

1. (a) *Haldane*: $r = (1 - e^{-2d})/2 \implies e^{-2d} = 1 - 2r \implies -2d = \log(1 - 2r) \implies d = -\frac{1}{2} \log(1 - 2r)$
- (b) *Kosambi*: $r = \tanh(2d)/2 = \frac{1}{2}[\exp(4d) - 1]/[\exp(4d) + 1] \implies (2r)[\exp(4d) + 1] = [\exp(4d) - 1] \implies \exp(4d)(1 - 2r) = 1 + 2r \implies \exp(4d) = (1 + 2r)/(1 - 2r) \implies d = \frac{1}{4} \log[(1 + 2r)/(1 - 2r)]$

r	cM	
	Haldane	Kosambi
0.01	1.0	1.0
0.05	5.3	5.0
0.10	11.2	10.1
0.20	25.5	21.2

2. Consider an interval of length d on a chromosome of length L .

Let $A = \{\text{no chiasma in interval}\}$ and $N = \text{total no. chiasmata on the chromosome}$.

$$\begin{aligned}
 \text{Map function: } M(d) &= [1 - \Pr(A)]/2 \\
 &= [1 - \sum_{n=0}^{\infty} \Pr(N = n \text{ and } A)]/2 \\
 &= [1 - \sum_{n=0}^{\infty} \Pr(N = n) \Pr(A|N = n)]/2 \\
 &= [1 - \sum_{n=0}^{\infty} p_n (1 - d/L)^n]/2
 \end{aligned}$$

In the case $p_n = e^{-2L} (2L)^n / n!$, we obtain:

$$\begin{aligned}
 M(d) &= [1 - \sum_{n=0}^{\infty} \frac{e^{-2L} (2L)^n}{n!} (1 - d/L)^n]/2 \\
 &= [1 - \sum_{n=0}^{\infty} \frac{1}{n!} e^{-2L} [2L(\frac{L-d}{L})]^n]/2 \\
 &= [1 - e^{-2d} \sum_{n=0}^{\infty} \frac{1}{n!} e^{-2(L-d)} [2(L-d)]^n]/2 \\
 &= [1 - e^{-2d}]/2
 \end{aligned}$$

3. Let the chromosome be represented by the interval $[0, L]$.

Consider any finite set of disjoint subintervals I_1, I_2, \dots, I_k . Let $n_i = \text{no. chiasmata in interval } I_i \text{ (on the four-strand bundle)}$ and $m_i = \text{no. crossovers in } I_i \text{ (on a random meiotic product)}$.

We wish to show that the $\{m_i\}$ are independent and $m_i \sim \text{Poisson}(|I_i|)$.

Since the chiasma process is a Poisson process, the $\{n_i\}$ are independent with $n_i \sim \text{Poisson}(2|I_i|)$.

Under no chromatid interference (NCI), the chiasmata are “thinned” independently with probability 1/2 to get the crossover process. Since the $\{n_i\}$ are independent and since the thinning in the disjoint subintervals are independent, it should be clear that the $\{m_i\}$ are independent.

So, we have $m_i|n_i \sim \text{Binomial}(n_i, 1/2)$ and $n_i \sim \text{Poisson}(2|I_i|)$, and we need to show $m_i \sim \text{Poisson}(|I_i|)$. Let $d = |I_i|$ and drop the subscripts i , to save a few keystrokes.

$$\begin{aligned}
\Pr(m = j) &= \sum_{k=j}^{\infty} \Pr(n = k \text{ and } m = j) \\
&= \sum_{k=j}^{\infty} \Pr(n = k) \Pr(m = j|n = k) \\
&= \sum_{k=j}^{\infty} \frac{e^{-2d}(2d)^k}{k!} \binom{k}{j} \left(\frac{1}{2}\right)^k \\
&= \sum_{k=j}^{\infty} \frac{e^{-2d} d^k}{k!} \left(\frac{k!}{j!(k-j)!}\right) \\
&= \left(\frac{e^{-d} d^j}{j!}\right) \sum_{k=j}^{\infty} \frac{e^{-d} d^{k-j}}{(k-j)!} \\
&= e^{-2d} d^j / j!
\end{aligned}$$

So $m_i \sim \text{Poisson}(|I_i|)$, and we’re done.

4. Let $e_i(v) = \Pr(\text{observe } O_i | v_i = v)$.

v	O					
	A	H	B	C	D	-
AA	$1 - \epsilon$	$\epsilon/2$	$\epsilon/2$	ϵ	$1 - \epsilon/2$	1
AB	$\epsilon/2$	$1 - \epsilon$	$\epsilon/2$	$1 - \epsilon/2$	$1 - \epsilon/2$	1
BA	$\epsilon/2$	$1 - \epsilon$	$\epsilon/2$	$1 - \epsilon/2$	$1 - \epsilon/2$	1
BB	$\epsilon/2$	$\epsilon/2$	$1 - \epsilon$	$1 - \epsilon/2$	ϵ	1

Note: Think of C = H or B, D = A or H, - = A or H or B, so, for example, $\Pr(C|v) = \Pr(H|v) + \Pr(B|v)$.

- 5.

$$\begin{aligned}
&B_i(\delta) < B_{i-1}(\delta) \\
\iff &\ln \text{RSS}_i + i\delta \left(\frac{\ln n}{n}\right) < \ln \text{RSS}_{i-1} + (i-1)\delta \left(\frac{\ln n}{n}\right) \\
\iff &\ln \left(\frac{\text{RSS}_{i-1}}{\text{RSS}_i}\right) > \delta \frac{\ln n}{n} \\
\iff &\frac{n}{2} \log_{10} \left(\frac{\text{RSS}_{i-1}}{\text{RSS}_i}\right) > \frac{\delta}{2} \log_{10} n
\end{aligned}$$

(and similarly for i vs $i + 1$). Thus, if $\frac{\text{RSS}_{i-1}}{\text{RSS}_i}$ is monotonically decreasing in i , then minimizing the BIC- δ criterion is equivalent to using $\frac{\delta}{2} \log_{10} n$ as a threshold for the conditional LOD score.