

Gibbs Sampling

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Introduction

Using Bayesian statistics we can incorporate prior knowledge into the estimation of our unknown parameters. We incorporate our prior knowledge of the unknown parameter through a prior distribution. Then, we will update our beliefs about θ with observed data and we will end up with what we call a posterior distribution. We will use the posterior to estimate the parameters of interest. Gibbs sampling is a type of Markov Chain Monte Carlo sampling that approximates posterior distributions when we can't sample from the posterior distributions directly. This method is an alternative to Metropolis-Hastings. In a nutshell, Gibbs sampling is a method that samples from separate conditional distributions and is most useful when the joint posterior distribution is unknown or hard to sample from. Each event is dependent on the last event; and it is only dependent on the last event (like in a standard Markov Chain).

Motivation

Gibbs sampling is a useful algorithm for Bayesian estimation. We think that incorporating prior knowledge (priors) and data in the estimation of a parameter can be useful in different fields including quantitative finance and bio statistics. Some people might be familiar with the Metropolis-Hastings algorithm to implement a simulation and to sample our posterior distributions. Gibbs is a suitable alternative to Metropolis-Hastings depending on the information one has available. We were interested in the applications of Monte Carlo simulations in finance and also wanted to explore other Bayesian Statistics algorithms more deeply.

Background Knowledge

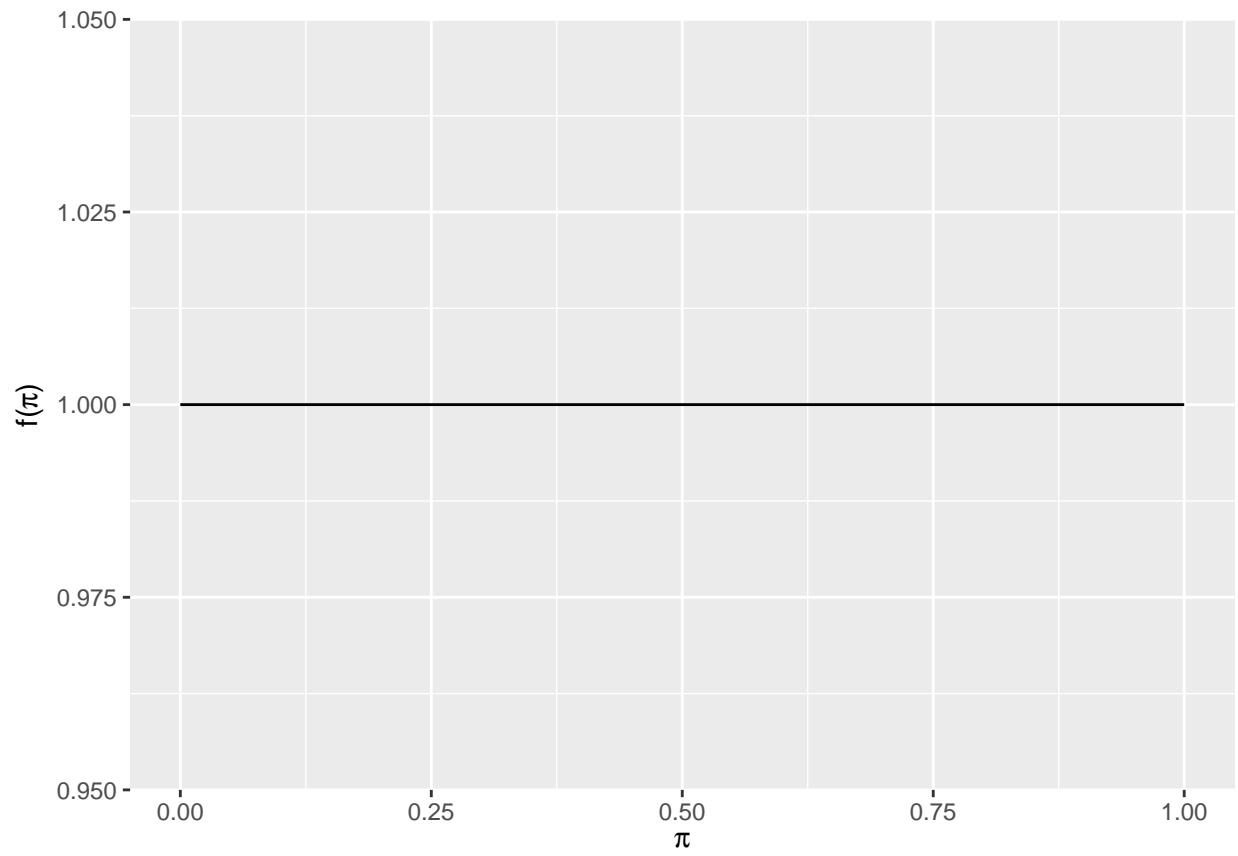
Bayesian Statistics

Bayesian Statistics is a different philosophy of statistics. Most statistical modeling that people use are under the frequentist school of thought. Frequentists methods depend only on the observed data. This means that interpretations and inference can only be from the sample that was studied or collected. Bayesian methods incorporate prior beliefs into the model before the data is received.

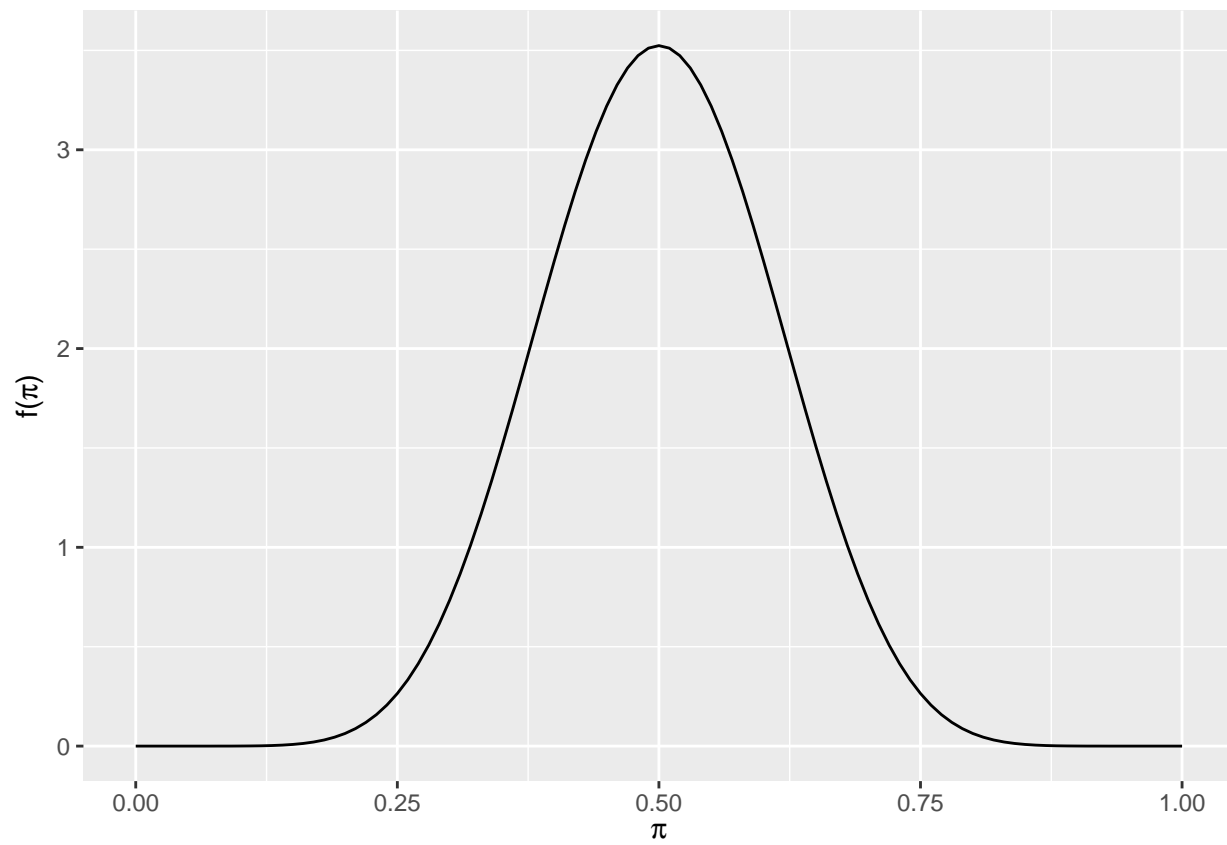
Example 1

[EXPLAIN EXAMPLE, EXPLAIN WHAT DOES IT MEAN THAT IT HAS BETA PRIOR] Prior: A prior is a probability distribution that shows our beliefs before any data is taken into account. A common distribution used for a prior is a beta model. Priors incorporate both the mean and the variance of your previous beliefs about the data. Priors with a greater spread indicate a less confident belief. [EXPLAIN WHAT YOU ARE DOING HERE WITH THIS GRAPHS]

