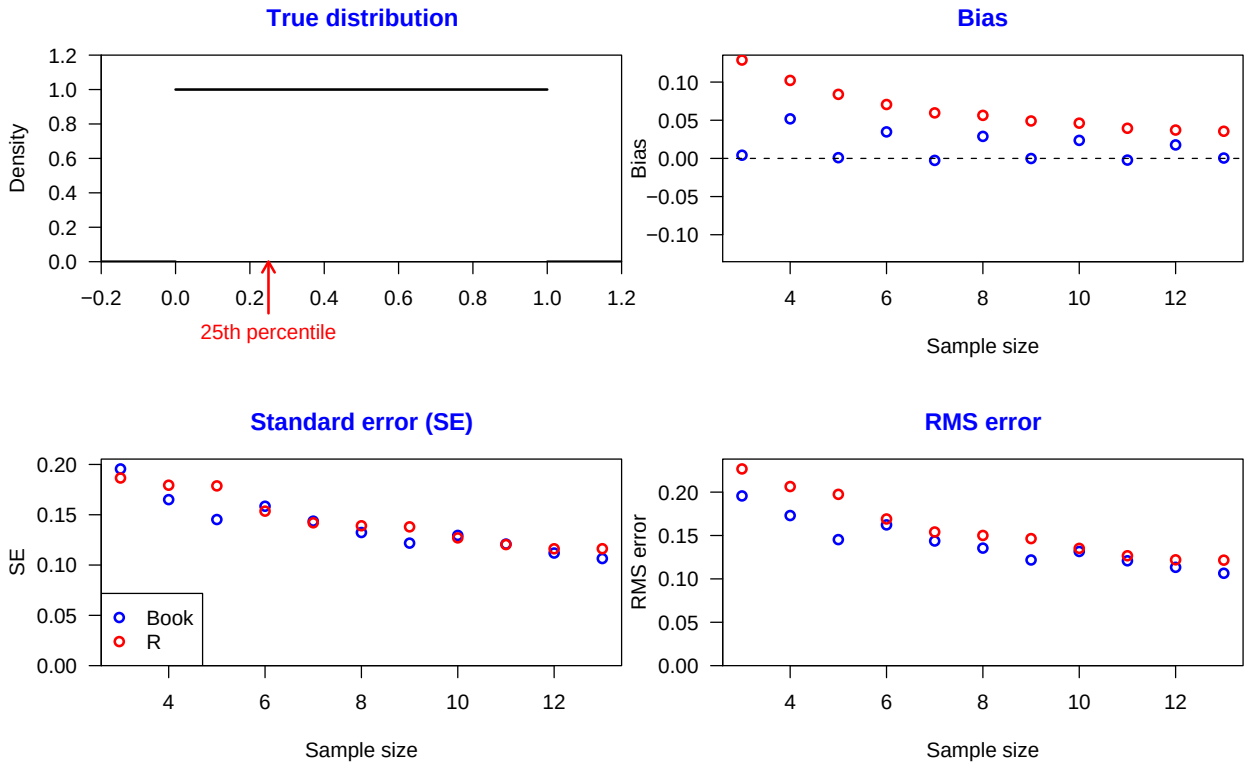





# Truth = normal distribution

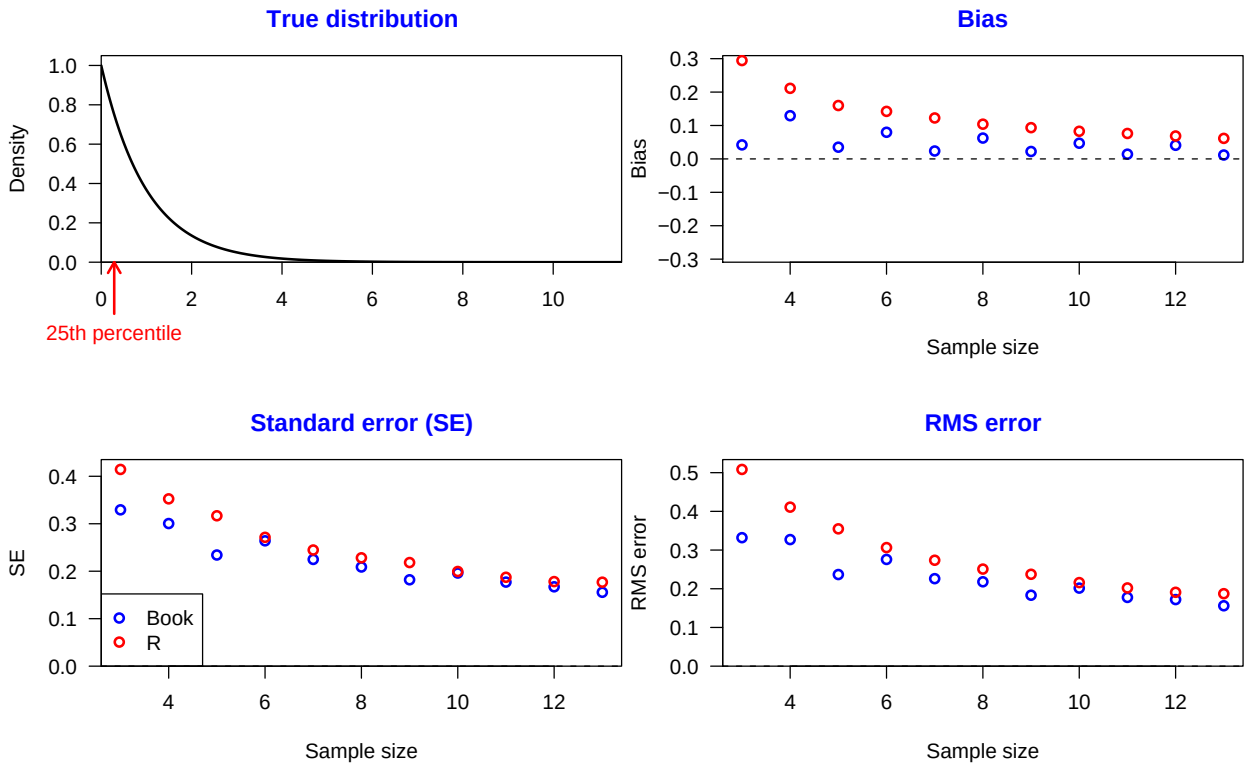


# Truth = uniform distribution

