May 27, 2022

Due 06/10/2022

## Problem 1.

In this problem, we will apply LDA to human ancestry discovery. In applications of population genetics, it is often useful to classify individuals in a sample into populations. An underlying assumption is that there are K ancestor populations, and each individual is an admixture of the ancestor populations. For each individual, we measure some genetic data about them, called genotype data. Each genotype is a locus that can take a discrete count value, individuals with similar genotypes are expected to belong to the same ancestor populations. We can derive the admixture coefficients  $\theta$  for each individual by running an LDA model, where the documents are individuals, and the words are the genotype.

Now let us assume the  $\beta$  matrix is known, and focus on variational inference of the population mixture  $\theta$  and the genotype ancestry (topic) assignments z for any individual. The variational distribution used to approximate the posterior (for each individual) is

$$q_i(\theta, z | \gamma, \phi) = q(\theta_i | \gamma_i) \prod_{n=1}^{N_i} q(z_{in} | \phi_{in}), \quad i = 1, \dots, M$$

The data matrix provides data about M = 100 individuals, each represented by a vocabulary of N = 200 genotype loci. This data has been preprocessed into a count matrix D of size  $M \times N$ .  $D_{ij}$  is the number of occurrences of genotype j in individual i, and  $\sum_j D_{ij}$  is the number of genotype loci in an individual. We learnt the LDA topic model over K = 4 ancestor populations, and the data matrix and the known  $\beta$  matrix can be downloaded from the course website. The value of  $\alpha$  is 0.1. You may use the following code to load the data in python.

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import pickle
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(1) Derive the variational inference update equations for estimating  $\gamma$  and  $\phi$ . [5pts] (2) For individual one, run LDA inference to find  $\phi$  for each genotype locus, store it as a matrix of size  $n_1 \times K$  (where  $n_1 : \sum_{1j} I(D_{1j} \neq 0), I(\cdot)$  being the indicator function, is the number of non-zero genotypes present in individual 1), and plot it as an image in your write up. Don't forget to show the colormap using the colorbar function to allow the colors in the image to be mapped to numbers! [10pts]

(3) We will construct a matrix  $\Theta$  of size  $M \times K$  to represent the ancestor assignments for all individuals in the population. For each individual *i*, run LDA inference to find  $\gamma$ , and store it as row of  $\Theta$ , i.e.  $\Theta_i = \gamma$ . Visualize  $\Theta$  as an image. [5pts] (4) Report the number of iterations needed to get to convergence for running inference on all M individuals (you may use absolute change less than 1*e*-3 as the convergence criteria). [5pts]

(5) Repeat the experiment for  $\alpha = 0.01, \alpha = 1, \alpha = 10$ , and for each of  $\alpha$ , visualize the  $\Theta$  matrix summarizing the ancestor population assignments for all individuals. Discuss the changes in the ancestor population assignments to the individuals as  $\alpha$  changes. Does the mean number of iterations required for convergence for inference change as  $\alpha$  changes? [10pts]

## Problem 2.

A simple first-order autoregressive process, AR(1), is defined as follows:

$$y_t = a + by_{t-1} + \epsilon_t, \quad t \ge 1, \quad y_0 = a,$$

where a and b are some constants and  $\epsilon_t \sim \mathcal{N}(0,1)$  is a Gaussian noise. AR(1) defines a distribution over sequence of discrete values,  $\{y_0, y_1, \ldots\}$  (to sample from this distribution, you can simply run the forward autoregressive recursion).

Derive a mean,  $\mu(t)$ , and a kernel, k(t, t'), functions for a Gaussian process that defines a distribution over functions, y(t), that coincides with AR(1) for all  $t \ge 1$ . [15pts]

## Problem 3.

Let  $G_0$  be a distribution over  $\Theta$  and let  $\alpha$  be a positive scalar. For any finite, measurable partition  $A_1, \ldots, A_r$  of  $\Theta$ , G is defined to be a Dirichlet process with base distribution  $G_0$  and concentration parameter  $\alpha_0$ , denoted by  $G \sim DP(\alpha_0 G_0)$ , if

$$(G(A_1),\ldots,G(A_r)) \sim \operatorname{Dir}(\alpha_0 G_0(A_1),\ldots,\alpha_0 G_0(A_r)).$$

Suppose we have observation  $X_1, \ldots, X_n$ , which we assume are drawn from G. Assuming we have the prior  $G \sim DP(\alpha_0 G_0)$ , derive the posterior distribution for  $G|X_1, \ldots, X_n$ . [15pts]

## Problem 4.

Consider the following DP mixture of Gaussian model in  $\mathbb{R}^2$ 

$$y_i | \theta_i \sim \mathcal{N}(\theta_i, \sigma_y^2 I_2)$$
  

$$\theta_i | G \sim G, \quad i = 1, \dots, n$$
  

$$G \sim \mathrm{DP}(\alpha_0 G_0)$$
  

$$G_0 = \mathcal{N}(0, \sigma_0^2 I_2)$$

Let  $\sigma_0 = 5, \sigma_y = 1$ . The data on the course website were generated by sampling from this model for a particular choice of  $\alpha_0$ .

(1) Generate 1000 samples from the model with  $\alpha_0 = 0.1, 1, 5, 10$  respectively, and show the scatter plots of your samples. [5pts]

(2) Download the data from the course website. Implement a collapsed Gibbs sampler for this model in which the  $\theta$  parameter have been integrated out. Fix  $\alpha_0 = 1$ . Run the sampler, show the scatter plots of the data and the samples of the unique  $\phi_i$ 's at the first few iterations (e.g., 1, 5, 10, 20, 50). When the sampler appears to have converged, use the subsequent samples to plot a histogram of the posterior distribution of the number of occupied tables. [15pts]

(3) Now place a vague gamma prior on  $\alpha_0$ . Again plot a histogram of the posterior distribution of the number of occupied tables. Also plot a histogram of the posterior distribution of  $\alpha_0$ . Explore the sensitivity of your results to the choice of parameters for the gamma distribution. [10pts]

[5pts]

(4) Interpret your results.

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