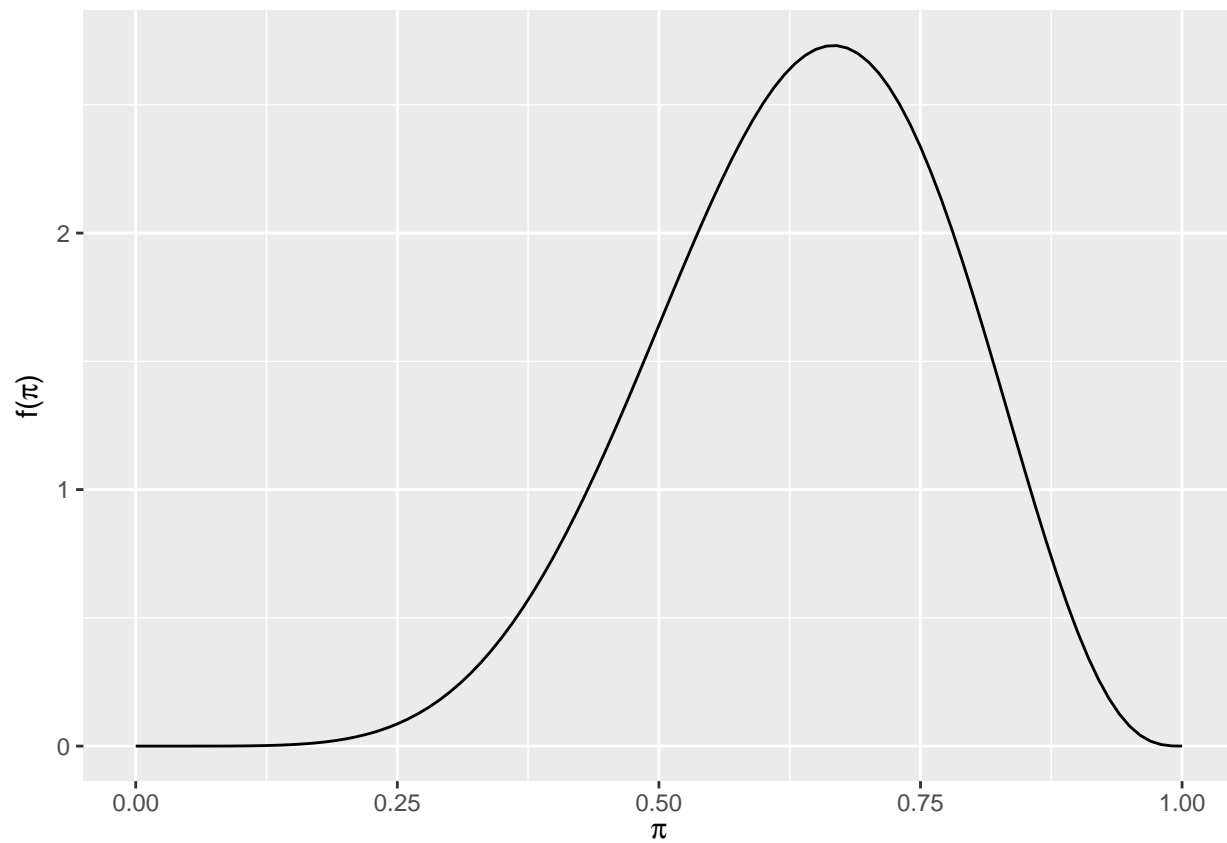
```
plot_beta(alpha = 1, beta = 1)
```



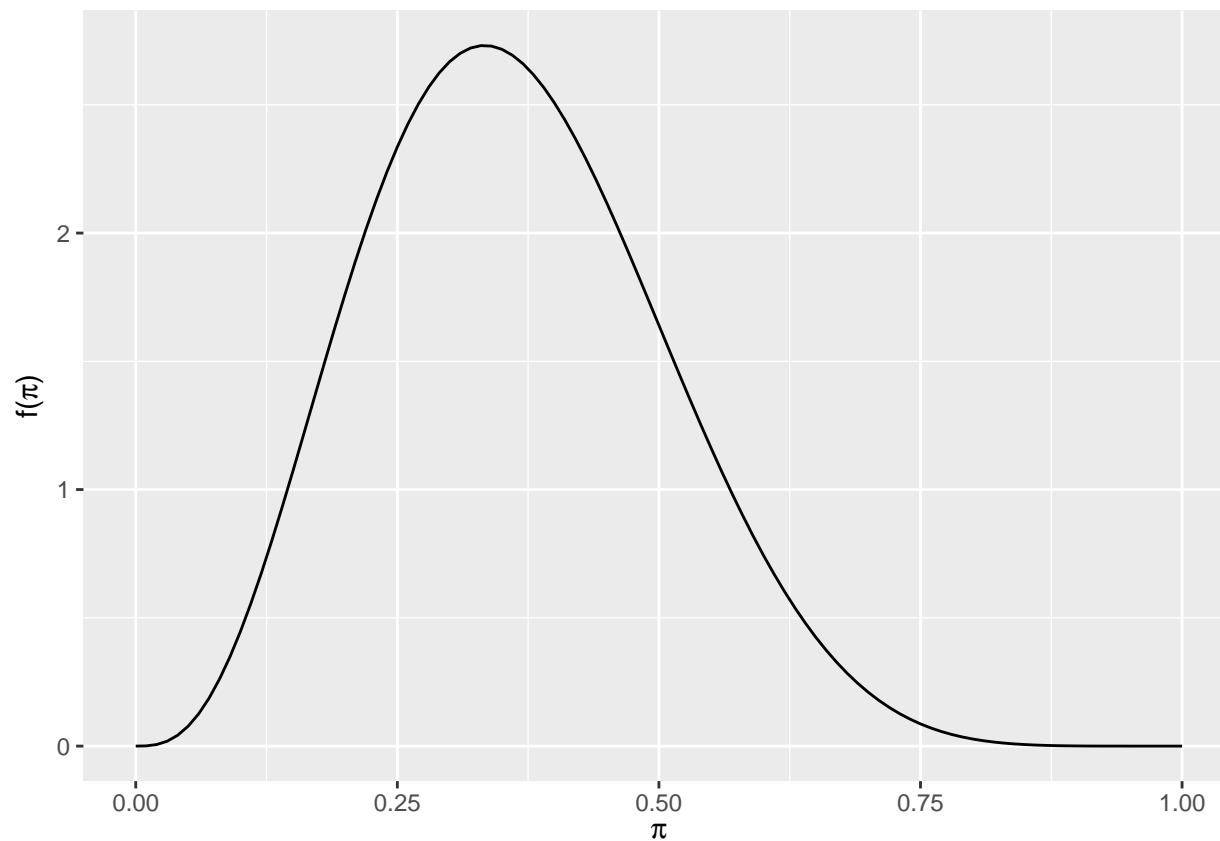
```
plot_beta(alpha = 10, beta = 10)
```



```
plot_beta(alpha = 7, beta = 4)
```