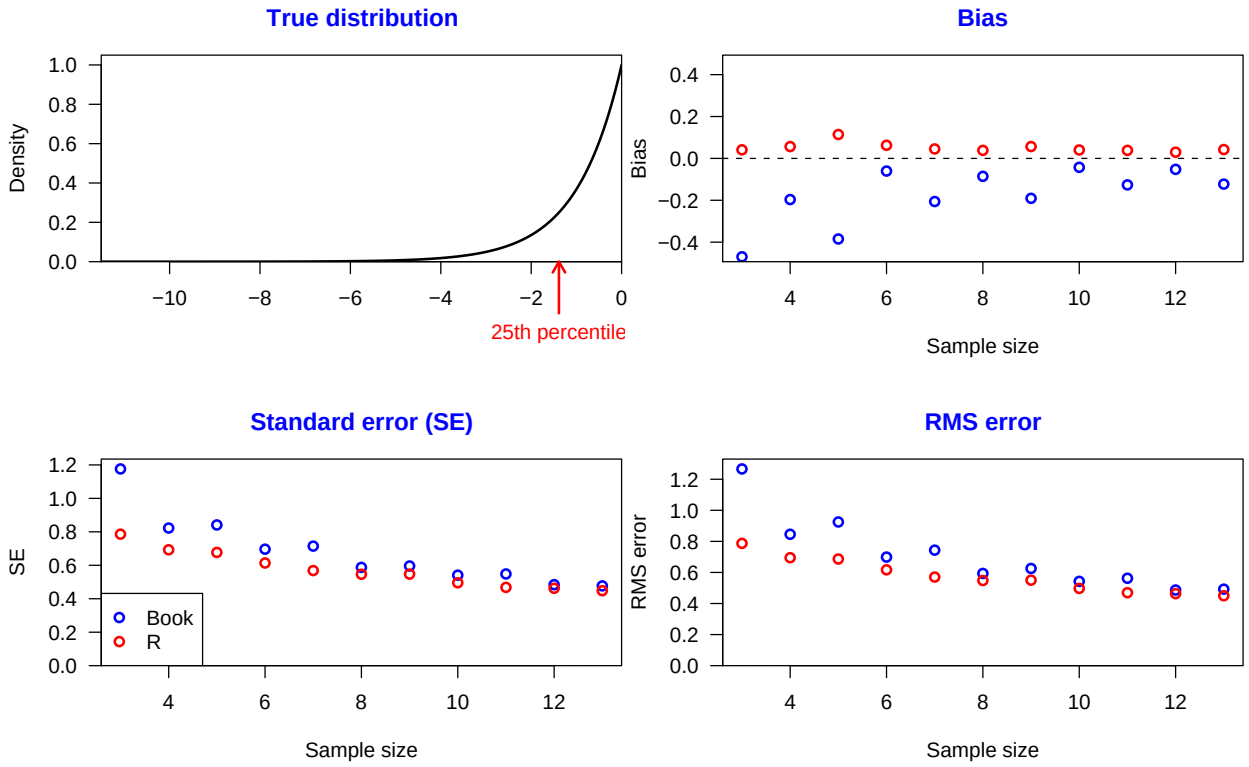





# Truth = exponential distribution



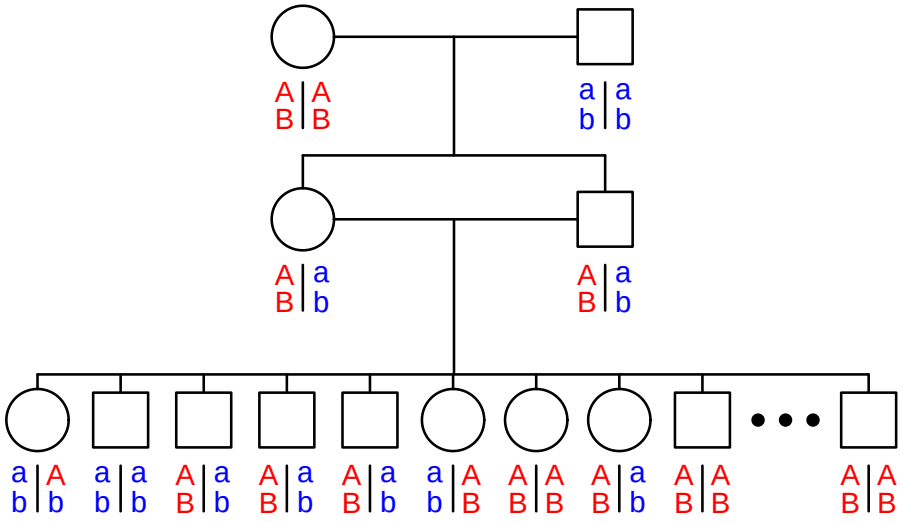
# Truth = negative of exponential






# Example 2

Consider the problem of estimating the recombination fraction (call it $\theta$) between two genetic markers in an intercross.



Note: We won't observe the haplotypes.

