

Additional file 2. Expectations of $\sum_m \sigma_m^2$, $\sum_m \sigma_m^4$, $\sum_m \rho_m$, $\sum_m \rho_m^2$, $\sum_m \rho_m^2 / \sigma_m^2$ and $\sum_m 1/\sigma_m^2$

The elements were defined as $\tau = \sum_m \sigma_m^2$ and $\tau_2 = \sum_m \sigma_m^4$, $\rho_m = \frac{n_r \sigma_m^2}{\lambda_\beta + n_r \sigma_m^2}$ and $\sigma_m^2 = 2p_m(1 - p_m)$.

The parameter $\lambda_\beta = \sigma_e^2 / \sigma_\beta^2 = n_M \frac{\sigma_m^2 \sigma_e^2}{\sigma_a^2}$. Notations will be simplified forgetting the m indice.

For the U shaped distribution, following Goddard [16], the allele frequencies p_m were assumed to be distributed with a density $f(p) = k/2p(1 - p)$ with $k = 1/\log 2N_e$, and N_e the effective size of the reference population.

The demonstration largely uses decomposition in simple elements:

$$\lambda_\beta + n_r \sigma_m^2 = 2n_r [h + p(1 - p)] \quad \text{and} \quad h + p(1 - p) = -(p - \beta_1)(p - \beta_2)$$

With $h = \frac{\lambda_\beta}{2n_r}$, $\beta_1 = \frac{1-\omega}{2}$, $\beta_2 = \frac{1+\omega}{2}$ and $\omega = \sqrt{1 + 4h}$ (note that $\beta_1 + \beta_2 = 1$, $\beta_1 - \beta_2 = -\omega$, $\beta_1 \beta_2 = -h$)

The decompositions are $\frac{1}{h+p(1-p)} = -\frac{1}{(\beta_1 - \beta_2)} \left[\frac{1}{(p - \beta_1)} - \frac{1}{(p - \beta_2)} \right]$ and $\frac{1}{[(p - \beta_1)(p - \beta_2)]^2} = \frac{1}{(\beta_1 - \beta_2)^3} \left[-\frac{2}{p - \beta_1} + \frac{\beta_1 - \beta_2}{(p - \beta_1)^2} + \frac{2}{p - \beta_2} + \frac{\beta_1 - \beta_2}{(p - \beta_2)^2} \right]$ giving

$$\int \frac{1}{h+p(1-p)} dp = \frac{1}{(\beta_1 - \beta_2)} \left[\log \left(\frac{p - \beta_2}{p - \beta_1} \right) \right] = A \quad [1]$$

$$\int \frac{1}{[h+p(1-p)]^2} dp = \frac{1}{(\beta_1 - \beta_2)^3} \left[2 \log \left(\frac{p - \beta_2}{p - \beta_1} \right) - (\beta_1 - \beta_2) \left(\frac{1}{p - \beta_1} + \frac{1}{p - \beta_2} \right) \right] = B \quad [2]$$

$$1) \quad E[\sigma^2] = E[2p(1 - p)]$$

a- U shaped distribution

$$E[\sigma^2] = \int_{1/N_e}^{1-1/N_e} 2p(1 - p) f(p) dp = k(1 - 2/N_e) \text{ following [16] about integration bounds.}$$

When N_e is large: $E[\sigma^2] = k$

b- Uniform distribution

$$E[\sigma^2] = \int_0^1 2p(1 - p) u(p) dp = 2 \left[\frac{p^2}{2} \right]_0^1 - 2 \left[\frac{p^3}{3} \right]_0^1 = \frac{1}{3}$$

$$2) \quad E[\sigma^4] = E[[2p(1 - p)]^2]$$

a- U shaped distribution

$$E[\sigma^4] = \int_{1/N_e}^{1-1/N_e} [2p(1 - p)]^2 f(p) dp = 2k \int_{1/N_e}^{1-1/N_e} p(1 - p) dp = 2k \left[\frac{1}{6} - \frac{1}{N_e^2} + \frac{2}{3N_e^3} \right]$$