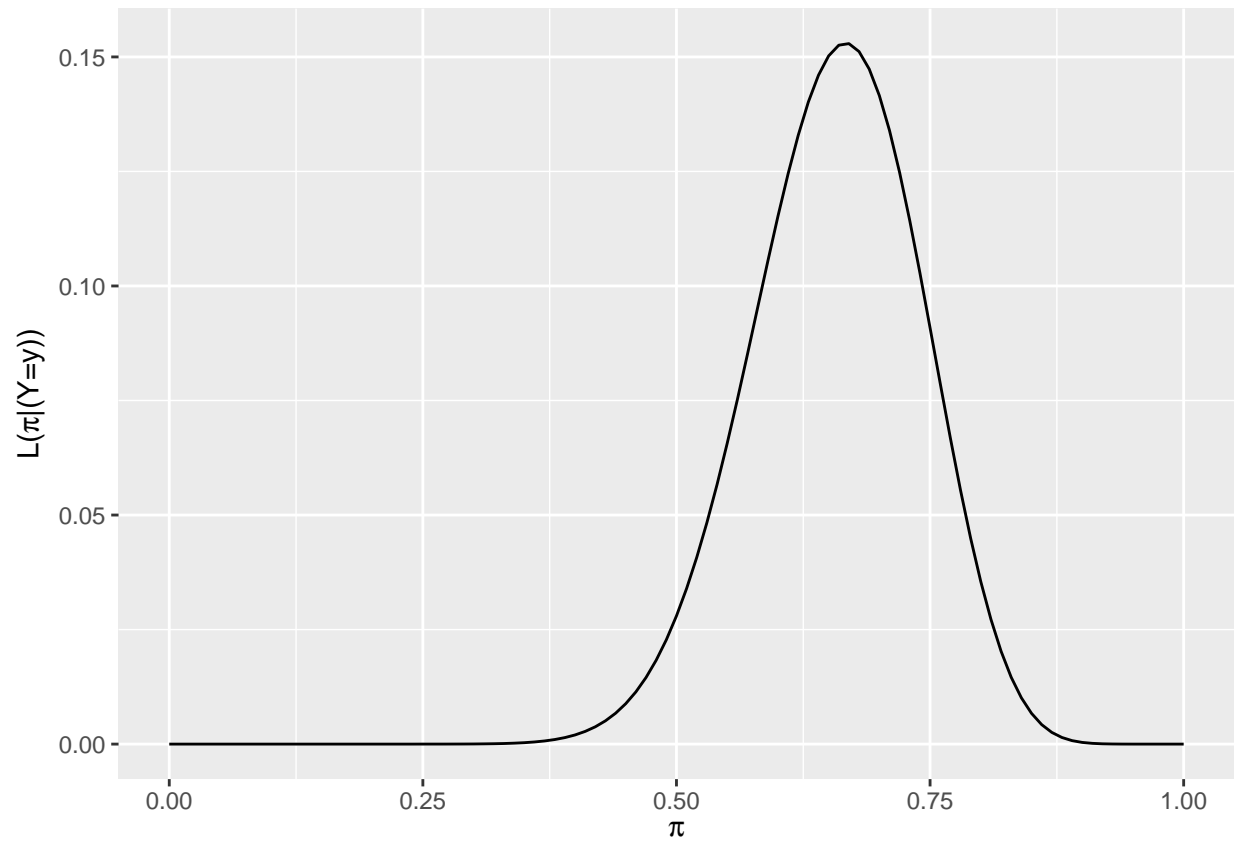


```
plot_beta(alpha = 4, beta = 7)
```



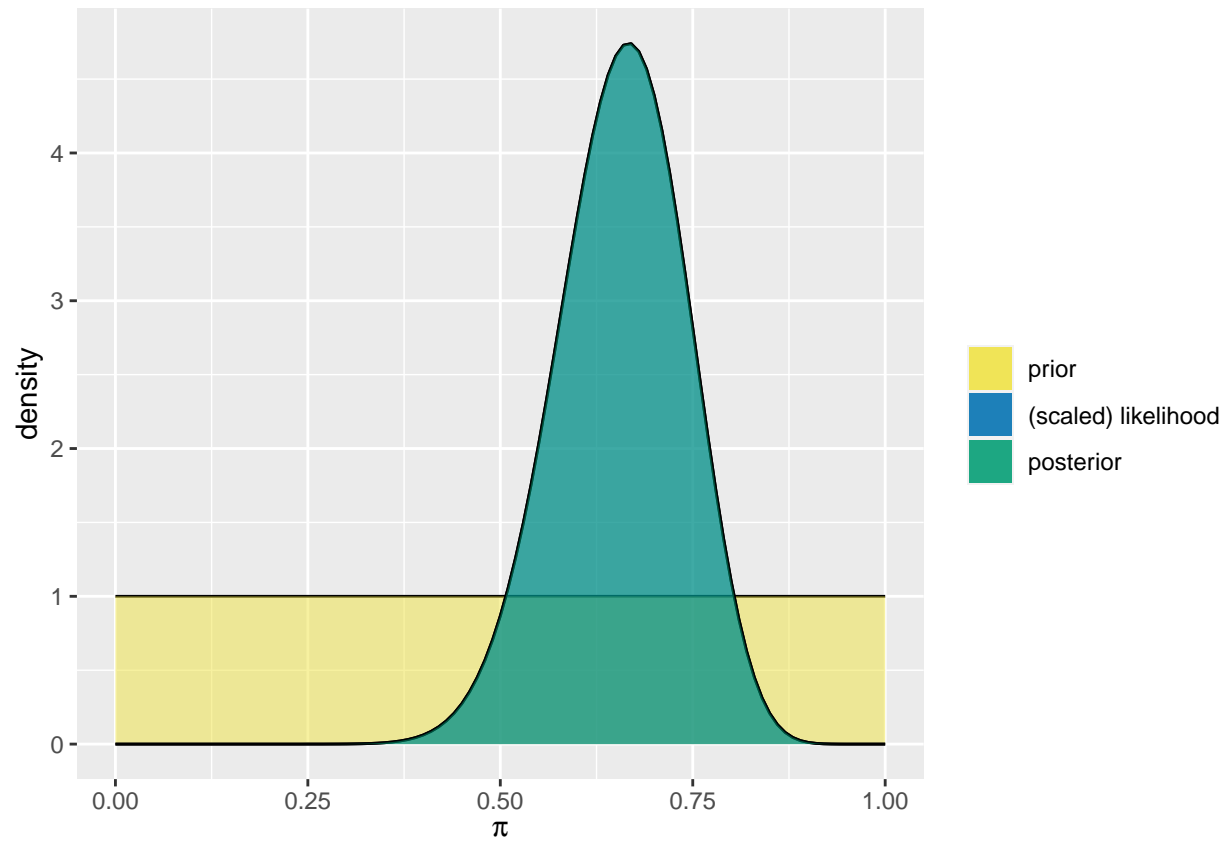
Likelihood is the distribution of the data. In this example the outcome variable is binary on whether the person was voted for or not. So we have a binomial distribution of the likelihood. [EXPLAIN WHAT THE GRPAHS ARE AND THE PURPOSE]

```
plot_binomial_likelihood(20,30)
```