# Example 2

## Data

| | AA | Aa | aa |
|---|---|---|---|
| BB | 58 | 9 | 0 |
| Bb | 8 | 95 | 14 |
| bb | 1 | 12 | 53 |

## Probabilities

| | AA | Aa | aa |
|---|---|---|---|
| BB | $\frac{1}{4}(1-\theta)^2$ | $\frac{1}{2}\theta(1-\theta)$ | $\frac{1}{4}\theta^2$ |
| Bb | $\frac{1}{2}\theta(1-\theta)$ | $\frac{1}{2}[\theta^2+(1-\theta)^2]$ | $\frac{1}{2}\theta(1-\theta)$ |
| bb | $\frac{1}{4}\theta^2$ | $\frac{1}{2}\theta(1-\theta)$ | $\frac{1}{4}(1-\theta)^2$ |

Question: Possible estimates of the recombination fraction, $\theta$?



# Maximum likelihood estimation

Likelihood function: $\mathrm{L}(\theta) = \Pr(\text{data} \mid \theta)$

Log likelihood: $\mathrm{l}(\theta) = \ln \Pr(\text{data} \mid \theta)$

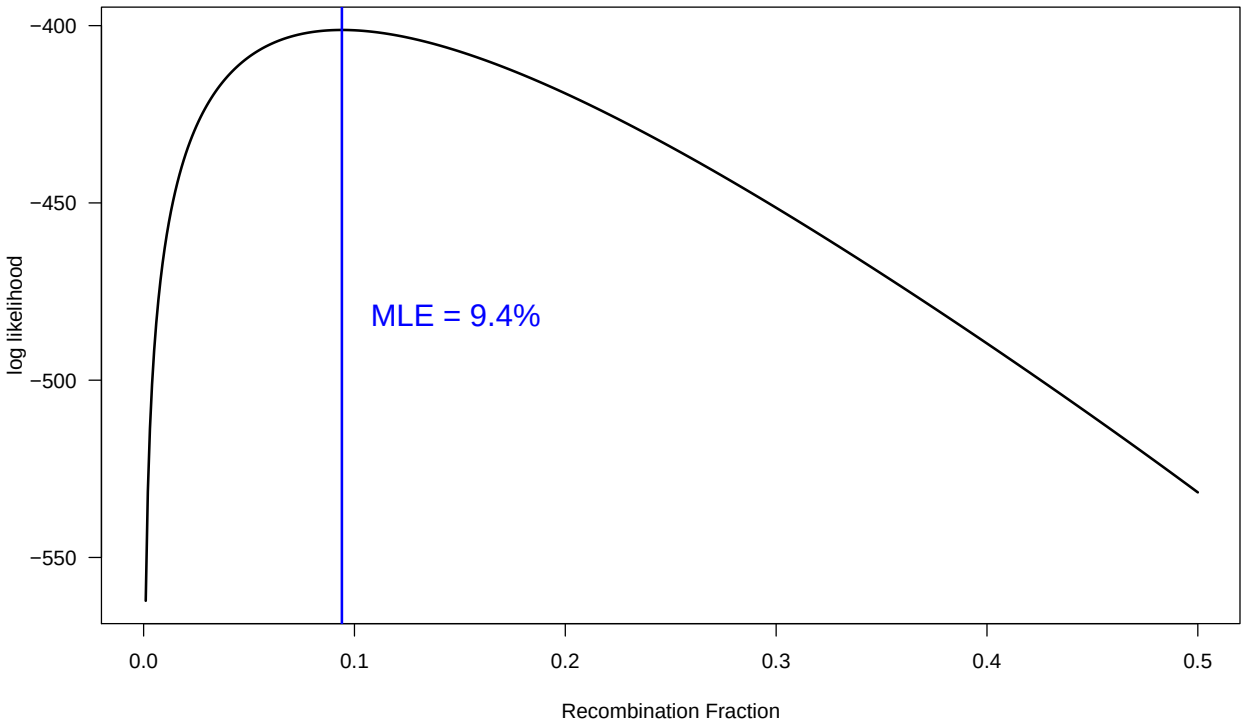
Maximum likelihood estimate:

> Choose, as the estimate of $\theta$, the value of $\theta$ for which $\mathrm{L}(\theta)$ is maximized.

