## Expectation Maximization

## Sorin Istrail

Department of Computer Science Brown University, Providence sorin@cs.brown.edu

31 October 2006

• In this lecture, we outline the haplotype phasing algorithm of Excoffier & Slatin [].

A 10

- In this lecture, we outline the haplotype phasing algorithm of Excoffier & Slatin [].
- Our domain is the set D = {x<sub>1</sub>, x<sub>2</sub>, ..., x<sub>n</sub>} of n samples, where each sample x<sub>i</sub> is drawn independently from a probability density p(x|θ).

- In this lecture, we outline the haplotype phasing algorithm of Excoffier & Slatin [].
- Our domain is the set D = {x<sub>1</sub>, x<sub>2</sub>, ..., x<sub>n</sub>} of n samples, where each sample x<sub>i</sub> is drawn independently from a probability density p(x|θ).
- The vector of parameters θ = (θ<sub>1</sub>,...,θ<sub>d</sub>) ∈ ℝ<sup>d</sup> is fixed but unknown and we would like to estimate θ from the sampled data.

- In this lecture, we outline the haplotype phasing algorithm of Excoffier & Slatin [].
- Our domain is the set D = {x<sub>1</sub>, x<sub>2</sub>, ..., x<sub>n</sub>} of n samples, where each sample x<sub>i</sub> is drawn independently from a probability density p(x|θ).
- The vector of parameters θ = (θ<sub>1</sub>,...,θ<sub>d</sub>) ∈ ℝ<sup>d</sup> is fixed but unknown and we would like to estimate θ from the sampled data.

## Recall that the log likelihood of $\theta$ with respect to $\mathcal D$ is given by the likelihood function

$$l(\theta) = \log p(\mathcal{D}|\theta) = \sum_{k=1}^{n} p(x_k|\theta)$$

Recall that the log likelihood of  $\theta$  with respect to  $\mathcal D$  is given by the likelihood function

$$l(\theta) = \log p(\mathcal{D}|\theta) = \sum_{k=1}^{n} p(x_k|\theta)$$

and the maximum log-likelihood estimate of  $\theta$ , denoted  $\hat{\theta}$ , is the value of  $\theta$  that maximizes  $p(\mathcal{D}|\theta)$ 

$$\hat{ heta} = rg\max_{ heta} \log p(\mathcal{D}| heta)$$

 In the missing data problem, we have both observed and missing features x<sub>k</sub> = {x<sub>kg</sub>, x<sub>kb</sub>}.

A 10

- In the missing data problem, we have both observed and missing features x<sub>k</sub> = {x<sub>kg</sub>, x<sub>kb</sub>}.
- For missing features, we need to perform statistical inference to infer the most likely value of the missing feature.

- In the missing data problem, we have both observed and missing features x<sub>k</sub> = {x<sub>kg</sub>, x<sub>kb</sub>}.
- For missing features, we need to perform statistical inference to infer the most likely value of the missing feature.
- Our central equation is the following.

$$Q(\theta; \theta^{i}) = \mathcal{E}_{\mathcal{D}_{b}}[Inp(\mathcal{D}_{g}, \mathcal{D}_{b}|\theta)|\mathcal{D}_{b}; \theta^{i}]$$

- In the missing data problem, we have both observed and missing features x<sub>k</sub> = {x<sub>kg</sub>, x<sub>kb</sub>}.
- For missing features, we need to perform statistical inference to infer the most likely value of the missing feature.
- Our central equation is the following.

$$Q(\theta; \theta^{i}) = \mathcal{E}_{\mathcal{D}_{b}}[Inp(\mathcal{D}_{g}, \mathcal{D}_{b}|\theta)|\mathcal{D}_{b}; \theta^{i}]$$

• Q is a function of  $\theta$  with  $\theta^i$  assumed fixed.

- In the missing data problem, we have both observed and missing features x<sub>k</sub> = {x<sub>kg</sub>, x<sub>kb</sub>}.
- For missing features, we need to perform statistical inference to infer the most likely value of the missing feature.
- Our central equation is the following.

$$Q(\theta; \theta^{i}) = \mathcal{E}_{\mathcal{D}_{b}}[Inp(\mathcal{D}_{g}, \mathcal{D}_{b}|\theta)|\mathcal{D}_{b}; \theta^{i}]$$

• Q is a function of  $\theta$  with  $\theta^i$  assumed fixed.

• Suppose we have an initial parameter vector  $\theta^i$  that is the current best estimate for the full distribution.

- Suppose we have an initial parameter vector  $\theta^i$  that is the current best estimate for the full distribution.
- We will use the candidate vector  $\theta^i$  to obtain an improved estimate  $\theta^{i+1}$ .

- Suppose we have an initial parameter vector  $\theta^i$  that is the current best estimate for the full distribution.
- We will use the candidate vector  $\theta^i$  to obtain an improved estimate  $\theta^{i+1}$ .
- Given  $\theta^i$ , the bound on the right hand side calculates the likelihood of the data including the unknown  $\mathcal{D}_b$  marginalized with respect to the current distribution (given by  $\theta^i$ ).