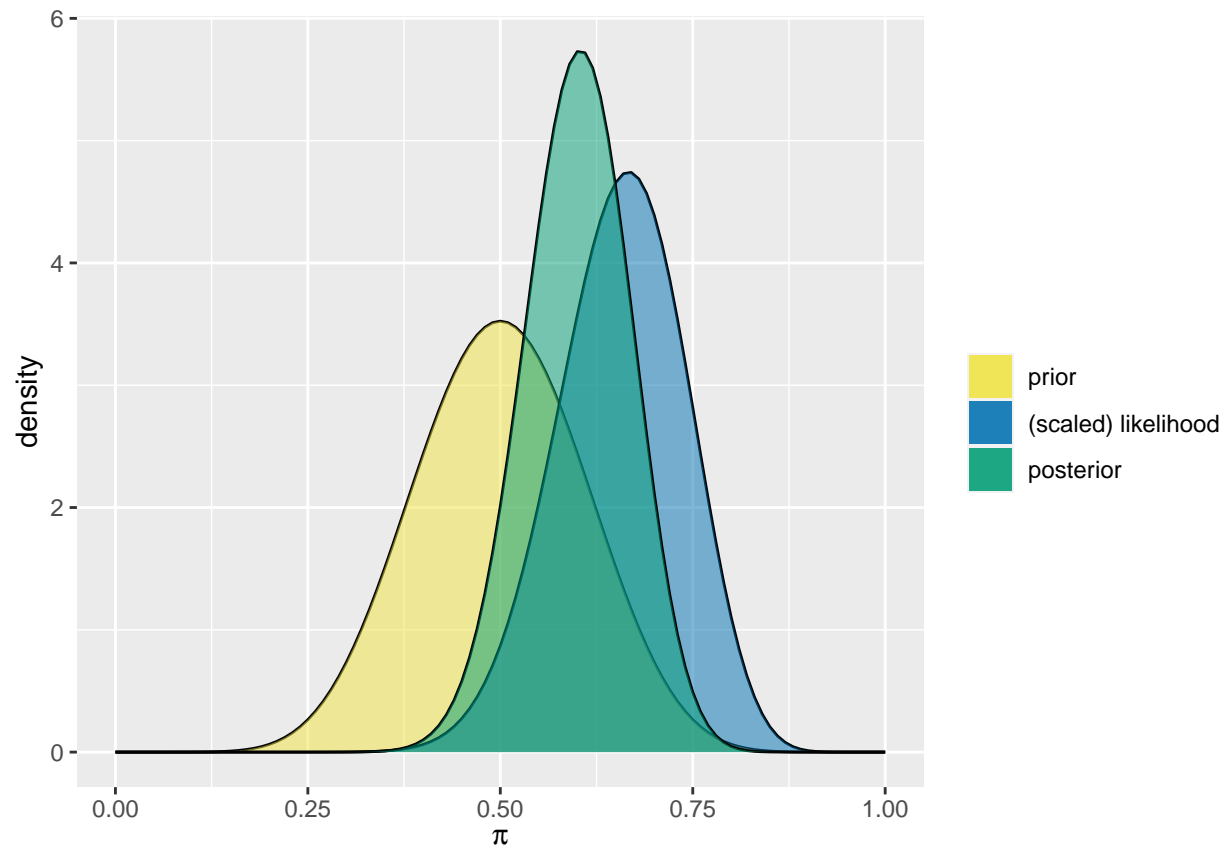


Posterior The posterior is simply a combination of the prior and data. In this case we conveniently chose conjugate distribution in which the prior and posterior share the same distribution. We combine our data with our prior model to form the posterior distribution. As more data is incorporated more weight shifts to the data from the prior. [EXPLAIN WHAT THE GRPAHS ARE AND THE PURPOSE]

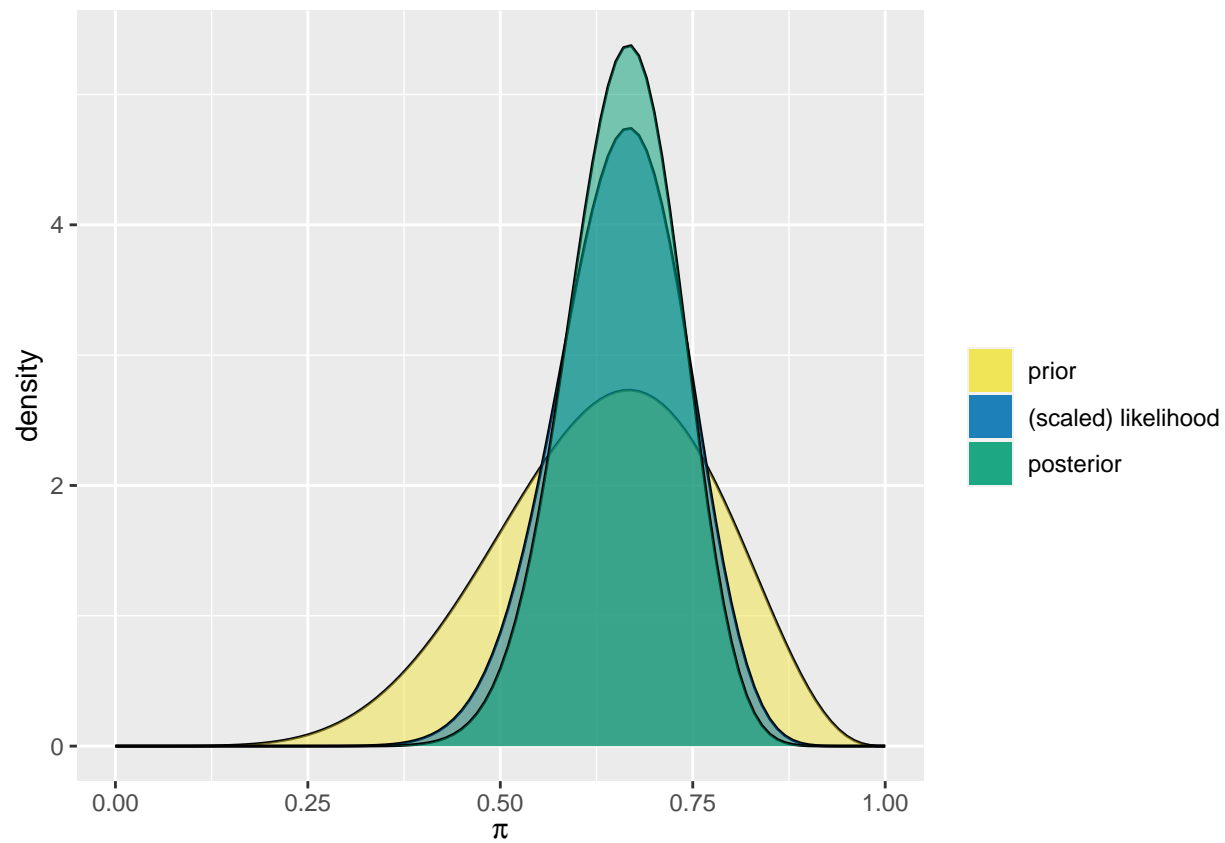
```
plot_beta_binomial(alpha = 1, beta = 1, y = 20, n= 30)
```



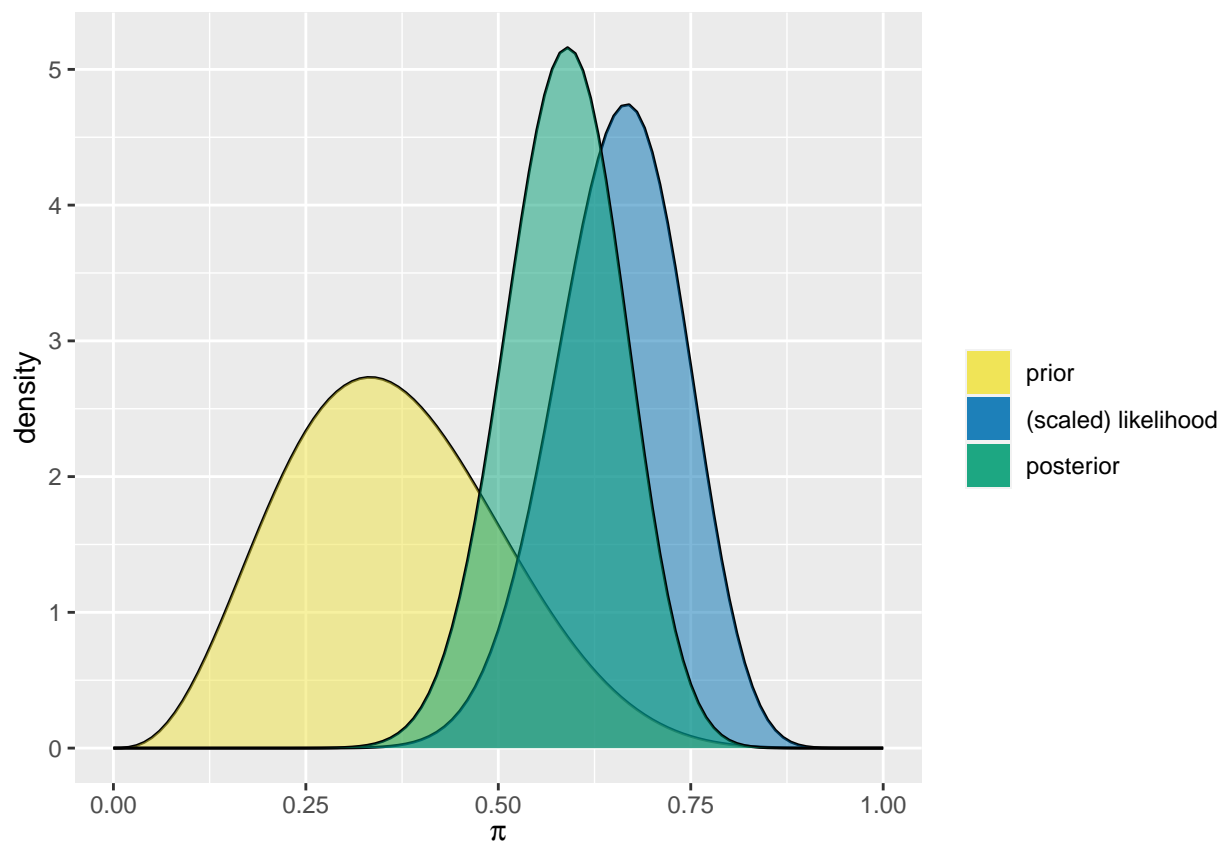
```
plot_beta_binomial(alpha = 10, beta = 10, y = 20, n= 30)
```



```
plot_beta_binomial(alpha = 7, beta = 4, y = 20, n = 30)
```

```
plot_beta_binomial(alpha = 4, beta = 7, y = 20, n= 30)
```



Monte Carlo <https://towardsdatascience.com/an-overview-of-monte-carlo-methods-675384eb1694>

Monte Carlo simulations randomly sample points within a region to approximate a distribution. The example above is a simple illustration of a uniform distribution for an estimate for π . This samples the proportion of points within the square region that fall within the circle's bounds. The proportion would be equal to $\frac{\pi}{4}$ since we are only interested in one fourth of the circle. As we sample more, our estimation for π gets closer to the actual distribution. This is due to the Central Limit Theorem. Monte Carlo simulations work well when the posterior distribution is easy to sample from. However, it is not always possible to sample from the posterior distribution, nor is it always efficient.