For the example,

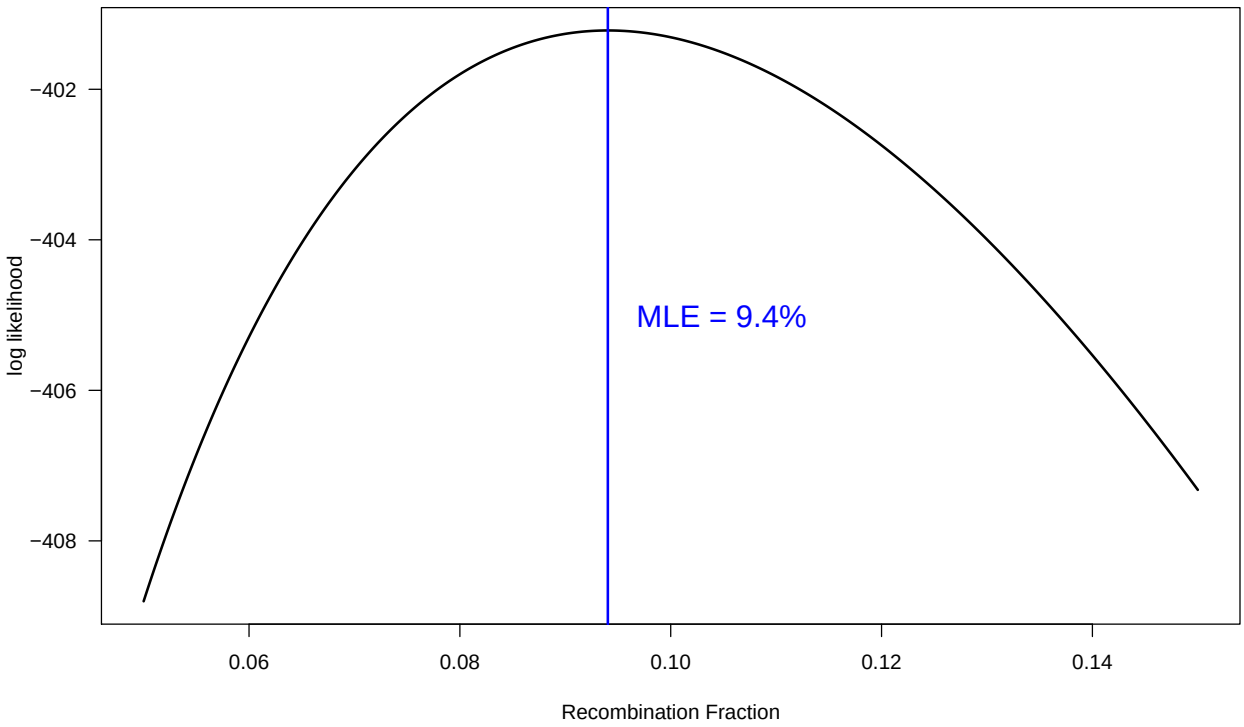
$$\mathrm{L}(\theta) \propto \left\{\tfrac{1}{4}(1-\theta)^2\right\}^{(58+53)} \times \left\{\tfrac{1}{2}\theta(1-\theta)\right\}^{(9+8+14+12)} \times \left\{\tfrac{1}{4}\theta^2\right\}^{(1+0)} \times \left\{\tfrac{1}{2}[\theta^2+(1-\theta)^2]\right\}^{95}$$

