

Robust

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August 4, 2016

1 Model

Let $\{y_i\}_{i=1}^n$ denote the coverage at position i for n loci across the chromosome/genome.

Let $\{s_i\}_{i=1}^n$ denote the segment number for each of those locations where $s_i \in \{1, \dots, S\}$ and S is the total number of segments.

$$p(y_i | s_i = j, w, \mu, \sigma^2) = (1 - \rho_j) \underbrace{\sum_{c=1}^C w_{0,c} f(y_i; \mu_{0,c}, \sigma_{0,c}^2)}_{\text{Common}} + \rho_j \underbrace{\sum_{k=1}^K w_{j,k} f(y_i; \mu_{j,k}, \sigma_{j,k}^2)}_{\text{Segment-specific}} \quad (1)$$

where f is the density function for the Normal distribution, $\sum_{k=1}^K w_{j,k} = 1$ and $\sum_{c=1}^C w_{0,c} = 1$.

2 EM Updates

2.1 E updates

For each data point y_i , ψ_i gives the probability it is in a common (rather than segment specific) component.

$$\psi_i = \frac{(1 - \rho_j) \sum_{c=1}^C w_{0,c} f(y_i; \mu_{0,c}, \sigma_{0,c}^2)}{(1 - \rho_j) \sum_{c=1}^C w_{0,c} f(y_i; \mu_{0,c}, \sigma_{0,c}^2) + \rho_j \sum_{k=1}^K w_{j,k} f(y_i; \mu_{j,k}, \sigma_{j,k}^2)} \quad (2)$$

For each data point y_i , $\phi_{i,c}$ gives the probability it is in component c given it's in a common component. $\nu_{i,k}$ gives the equivalent for segment specific components.

$$\phi_{i,c} = \frac{w_{0,c} f(y_i; \mu_{0,c}, \sigma_{0,c}^2)}{\sum_{c=1}^C w_{0,c} f(y_i; \mu_{0,c}, \sigma_{0,c}^2)},$$
$$\nu_{i,k} = \frac{w_{s_i,k} f(y_i; \mu_{s_i,k}, \sigma_{s_i,k}^2)}{\sum_{k=1}^K w_{s_i,k} f(y_i; \mu_{s_i,k}, \sigma_{s_i,k}^2)}$$

2.2 M updates

M update for ρ , a global common vs segment specific weighting:

$$\rho_j = 1 - \frac{\sum_{i:s_i=j} \psi_i}{n}$$

M updates for common component parameters:

$$w_{0,c} = \frac{\sum_{i=1}^n \psi_i \phi_{i,c}}{\sum_{i=1}^n \psi_i}$$

$$\mu_{0,c} = \frac{\sum_{i=1}^n \psi_i \phi_{i,c} y_i}{\sum_{i=1}^n \psi_i \phi_{i,c}}$$

$$\sigma_{0,c}^2 = \frac{\sum_{i=1}^n \psi_i \phi_{i,c} (y_i - \mu_{0,c})^2}{\sum_{i=1}^n \psi_i \phi_{i,c}}$$

M updates for segment specific component parameters:

$$w_{j,k} = \frac{\sum_{i:s_i=j} (1 - \psi_i) \nu_{i,k}}{\sum_{i:s_i=j} (1 - \psi_i)}$$

$$\mu_{j,k} = \frac{\sum_{i:s_i=j} (1 - \psi_i) \nu_{i,k} y_i}{\sum_{i:s_i=j} (1 - \psi_i) \nu_{i,k}}$$

$$\sigma_{j,k}^2 = \frac{\sum_{i:s_i=j} (1 - \psi_i) \nu_{i,k} (y_i - \mu_{j,k})^2}{\sum_{i:s_i=j} (1 - \psi_i) \nu_{i,k}}$$