Markov Chains

Markov Chains are an example of a random walk. Random walks are a series of random moves through space in succession. Random walks use a combination of past events in the probability to determine the next step. Markov Chains are a special case in which only the previous step/location is used to determine the probability distribution of the next step. The following notation represents this process $P(X_{n+1} = x | X_n = x_n)$ meaning the probability distribution of move $n+1$ is only conditioned on the result of the previous move n . It is important to talk about the fact that Markov Chains are dependent on the previous move and are not an independent event.

[NEED A PICTURE]

Gibbs Sampling Overview

Gibbs Sampling is a specific type of MCMC sampling that is used when it is hard to sample from the joint pdf or pmf or when the joint pdf (or pmf) is unknown. To perform Gibbs sampling you must know the conditional distributions of both variables.

Markov Chains Monte Carlo (MCMC)

MCMC is the application of Markov Chains to simulate probability models. Two important characteristics are that MCMC samples aren't taken from the posterior pdf and that the samples aren't independent. The fact that the samples aren't independent reflects the "chain" nature of the algorithm. For example in the $N - length$ MCMC sample (Markov chain) $\{\theta^{(1)}, \theta^{(2)}, \dots, \theta^{(N)}\}$, when constructing the chain $\theta^{(2)}$ is drawn from some model that depends upon $\theta^{(1)}$, $\theta^{(3)}$ is drawn from some model that depends on $\theta^{(2)}$ and so on.

We can say that the $(i+1)$ st chain value $\theta^{(i+1)}$ has a conditional pdf $f(\theta^{(i+1)}|\theta^{(i)}, y)$ is drawn from a model that depends on data y and the previous chain value $\theta^{(i)}$. It's important to note that by the Markov property, $\theta^{(i+1)}$ depends on the preceding chain values only through $\theta^{(i)}$, the most recent value. The only information we need to simulate $\theta^{(i+1)}$ is the value of $\theta^{(i)}$. Therefore, each value can be sampled from a different model, and none of these models are the target posterior. The pdf from which a Markov Chain value is simulated is not equivalent to the posterior pdf.

$$f(\theta^{(i+1)}|\theta^{(i)}, y) \neq f(\theta^{(i+1)}|y)$$

We will conduct the MCMC simulation using the rstan package (Guo and Weber 2020). There are two essential steps to all rstan analyses, first we define the Bayesian model structure and then simulate the posterior. We will use a Beta-Binomial example.

STEP 1: DEFINE the model

Data: Y is the observed number of success trials. We specify that Y is between 10 and 0. *Parameters:* The model depends on π , therefore we must specify that π can be any real number from 0 to 1. *Model:* We need to specify the model for the data and the model for the prior.

```
# STEP 1: DEFINE the model
bb_model <- "
  data {
    int<lower = 0, upper = 10> Y;
  }
  parameters {
    real<lower = 0, upper = 1> pi;
  }
  model {
    Y ~ binomial(10, pi);
    pi ~ beta(2, 2);
  }
"
```

STEP 2: Simulate the posterior

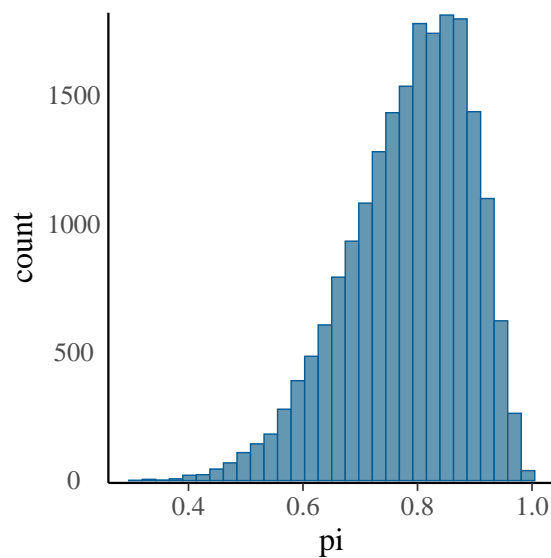
We simulate the posterior using the stan() function. This function designs and runs an MCMC algorithm to produce an approximate sample from the Beta-Binomial posterior. The model code argument requires a string that defines the model. The data argument requires a list of observed data. The chains argument

specifies how many parallel Markov Chains we are running. Since we are running four chains we will have four π values. The “iter” argument specifies the number of iterations or length for each chain. The first half of this iterations are thrown out as “burn in” samples (samples that we use to calibrate our model). To keep our random results constant we utilize the seed argument within the stan() function.

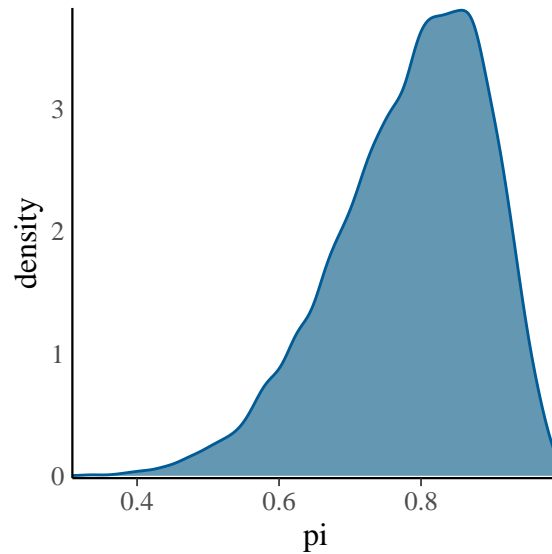
As you can see in Figure 1, when observing the distribution of the sampled π values we approximate the target Beta(11,3) posterior model of π . The target pdf is next to it. (Alicia A. Johnson 2022)

```
# Histogram of the Markov chain values  
mcmc_hist(bb_sim, pars = "pi") +  
  yaxis_text(TRUE) +  
  ylab("count")
```

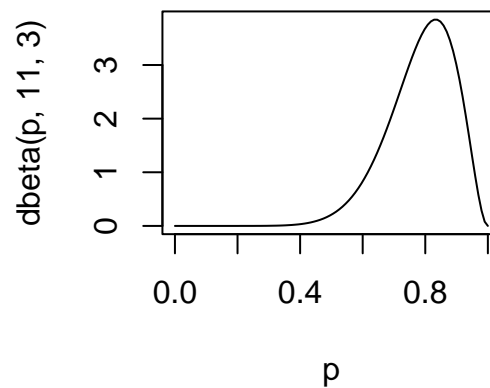
```
## ‘stat_bin()’ using ‘bins = 30’. Pick better value with ‘binwidth’.
```



```
# Density plot of the Markov chain values  
p = seq(0, 1, length=100)  
mcmc_dens(bb_sim, pars = "pi") +  
  yaxis_text(TRUE) +  
  ylab("density")
```



```
plot(p, y = dbeta(p, 11, 3), type='l')
```



Metropolis-Hastings algorithm

If we weren't able to recognize the posterior model of μ in a Normal-Normal model, we could approximate it using the MCMC simulation. Metropolis-Hastings algorithm helps automate the decision of what values of μ to sample and with what frequency. This algorithm iterates through a two step process. If we are in the location $\mu^{(i)} = \mu$ we select the next value to sample first by proposing a random location μ' and then we decide whether to stay at the current location or to stay at the current location $\mu^{(i+1)} = \mu$.