- Suppose we have an initial parameter vector  $\theta^i$  that is the current best estimate for the full distribution.
- We will use the candidate vector  $\theta^i$  to obtain an improved estimate  $\theta^{i+1}$ .
- Given  $\theta^i$ , the bound on the right hand side calculates the likelihood of the data including the unknown  $\mathcal{D}_b$  marginalized with respect to the current distribution (given by  $\theta^i$ ).
- The EM algorithm will select  $\theta^{i+1}$  as the best such candidate, i.e.,

$$\theta^{i+1} = \arg \max_{\theta} Q(\theta; \theta^i).$$

- Suppose we have an initial parameter vector  $\theta^i$  that is the current best estimate for the full distribution.
- We will use the candidate vector  $\theta^i$  to obtain an improved estimate  $\theta^{i+1}$ .
- Given  $\theta^i$ , the bound on the right hand side calculates the likelihood of the data including the unknown  $\mathcal{D}_b$  marginalized with respect to the current distribution (given by  $\theta^i$ ).
- The EM algorithm will select  $\theta^{i+1}$  as the best such candidate, i.e.,

$$\theta^{i+1} = \arg \max_{\theta} Q(\theta; \theta^i).$$

 Note that by choosing different initial candidates θ<sup>0</sup>, the output of the EM algorithm possibly varies.

- Note that by choosing different initial candidates  $\theta^0$ , the output of the EM algorithm possibly varies.
- Therefore, in order to compute a good candidate for a global optimal solution, the EM algorithm is often run many times from different initial values θ<sup>0</sup>.

- Note that by choosing different initial candidates  $\theta^0$ , the output of the EM algorithm possibly varies.
- Therefore, in order to compute a good candidate for a global optimal solution, the EM algorithm is often run many times from different initial values θ<sup>0</sup>.

The entire algorithm is given as follows.

Image: A = A

 The entire algorithm is given as follows.

**Input:** Observed data  $\mathcal{D}$ , initial estimate  $\theta^0$ , margine of error  $\epsilon$ **Output:** Maximum likelihood parameters  $\hat{\theta}$ 

1. Initialize 
$$i = 0$$
.  
While  $Q(\theta^{i+1}, \theta^i) - Q(\theta^i, \theta^{i-1}) \le \epsilon$   
(i) E-Step: Compute  $Q(\theta; \theta^i)$   
(ii) Max step:  $\theta^{i+1} = \arg \max_{\theta} Q(\theta; \theta^i)$   
(iii)  $i = i + 1$   
Return n  $\hat{\theta} \leftarrow \theta^{i+1}$ 

• In this problem, we have m different phenotype with frequencies  $P_1, P_2, \ldots P_m$ .

- **→** → **→** 

- In this problem, we have m different phenotype with frequencies  $P_1, P_2, \ldots P_m$ .
- Given observed samples  $n_1, n_2 \dots n_m$  of the phenotypes, the likelihood of the samples given the frequencies are given by the likelihood equation

- In this problem, we have m different phenotype with frequencies  $P_1, P_2, \ldots P_m$ .
- Given observed samples  $n_1, n_2 \dots n_m$  of the phenotypes, the likelihood of the samples given the frequencies are given by the likelihood equation

$$P(Samples|P_1,...,P_m) = \frac{n!}{n_1!n_2!\cdots n_m!}P_1^{n_1}P_2^{n_2}\dots P_m^{n_m}$$

- In this problem, we have m different phenotype with frequencies  $P_1, P_2, \ldots P_m$ .
- Given observed samples  $n_1, n_2 \dots n_m$  of the phenotypes, the likelihood of the samples given the frequencies are given by the likelihood equation

$$P(Samples|P_1,...,P_m) = \frac{n!}{n_1!n_2!\cdots n_m!}P_1^{n_1}P_2^{n_2}\dots P_m^{n_m}$$

• Under the assumption of Hardy-Weinberg equilibrium, or random mating, the likelihood of the haplotype frequencies given the phenotypic counts is given by

- In this problem, we have m different phenotype with frequencies  $P_1, P_2, \ldots P_m$ .
- Given observed samples  $n_1, n_2 \dots n_m$  of the phenotypes, the likelihood of the samples given the frequencies are given by the likelihood equation

$$P(Samples|P_1,...,P_m) = \frac{n!}{n_1!n_2!\cdots n_m!}P_1^{n_1}P_2^{n_2}\dots P_m^{n_m}$$

• Under the assumption of Hardy-Weinberg equilibrium, or random mating, the likelihood of the haplotype frequencies given the phenotypic counts is given by

$$I(P_1,...,P_h) = a_1 \cdot \prod_{j=1}^m (\sum_{i=1}^{C_j} P(h_{ik}h_{ie}))^{n_i}$$

- In this problem, we have m different phenotype with frequencies  $P_1, P_2, \ldots P_m$ .
- Given observed samples  $n_1, n_2 \dots n_m$  of the phenotypes, the likelihood of the samples given the frequencies are given by the likelihood equation

$$P(Samples|P_1,...,P_m) = \frac{n!}{n_1!n_2!\cdots n_m!}P_1^{n_1}P_2^{n_2}\dots P_m^{n_m}$$

• Under the assumption of Hardy-Weinberg equilibrium, or random mating, the likelihood of the haplotype frequencies given the phenotypic counts is given by

$$I(P_1,...,P_h) = a_1 \cdot \prod_{j=1}^m (\sum_{i=1}^{C_j} P(h_{ik}h_{ie}))^{n_i}$$