# Examples for large sample credible intervals

# Example 1: Prevalence of Recessive Gene

If gene frequencies are in equilibrium, the genotypes AA, Aa, and aa occur with probabilities  $(1-\theta)^2$ ,  $2\theta(1-\theta)$ , and  $\theta^2$  respectively, where  $\theta$  represents the overall prevalence of the recessive a gene in the population. Plato et al. (1964) published the following data on a haptoglobin type in a sample of 190 people:

Haptoglobin Type	AA	Aa	aa
Count	112	68	10

Let's regard the vector  $\mathbf{x} = (x_1, x_2, x_3) = (112, 68, 10)$  as a realization of the random variable  $\mathbf{X} \sim \text{Multinomial}((1-\theta)^2, 2\theta(1-\theta), \theta^2)$ .

To save some time/allow us to focus on the results of interest here, I'll give you the likelihood function, its first and second derivatives with respect to  $\theta$ , and the form of the posterior:

# **Preliminary Results**

## General form of Multinomial pmf

$$f(\mathbf{x}|\mathbf{p}) = \frac{n!}{x_1! x_2! \cdots x_k!} p_1^{x_1} p_2^{x_2} \cdots p_k^{x_k}$$

#### Likelihood function

In our example,  $p_1 = (1 - \theta)^2$ ,  $p_2 = 2\theta(1 - \theta)$ , and  $p_3 = \theta^2$ .

$$\mathcal{L}(\theta|\mathbf{x}) = f(\mathbf{x}|\theta) = \{(1-\theta)^2\}^{x_1} \{2\theta(1-\theta)\}^{x_2} \{\theta^2\}^{x_3}$$

I'm going to leave this in terms of  $x_1, x_2$ , and  $x_3$  for now.

# Log-likelihood function

$$\ell(\theta|\mathbf{x}) = \log[\mathcal{L}(\theta|\mathbf{x})]$$

$$= \log\left[\{(1-\theta)^2\}^{x_1}\{2\theta(1-\theta)\}^{x_2}\{\theta^2\}^{x_3}\right]$$

$$= x_1 \log\{(1-\theta)^2\} + x_2 \log\{2\theta(1-\theta)\} + x_3 \log\{\theta^2\}$$

#### First and second derivatives of log-likelihood function

The first derivative of the log-likelihood is:

$$\frac{d}{d\theta}\ell(\theta|\mathbf{x}) = \dots = \frac{-2x_1\theta}{\theta(1-\theta)} + \frac{x_2(1-2\theta)}{\theta(1-\theta)} + \frac{2x_3(1-\theta)}{\theta(1-\theta)}$$

The second derivative of the log-likelihood is:

$$\frac{d^2}{d\theta^2}\ell(\theta|\mathbf{x}) = \dots = -\frac{2x_1 + x_2}{(1-\theta)^2} - \frac{2x_3 + x_2}{\theta^2}$$

## Maximum likelihood estimator

Setting the first derivative equal to 0, we obtain a maximum likelihood estimator of  $\hat{\theta}^{MLE} = \frac{X_2 + 2X_3}{2n}$ .

It can be verified that this gives a global maximum of the likelihood function.

#### Posterior Distribution

Suppose we adopt a prior of  $\Theta \sim \text{Uniform}(0,1)$ 

The prior distribution for  $\Theta$  has density  $f_{\Theta}(\theta) = \begin{cases} 1 \text{ if } \theta \in [0,1] \\ 0 \text{ otherwise} \end{cases}$ .

Additionally, in part (a) we showed that  $f_{\mathbf{X}|\Theta}(\mathbf{x}|\theta) = \{(1-\theta)^2\}^{x_1} \{2\theta(1-\theta)\}^{x_2} \{\theta^2\}^{x_3}$ . Applying Bayes' Rule, we find that

$$f_{\Theta|\mathbf{X}}(\theta|\mathbf{x}) = \dots = \begin{cases} c\{(1-\theta)^2\}^{x_1} \{2\theta(1-\theta)\}^{x_2} \{\theta^2\}^{x_3} & \text{if } \theta \in [0,1] \\ 0 & \text{otherwise} \end{cases}$$

The integral is kindof annoying, but can be done.

# Problems for you

#### 1. Find a large-sample normal approximation to the posterior distribution for $\theta$ .

You should find the numeric values of the mean and variance of this normal approximation based on our data set.

Mean:

```
mle <- (68 + 2 * 10)/(2 * 190)
mle
```

## [1] 0.2315789

Variance:

$$\frac{d^2}{d\theta^2} \ell(\theta|x_1, \dots, x_n)|_{\theta = \hat{\theta}^{MLE}} = -\frac{2 \cdot 112 + 68}{\left(1 - \hat{\theta}^{MLE}\right)^2} - \frac{2 \cdot 10 + 68}{\left(\hat{\theta}^{MLE}\right)^2}$$
 second\_deriv\_loglik <- -(2 \* 112 + 68)/(1 - mle)^2 - (2 \* 10 + 68)/(mle^2) post\_approx\_var <- -1/second\_deriv\_loglik post\_approx\_var

## [1] 0.0004682898

Therefore, the posterior distribution for  $\theta$  is approximately Normal (0.232, 0.000468)

# 2. Add a plot of the pdf of the normal approximation to the plot of the actual posterior below.

I used Wolfram Alpha to figure out how to calculate the constant c. That's the first 4 lines of the calculation of the log density in the dposterior function below:

```
library(ggplot2)
dposterior <- function(theta, x_1, x_2, x_3, log = FALSE) {</pre>
    n \leftarrow x_1 + x_2 + x_3
    log_d_posterior <- sum(log(seq_len(2 * n + 1))) -</pre>
        x_2 * log(2) -
        sum(log(seq_len(2 * x_1 + x_2))) -
        sum(log(seq_len(x_2 + 2 * x_3))) +
        2 * x_1 * log(1 - theta) +
        x_2 * log(2 * theta * (1 - theta)) +
        2 * x_3 * log(theta)
    if(log) {
        return(log_d_posterior)
    } else {
        return(exp(log_d_posterior))
    }
}
ggplot(data = data.frame(theta = c(0, 1)), mapping = aes(x = theta)) +
    stat_function(fun = dposterior, args = list(x_1 = 112, x_2 = 68, x_3 = 10), n = 1001) +
    stat_function(fun = dnorm, args = list(mean = 0.232, sd = sqrt(0.000468)), color = "orange")
  15 -
  10 -
   5 -
                           0.25
                                               0.50
       0.00
                                                                   0.75
                                                                                       1.00
                                              theta
```

3. Find and interpret an approximate Bayesian 95% credible interval for  $\theta$  based on the normal approximation.

```
qnorm(c(0.025, 0.975), mean = 0.232, sd = sqrt(0.000468))
```

**##** [1] 0.1895995 0.2744005

There is probability 0.95 that the proportion of people in the population carrying the recessive gene is between about 0.19 and 0.27.