There are special cases of the Metropolis-Hastings that involve a different sampling decision criteria such as the Gibbs sampling, the Monte Carlo and the Metropolis algorithms. In this report we will be focusing on the Gibbs Sampling algorithm.

Gibbs Sampling

Example 1: Bernoulli Distribution:

Example from Youtube

We start with an example of two random variables A, B with a Bernoulli distribution. We now find their conditional distributions

$$P(A|B=0) \in P(A=1) = \frac{4}{5}, P(A=0) = \frac{1}{5}$$

$$P(A|B=1) \in P(A=1) = \frac{2}{5}, P(A=0) = \frac{3}{5}$$

$$P(B|A=0) \in P(B=1) = \frac{3}{4}, P(B=0) = \frac{1}{4}$$

$$P(B|A=1) \in P(B=1) = \frac{1}{3}, P(B=0) = \frac{2}{3}$$

1. Pick specific starting value of (x_0, y_0) Here we pick $(A_0), (B_0)$
2. Condition on B_0
3. Your distribution is now $P(A=0) = 1/5$ and $P(A=1) = 4/5$
4. Randomly sample
5. Your random sample leads to $A=1$
6. Condition on A_1
7. Your distribution is now $P(B=0) = 2/3$ and $P(B=1) = 1/3$
8. Randomly sample
9. Your random sample leads to $B=1$
10. Condition on B_1 Now the process repeats thousands of times until and each move is recorded. This algorithm then approximates well the true probability distribution after thousand of trials. More trials will lead to a better approximation.

Example 2: Normal Distribution

Now suppose we have data from a normal distribution where both the mean **and** variance are unknown. For convenience, we'll parameterize this model in terms of the *precision* $\gamma = \frac{1}{\sigma^2}$ instead of the variance σ^2 .

$$Y | \mu, \gamma \sim N\left(\mu, \frac{1}{\gamma}\right)$$

Suppose we put the following *independent* priors on the mean μ and precision γ :

$$\mu \sim N(m, v)$$

$$\gamma \sim \text{Gamma}(a, b)$$

1. Write down the joint posterior distribution for μ, γ . Does this look like a recognizable probability distribution?

ANSWER: No, this is not a recognizable posterior:

$$\begin{aligned}
g(\mu, \gamma | y) &\propto f(y | \mu, \gamma) f(\mu, \gamma) \\
&= f(y | \mu, \gamma) f(\mu) f(\gamma), \text{ since } \mu, \gamma \text{ independent} \\
&= \left[(2\pi)^{-\frac{1}{2}} \gamma^{\frac{1}{2}} e^{-\frac{1}{2}\gamma(y-\mu)^2} \right] \left[(2\pi v)^{-\frac{1}{2}} e^{-\frac{1}{2v}(\mu-m)^2} \right] \left[\frac{b^a}{\Gamma(a)} \gamma^{a-1} e^{-b\gamma} \right] \\
&\propto \gamma^{\frac{1}{2}} e^{-\frac{1}{2}\gamma(y-\mu)^2} e^{-\frac{1}{2v}(\mu-m)^2} \gamma^{a-1} e^{-b\gamma} \\
&= \gamma^{\frac{1}{2}+a-1} e^{-\frac{1}{2}\gamma(y-\mu)^2 - \frac{1}{2v}(\mu-m)^2 - b\gamma} \\
&= \gamma^{\frac{1}{2}+a-1} e^{-\frac{1}{2}[\gamma(y-\mu)^2 + \frac{1}{v}(\mu-m)^2 + 2b\gamma]} \\
&= \gamma^{\frac{1}{2}+a-1} e^{-\frac{1}{2}[\gamma y^2 - 2\mu y \gamma + \gamma \mu^2 + \mu^2/v - 2m\mu/v + m^2/v + 2b\gamma]} \\
&= \gamma^{\frac{1}{2}+a-1} e^{-\frac{1}{2}[\gamma(y^2+2b) - 2\mu(y\gamma+m/v) + \mu^2(\gamma+1/v) + m^2/v]} \\
&\propto \gamma^{\frac{1}{2}+a-1} e^{-\frac{1}{2}[\gamma(y^2+2b) - 2\mu(y\gamma+\frac{m}{v}) + \mu^2(\frac{1}{v}+\gamma)]}
\end{aligned}$$

You should have answered “no” to Question 1, meaning that we can’t use our usual techniques here to find Bayes estimators for μ or γ since we don’t have a recognizable posterior distribution. Instead, we’ll use a computational technique known as *Gibbs Sampling* to generate samples from this posterior distribution. Gibbs Sampling is particularly useful when we have more than one parameter, and the basic idea involves reducing our problem to a series of calculations involving one parameter at a time. In order to perform Gibbs Sampling, we need to find the conditional distributions

$$g(\mu | y, \gamma) \propto f(y | \mu, \gamma) f(\mu)$$

$$g(\gamma | y, \mu) \propto f(y | \mu, \gamma) f(\gamma)$$

We will use these conditional distributions to sample from the joint posterior $g(\mu, \gamma | y)$ according to the following algorithm:

- (1) Start with initial values $\mu^{(0)}, \gamma^{(0)}$.
- (2) Sample $\mu^{(t+1)} \sim g(\mu | y, \gamma = \gamma^{(t)})$.
- (3) Sample $\gamma^{(t+1)} \sim g(\gamma | y, \mu = \mu^{(t+1)})$.
- (4) Repeat many times.

It turns out that the resulting $\mu^{(0)}, \mu^{(1)}, \dots, \mu^{(N)}$ and $\gamma^{(0)}, \gamma^{(1)}, \dots, \gamma^{(N)}$ are samples from the joint posterior distribution $g(\mu, \gamma | Y)$, and we can use these sampled values to estimate quantities such as the posterior mean of each parameter $\hat{E}(\mu | y) = \frac{1}{N} \sum_{i=1}^N \mu^{(i)}$, $\hat{E}(\gamma | y) = \frac{1}{N} \sum_{i=1}^N \gamma^{(i)}$. Note that in practice we typically remove the initial iterations, known as the “burn-in” period: e.g., $\hat{E}(\mu | y) = \frac{1}{N-B} \sum_{i=B}^N \mu^{(i)}$.

2. Show that the conditional distributions $g(\mu | y, \gamma), g(\gamma | y, \mu)$ are proportional to $f(y | \mu, \gamma) f(\mu), f(y | \mu, \gamma) f(\gamma)$, respectively, as stated above.

ANSWER:

$$\begin{aligned}
g(\mu | y, \gamma) &= \frac{f(\mu, y, \gamma)}{f(y, \gamma)} \\
&\propto f(\mu, y, \gamma), \text{ since } f(y, \gamma) \text{ doesn't depend on } \mu \\
&= f(y | \mu, \gamma) f(\mu, \gamma) \\
&= f(y | \mu, \gamma) f(\mu) f(\gamma), \text{ since } \mu, \gamma \text{ independent} \\
&\propto f(y | \mu, \gamma) f(\mu), \text{ since } f(\gamma) \text{ doesn't depend on } \mu
\end{aligned}$$

A similar argument can be used to show $g(\gamma | y, \mu) \propto f(y|\mu, \gamma)f(\gamma)$.

3. Use this result to show that $\mu | y, \gamma \sim N\left(\frac{y\gamma + \frac{m}{v}}{\gamma + \frac{1}{v}}, \left[\gamma + \frac{1}{v}\right]^{-1}\right)$ and $\gamma | y, \mu \sim \text{Gamma}\left(\frac{1}{2} + a, \frac{1}{2}(y - \mu)^2 + b\right)$.