# Example 2: Log likelihood function






# A closer view



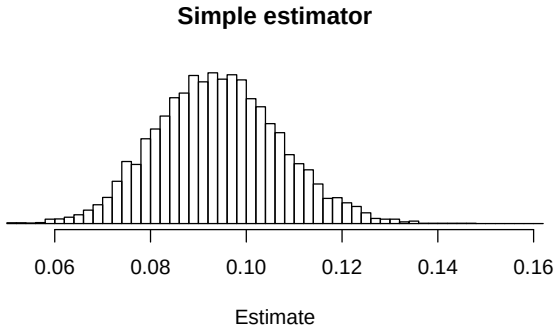
# A comparison of two estimators

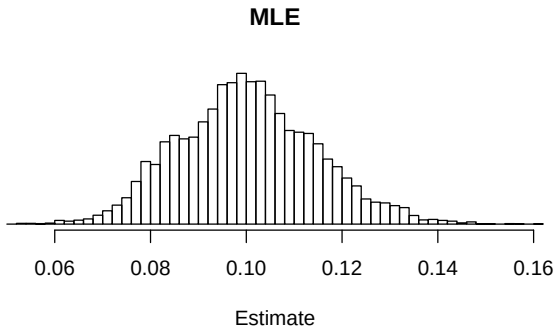
A simple estimator:

Assume double-heterozygotes are non-recombinant

For the case n = 250, $\theta$ = 0.10; the results of 10,000 simulations:

| | Simple | MLE |
|---|---|---|
| Bias | –0.005 | 0.000 |
| SE | 0.013 | 0.014 |
| RMSE | 0.014 | 0.014 |





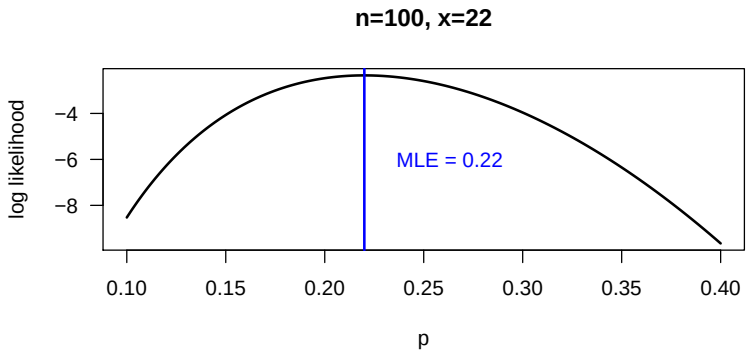

# Example 3

Suppose x $\sim$ binomial(n, p).

log likelihood function: $l(p) = \ln\left\{\binom{n}{x} p^x (1-p)^{(n-x)}\right\}$

