Assignment 1

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1 Part 1

1.1 Definitions

- *i*: Index over nucleotide sequences.
- P: Length of kmers to consider in the model.
- m: Indexes over all kmers of a given nucleotide sequence.
- j: Indexes over nucleotides of a given kmer m.
- k: Indexes over nucleotides (A, T, G, C).
- *l*: Indexes over models where 0 indicates model cooresponding to transcription factor bindind motif (foreground) and 1 indicates non-binding region (background).
- $X_{i,m,j,k}$: indicator variable that equals 1 if nucleotide j of kmer m of sequence i is equal to base k and equals 0 otherwise.
- $C_{i,m,l}$: Indicator variable that will equal 1 if kmer m of sequence i was drawn from model l and otherwise equals 0.
- X: The set of all $X_{i,m,i,k}$.
- C: The set of all $C_{i,m,l}$.
- $P(C_{i,m,l} = 1) = \lambda_l$ where l is either 0 or 1 indicating foreground or background.
- $P(X_{i,m,j,k} = 1 | C_{i,m,l} = 1) = \psi_{j,k}^{(l)}$, where $\sum_k \psi_{j,k}^{(l)} = 1$

This should define two 4 by P matries where each row sums to 1 and represents the probability of observing each nucleotide at a given position j of a kmer given kmer was selected from model l.

• The model parameters are denoted as $\theta = \{\lambda_l, \psi_{j,k}^{(l)}\}$

1.2 Complete log likelihood

$$Q(\theta|\mathbf{X}, \mathbf{C}) = \log(P(\mathbf{X}, \mathbf{C}|\theta)) \tag{1}$$

$$= \log \prod_{i} \prod_{m} P(X_{i,m}, C_{i,m} | \theta)$$
 (2)

$$= \log \prod_{i} \prod_{m} (\prod_{j} \prod_{k} \prod_{l} [P(C_{i,m,l} = 1 | \theta) P(X_{i,m,j,k} = 1 | C_{i,m,l} = 1, \theta)]^{X_{i,m,j,k}C_{i,m,l}})$$
(3)

$$= \log \prod_{i} \prod_{m} (\prod_{j} \prod_{k} \prod_{l} [\lambda_{l} \psi_{j,k}^{(l)}]) \tag{4}$$

$$= \sum_{i} \sum_{m} \sum_{j} \sum_{k} \sum_{l} X_{i,m,j,k} C_{i,m,l} \log[\lambda_l \psi_{j,k}^{(l)}]$$

$$\tag{5}$$

$$= \sum_{i} \sum_{m} \sum_{j} \sum_{k} \sum_{l} X_{i,m,j,k} C_{i,m,l} \log \lambda_{l} + \sum_{i} \sum_{m} \sum_{j} \sum_{k} \sum_{l} X_{i,m,j,k} C_{i,m,l} \log \psi_{j,k}^{(l)}$$
 (6)