ANSWER:

$$\begin{aligned}
g(\mu | y, \gamma) &\propto f(y | \mu, \gamma)f(\mu) \\
&= \left[(2\pi)^{-\frac{1}{2}}\gamma^{\frac{1}{2}}e^{-\frac{1}{2}\gamma(y-\mu)^2}\right] \left[(2\pi v)^{-\frac{1}{2}}e^{-\frac{1}{2v}(\mu-m)^2}\right] \\
&\propto e^{-\frac{1}{2}\gamma(y-\mu)^2 - \frac{1}{2v}(\mu-m)^2} \\
&= e^{-\frac{1}{2}\gamma(y^2 - 2\mu y + \mu^2) - \frac{1}{2v}(\mu^2 - 2\mu m + m^2)} \\
&\propto e^{-\frac{1}{2}\gamma(-2\mu y + \mu^2) - \frac{1}{2v}(\mu^2 - 2\mu m)} \\
&= e^{-\frac{1}{2}\left[\mu^2\left(\gamma + \frac{1}{v}\right) - 2\mu\left(y\gamma + \frac{m}{v}\right)\right]} \\
&= e^{-\frac{1}{2}\left(\gamma + \frac{1}{v}\right)\left[\mu^2 - 2\mu\left(\frac{y\gamma + \frac{m}{v}}{\gamma + \frac{1}{v}}\right)\right]} \\
&\propto e^{-\frac{1}{2}\left(\gamma + \frac{1}{v}\right)\left[\mu^2 - 2\mu\left(\frac{y\gamma + \frac{m}{v}}{\gamma + \frac{1}{v}}\right) + \left(\frac{y\gamma + \frac{m}{v}}{\gamma + \frac{1}{v}}\right)^2\right]} \\
&= \propto e^{-\frac{1}{2\left(\gamma + \frac{1}{v}\right)^{-1}}\left[\mu - \left(\frac{y\gamma + \frac{m}{v}}{\gamma + \frac{1}{v}}\right)\right]^2} \\
\Rightarrow \mu | y, \gamma &\sim N\left(\frac{y\gamma + \frac{m}{v}}{\gamma + \frac{1}{v}}, \left[\gamma + \frac{1}{v}\right]^{-1}\right)
\end{aligned}$$

$$\begin{aligned}
g(\gamma | y, \mu) &\propto f(y | \mu, \gamma)f(\gamma) \\
&= \left[(2\pi)^{-\frac{1}{2}}\gamma^{\frac{1}{2}}e^{-\frac{1}{2}\gamma(y-\mu)^2}\right] \left[\frac{b^a}{\Gamma(a)}\gamma^{a-1}e^{-b\gamma}\right] \\
&\propto \gamma^{\frac{1}{2}}\gamma^{a-1}e^{-\frac{1}{2}\gamma(y-\mu)^2}e^{-b\gamma} \\
&= \gamma^{\frac{1}{2}+a-1}e^{-\frac{1}{2}\gamma(y-\mu)^2-b\gamma} \\
&= \gamma^{\frac{1}{2}+a-1}e^{-\gamma\left(\frac{1}{2}(y-\mu)^2+b\right)} \\
\Rightarrow \gamma | y, \mu &\sim \text{Gamma}\left(\frac{1}{2} + a, \frac{1}{2}(y - \mu)^2 + b\right)
\end{aligned}$$

4. Suppose that we choose the following hyperparameters for our prior distributions— $m = 0, v = 1, a = 1, b = 1$ —and that we observe $y = 2$. Write code to implement this Gibbs Sampler.

ANSWER:

```

# set up priors
m <- 0
v <- 1
a <- 1
b <- 1

# set up data
y <- 2

```



```

# choose starting values by randomly sampling from our priors
# (this is just one possible way to choose starting values)
# (it's also useful to try out a few different starting values)
set.seed(1)
mu <- rnorm(1, mean = m, sd = sqrt(v))
gam <- rgamma(1, shape = a, rate = b)

# set up empty vectors to store samples
mus <- c()
gams <- c()

# store starting values in vectors of samples
mus[1] <- mu
gams[1] <- gam

# choose number of iterations
# (we'll start with 100, but in practice you'd choose something much bigger)
N <- 100

# run through Gibbs Sampling for a total of N iterations
for(i in 2:N){
  # update mu
  m1 <- y*gam + m/v
  m2 <- gam + 1/v
  mu <- rnorm(n = 1, mean = (m1)/(m2), sd = sqrt(1/m2))

  # update gamma
  g1 <- 0.5 + a
  g2 <- 0.5*(y-mu)^2 + b
  gam <- rgamma(n = 1, shape = g1, rate = g2)

  # store new samples
  mus[i] <- mu
  gams[i] <- gam
}

```

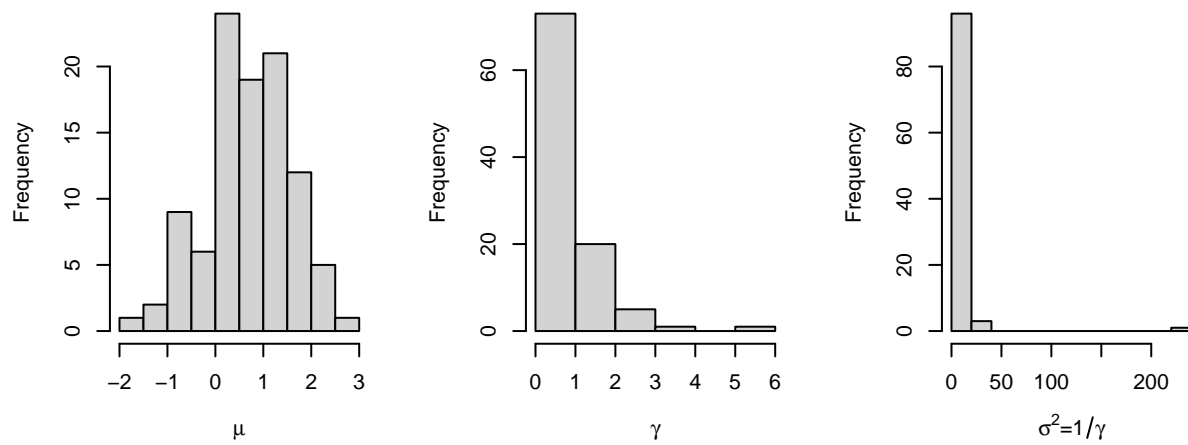
5. Look at a histogram of your posterior samples for μ, γ and $\sigma^2 = \frac{1}{\gamma}$.

ANSWER:

```

par(mfrow=c(1,3))
hist(mus, xlab = expression(mu), main = '')
hist(gams, xlab = expression(gamma), main = '')
hist(1/gams, xlab = expression(paste(sigma^2, '=', 1/gamma)), main = '')

```



6. Estimate the posterior mean and median of μ .

ANSWER:

```
# posterior mean
mean(mus)
```

```
## [1] 0.6840056
```

```
# posterior median
median(mus)
```

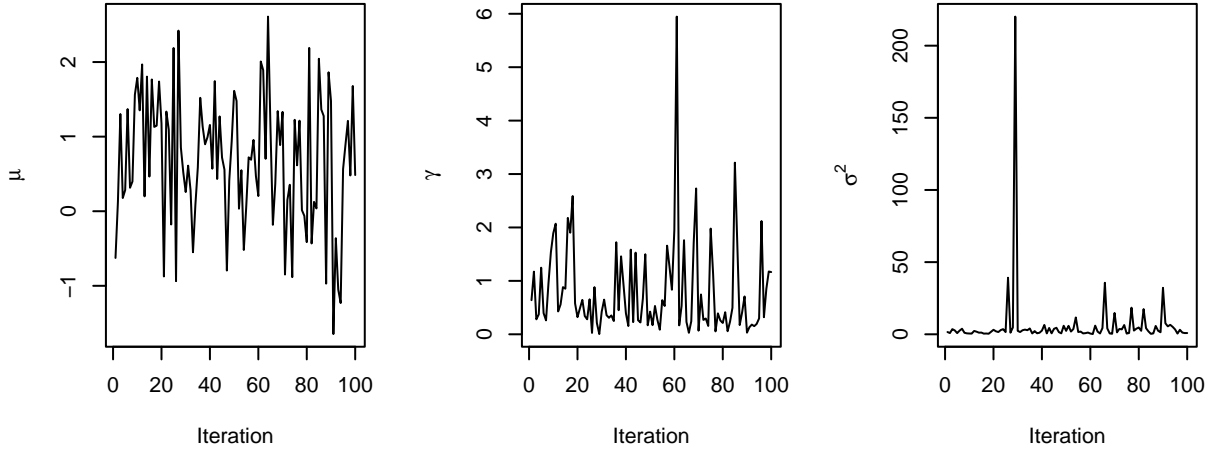
```
## [1] 0.6525553
```

8. Create a *trace plot* showing the behavior of the samples over the N iterations.

ANSWER:

```
iterations <- 1:N

par(mfrow=c(1,3))
plot(mus ~ iterations, xlab = 'Iteration', ylab = expression(mu), type = 'l')
plot(gams ~ iterations, xlab = 'Iteration', ylab = expression(gamma), type = 'l')
plot(1/gams ~ iterations, xlab = 'Iteration', ylab = expression(sigma^2), type = 'l')
```



9. As mentioned above, in practice we usually pick a burn-in period of initial iterations to remove. This decision is often motivated by the fact that, depending on your choice of starting value, it may take awhile for your chain of samples to look like it is “mixing” well. Play around with your choice of starting value above to see if you can find situations in which a burn-in period might be helpful.