$= x \ln(p) + (n-x)\ln(1-p) + \text{constant}$

MLE: the obvious thing: $\hat{p} = x/n$



# Example 4

Suppose $x_1, \ldots, x_n \sim$ iid $N(\mu, \sigma)$

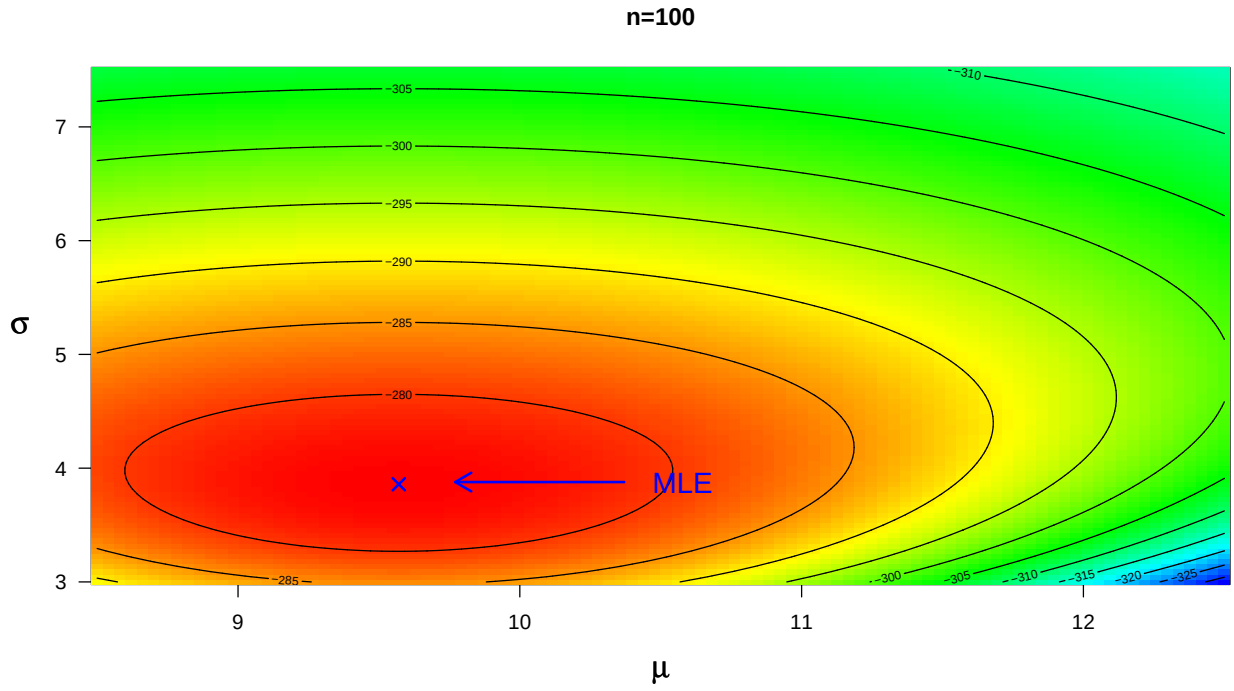
log likelihood function: $l(\mu, \sigma) = \ln \left\{ \prod_i \frac{1}{\sigma\sqrt{2\pi}} \exp\left[ -\frac{1}{2} \left( \frac{x-\mu}{\sigma} \right)^2 \right] \right\}$

MLEs: almost the obvious things:

$$\hat{\mu} = \bar{x} \qquad \hat{\sigma} = \sqrt{\sum (x_i - \bar{x})^2 / n}$$



# Example 4: the log likelihood surface



# About MLEs

Maximum likelihood estimation is a general procedure for finding a reasonable estimator

- In many cases, the MLE turns out to be the obvious thing.
- MLEs are often good (but not necessarily the best) estimators
  - Nearly unbiased
  - small SE
- Sometimes obtaining the MLEs requires hefty computation



# Example 5: ABO blood groups

| Phenotype | Genotype | Frequency |
|---|---|---|
| O | OO | $p_O^2$ |
| A | AA or AO | $p_A^2 + 2p_Ap_O$ |
| B | BB or BO | $p_B^2 + 2p_Bp_O$ |
| AB | AB | $2p_Ap_B$ |

Frequencies under the assumption of Hardy-Weinberg equilibrium.

# Example 5: Data

| Phenotype | No. subjects | % subjects |
|---|---|---|
| O | 117 | 46.8% |
| A | 98 | 39.2% |
| B | 29 | 11.6% |
| AB | 6 | 2.4% |
| **Total** | **250** | **100%** |

Question: Estimates of $p_A$, $p_B$, $p_O$?



# Example 5: Estimates

Simple estimates

$\tilde{p}_O = \sqrt{0.468} = 0.684$

$\tilde{p}_A$ to solve $\tilde{p}_A^2 + 2\tilde{p}_A 0.684 = 0.392$

$\Longrightarrow \quad \tilde{p}_A = 0.243$

$\tilde{p}_B = 0.024/(2\tilde{p}_A) = 0.072$

Log likelihood:

$l(p_O, p_A, p_B) =$

$$117 \ln(p_O^2) + 98 \ln(p_A^2 + 2p_A p_O) + 29 \ln(p_B^2 + 2p_B p_O) + 6 \ln(2p_A p_B)$$

# Example 5: log likelihood