

Text S1 Conditional-maximization steps of ECM algorithm

Take the expectation of $l_{c1}(\Psi)$ with respect to \mathbf{Z} , then we have the following Q function,

$$\begin{aligned} Q_1(\Psi; \hat{\Psi}^{(t)}) &= E_{\mathbf{Z}}[l_{c1}(\Psi)] \\ &= \sum_{j=1}^n \sum_{r=1}^m \sum_{r < s} E[I(Z_j = h_r h_s) | G_j, \hat{\Psi}^{(t)}] \ln [a\theta_r^2 I(r = s) + 2b\theta_r \theta_s I(r < s)] - n \ln T \\ &\doteq R - S \end{aligned}$$

There are $(m + 1)$ parameters to estimate (the parameter K and m haplotype frequencies) in the Q function and only one parameter is estimated in each CM-step. Further, note that when we maximize the Q function, there is a constraint condition that the sum of all the haplotype frequencies is equal to 1. So, there are m CM-steps in maximizing the Q function. Here, suppose that θ_1 is calculated by others. Let θ_x ($x = 2, 3, \dots, m$) denote the haplotype frequency which will be estimated in the x^{th} CM-step. As such, θ_1 can be estimated by $\hat{\theta}_1 = 1 - \hat{\theta}_2 - \dots - \hat{\theta}_x - \dots - \hat{\theta}_m = 1 - \hat{\theta}_x - A_1$. Then

$$\begin{aligned} T &= a \sum_{r=1}^m \theta_r^2 + 2b \sum_{r=1}^m \sum_{r < s} \theta_r \theta_s \\ &= 2(a-b)\theta_x^2 - 2(a-b)(1-A_1)\theta_x + \left[2(a-b) \sum_{r=2, r \neq x}^m \theta_r^2 \right. \\ &\quad \left. + 2(a-b) \sum_{r=2, r \neq x}^m \sum_{s \neq x, r < s} \theta_r \theta_s - 2(a-b)A_1 + a \right] \end{aligned}$$

To estimate the haplotype frequencies, take the first-order derivation of Q_1 with respect to θ_x and we can get the following equations,

$$\begin{aligned} \frac{\partial Q_1}{\partial \theta_x} &= \frac{\partial R}{\partial \theta_x} - \frac{\partial S}{\partial \theta_x} = 0 \\ \frac{\partial R}{\partial \theta_x} &= \sum_{j=1}^n \sum_{r=1}^m \frac{2^{I(r=x)}}{\theta_x} P(Z_j = h_x h_r | G_j, \hat{\Psi}^{(t)}) - \sum_{j=1}^n \sum_{r=1}^m \frac{2^{I(r=1)}}{\theta_1} P(Z_j = h_1 h_r | G_j, \hat{\Psi}^{(t)}) \\ \frac{\partial S}{\partial \theta_x} &= \frac{n}{T} [4(a-b)\theta_x - 2(a-b)(1-A_1)] \end{aligned}$$

Using the equations above, Equation (7) can be obtained.