Real World Application

Gibbs sampling has been used in the inference of population structure using multilocus genotype data (Jonathan K. Pritchard and Donnelly 2000). In other words, to infer the population of an individual using their genetic information. I will start defining some of the most important terms in the paper. A locus is the specific physical location of a gene or other DNA sequence on a chromosome, like a genetic street address. A genotype is the pair of alleles inherited from each parent for a particular gene, where an allele is a variation of a gene and a gene is the functional unit of heredity. For example, if a gene contains information on hair color one allele might code for brown and other for blond, a genotype would be the pair of alleles one coded for brown and the other for blond hair.

Assuming that each population is modeled by a characteristic set of allele frequencies, let X represent the genotypes of the sampled individuals, Z the populations of origin of individuals, and P the allele frequencies in all populations. Each allele at each locus in each genotype is an independent draw from the appropriate frequency distribution. This specifies the probability distribution $Pr(X | Z, P)$. Which is the probability that we draw a specific genotype given the population of origin of an individual and the allele frequency in a population.

Jonathan K. Pritchard, et. al used a Dirichlet distribution to model the probability that we observe specific genotypes

$$D \sim Dir(\alpha) = \frac{1}{Beta(\alpha)} \prod_{i=1}^J \theta_i^{\alpha_i - 1}, \text{ where } Beta(\alpha) = \frac{\prod_{i=1}^J \Gamma(\alpha_i)}{\Gamma(\sum_{i=1}^J \alpha_i)} \alpha = (\alpha_1, \dots, \alpha_J)$$

where D is a vector of J dimensions of the form $D = (\lambda_1, \lambda_2, \dots, \lambda_J)$, and $\alpha_i > 0$ and D belongs to the probability simplex where vectors are positive and the sum of their probability mass functions are always one. We use this distribution to model the allele frequencies $p = (p_1, p_2, \dots, p_J)$ knowing that these frequencies sum to 1.

The authors use a Dirichlet distribution given that it is a commonly used conjugate prior. Conjugate priors make the process of estimating a posterior easier given that the posterior will be in the same probability distribution family.

I will introduce some of their model notation. The authors assumed that each population is modeled by a characteristic set of allele frequencies. X denotes the genotypes of the sampled individuals, Z denotes the individual's unknown populations of origin, and P denotes the unknown allele frequency in all populations.

We adopt a Bayesian approach by specifying models priors $Pr(Z)$ and $Pr(P)$ for both Z and P . The authors used a Bayesian approach to estimate the quantities of interest. The authors specified model priors $Pr(Z)$ and $Pr(P)$ for both Z and P .

Having observed the genotypes, X , our knowledge of Z and P is given by the posterior distribution

$$Pr(Z, P | X) \propto Pr(Z)Pr(P)Pr(X | Z, P)$$

(1)

Where $Pr(Z)$ and $Pr(P)$ are the priors and $Pr(X | Z, P)$ is the likelihood function of a genotype given a population and allele frequency.

We can't compute this distribution exactly but we can obtain an approximate sample $(Z^{(1)}, P^{(1)}), (Z^{(2)}, P^{(2)}), \dots, (Z^{(M)}, P^{(M)})$ from $Pr(Z, P | X)$ from $Pr(Z, P | X)$ using Gibbs Sampling. Inference for Z and P may be based on summary statistics obtained from this sample. We will focus on a simpler model where each person is assumed to have originated in a single population.

Suppose we sample N individuals with paired chromosomes (diploid). We assume each individual originates in one of K populations, each with its own characteristic set of allele frequencies. The vectors X (observed genotypes), Z (populations of origin of the individuals), and P (the unknown allele frequencies in the populations). These vectors consist of the following elements.

$(x_l^{(i,1)}, x_l^{(i,2)})$ = genotype of the i th individual at the l th locus, where $i = 1, 2, \dots, N$ and $l = 1, 2, \dots, L$;

$Z^{(i)}$ = population from which individual i originated

p_{klj} = frequency of allele j at locus l in population k , where $k = 1, 2, \dots, K$ and $j = 1, 2, \dots, J_l$

where J_l is the number of distinct alleles observed at locus l , and these alleles are labeled $1, 2, \dots, J_l$.

Given the population of origin of each subject, the genotypes are assumed to be sampled by drawing alleles independently from the respective population frequency distributions $Pr(X | Z, P) = Pr(x_l^{(i,a)} = j | Z, P) = p_z(i)l_j$ (2) independently for each $x_l^{(i,a)}$ (allele for i th individual at l th locus). $p_z(i)l_j$ is the frequency of allele j at locus l in the population of origin of individual i and it's our likelihood function.

When defining the prior $P(Z)$ the authors assumed that before observing the genotypes we have no information about the population of origin of each subject. If the probability that individual i originated in population k is the same for all k , then

$$P(Z) = Pr(z^{(i)} = k) = \frac{1}{K} \quad (4)$$

independently for all individuals.

When defining the prior $Pr(P)$ the authors used the Dirichlet distribution to model the distribution on allele frequencies $p = (p_1, p_2, \dots, p_J)$. These frequencies have the property that they sum up to 1. This distribution specifies the probability of a particular set of allele frequencies p_{kl} for population k at locus l .

$$Pr(P) = p_{kl} \sim D = (\lambda_1, \lambda_2, \dots, \lambda_J) \quad (4)$$

in dependently for each k,l. The expected frequency of allele j for a population k is proportional to λ_j , and the variance of this frequency decreases as the sum of the λ_j pmf increases (as the sum of the pmf is closer to 1). The authors take $\lambda_1 = \lambda_2 = \dots = \lambda_{Jl} = 1.0$ which gives a uniform distribution on the allele frequencies allowing each λ_j to be equally likely.

Then the authors proceed to apply the Gibbs Sampling algorithm which can be described as follows.

Considering that our conditional distributions are

$$P(Z | X, P) \propto f(X | Z, P)f(Z)$$

And,

$$P(P | X, Z) \propto f(X | Z, P)f(P)$$

we can construct a Markov chain with stationary (target) multinomial distribution $Pr(Z, P | X)$ as follows:

Starting with the initial value $Z^{(0)}$ for Z (chosen randomly) we iterate over the following steps for $m=1,2,\dots$

Step 1. Sample $P^{(m)}$ from $Pr(P | X, Z^{(m-1)})$ Step 1. Sample $Z^{(m)}$ from $Pr(Z | X, P^{(m)})$

In step 1 we are estimating allele frequencies for each population given our sampled genotype and assuming that the population of origin of each individual is known. In step 2 we estimate the population of origin of each individual, given our sampled genotype and assuming that the population's allele frequencies are known. For sufficiently large m and c, $(Z^{(m)}, P^{(m)})$, $(Z^{(m+c)}, P^{(m+c)})$, $(Z^{(m+2c)}, P^{(m+2c)})$, ... will be approximately independent random samples from $Pr(Z, P | X)$

To be more specific, setting $\theta = (\theta_1, \theta_2) = (Z, P)$ and letting $\pi(Z, P) = Pr(Z, P | X)$, and starting with initial values $\theta^{(0)} = (\theta_1^{(0)}, \dots, \theta_r^{(0)})$, we iterate the following steps for $m = 1, 2, \dots$

Step 1. Sample $\theta_1^{(m)}$ from $\pi(\theta_1 | \theta_2^{(m-1)}, \theta_3^{(m-1)}, \dots, \theta_r^{(m-1)})$

Step 2. Sample $\theta_2^{(m)}$ from $\pi(\theta_2 | \theta_1^{(m)}, \theta_3^{(m-1)}, \dots, \theta_r^{(m-1)})$

Step r. Sample $\theta_r^{(m)}$ from $\pi(\theta_r | \theta_1^{(m)}, \theta_2^{(m)}, \dots, \theta_r^{(m-1)})$

We can show that if $\theta^{(m-1)} \sim \pi(\theta)$, then, $\theta^{(m)} \sim \pi(\theta)$, and so $\pi(\theta)$ is the stationary distribution of this Markov chain.

Discussion

Limitations, pro's, cons, alternatives.

References

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