When N_e is large: $E[\sigma^4] = k/3$

b- Uniform distribution

$$E[\sigma^4] = \int_0^1 [2p(1-p)]^2 u(p) dp = 4 \left(\left[\frac{p^3}{3} \right]_0^1 - 2 \left[\frac{p^4}{4} \right]_0^1 + \left[\frac{p^5}{5} \right]_0^1 \right) = \frac{2}{15}$$

$$3) E[\rho] = E \left[\frac{n_r \sigma^2}{\lambda_\beta + n_r \sigma^2} \right] = E \left[\frac{p(1-p)}{h+p(1-p)} \right]$$

a- U shaped distribution

$$E[\rho] = E \left[\frac{n_r \sigma^2}{\lambda_\beta + n_r \sigma^2} \right] = E \left[\frac{p(1-p)}{h+p(1-p)} \right] = \int_{1/N_e}^{1-1/N_e} \frac{p(1-p)}{h+p(1-p)} f(p) dp = \frac{k}{2} \int_{1/N_e}^{1-1/N_e} \frac{1}{h+p(1-p)} dp$$

$$\text{Using [1], } E[\rho] = \frac{k}{2(\beta_1 - \beta_2)} \log \left(\frac{(1-1/N_e - \beta_2)}{(1-1/N_e - \beta_1)} \times \frac{(1/N_e - \beta_1)}{(1/N_e - \beta_2)} \right)$$

$$\text{When } N_e \text{ is large: } E[\rho] = \frac{k}{-2\omega} \log \left(\left(\frac{1-\omega}{1+\omega} \right)^2 \right) = \frac{k}{\omega} \log \left(\left| \frac{1+\omega}{1-\omega} \right| \right) = \frac{k}{\omega} \theta$$

b- Uniform distribution

$$E[\rho] = \int_0^1 \frac{p(1-p)}{h+p(1-p)} dp$$

$$\text{From [1], } \int \frac{p(1-p)}{h+p(1-p)} dp = 1 - hA \text{ and } E[\rho] = 1 - h \frac{1}{(\beta_1 - \beta_2)} \left[\log \left(\frac{1-\beta_2}{1-\beta_1} \times \frac{\beta_1}{\beta_2} \right) \right]$$

$$E[\rho] = 1 - \frac{h}{-\omega} \log \left(\left(\frac{1-\omega}{1+\omega} \right)^2 \right) = 1 - \frac{2h}{\omega} \theta$$

4) $E[\rho^2]$

a- U shaped distribution

$$E[\rho^2] = \int_{1/N_e}^{1-1/N_e} \left(\frac{p(1-p)}{h+p(1-p)} \right)^2 f(p) dp = \frac{k}{2} \int_{1/N_e}^{1-1/N_e} \frac{p(1-p)}{(h+p(1-p))^2} dp$$

$$\text{From [1] and [2], } \int \frac{p(1-p)}{(h+p(1-p))^2} dp = A - hB$$

$$\frac{2}{k} E[\rho^2] = \frac{1}{(\beta_1 - \beta_2)} \left[\log \left(\frac{p-\beta_2}{p-\beta_1} \right) \right]_{1/N_e}^{1-1/N_e} - h \left\{ \frac{1}{(\beta_1 - \beta_2)^3} \left[2 \log \left(\frac{p-\beta_2}{p-\beta_1} \right) - (\beta_1 - \beta_2) \left(\frac{1}{p-\beta_1} + \frac{1}{p-\beta_2} \right) \right]_{1/N_e}^{1-1/N_e} \right\}$$

$$\text{We used } \left[\log \left(\frac{p-\beta_2}{p-\beta_1} \right) \right]_{1/N_e}^{1-1/N_e} = \log \left(\frac{(1-1/N_e - \beta_2)}{(1-1/N_e - \beta_1)} \times \frac{(1/N_e - \beta_1)}{(1/N_e - \beta_2)} \right) \sim \log \left(\left(\frac{\beta_1}{\beta_2} \right)^2 \right) = -2\theta$$

$$\text{and } \left[\left(\frac{1}{p-\beta_1} \right) \right]_{1/N_e}^{1-1/N_e} = \frac{2/N_e - 1}{(1-1/N_e - \beta_1)(1/N_e - \beta_1)} \sim \frac{1}{\beta_1 \beta_2} = -\frac{1}{h}$$

$$\frac{2}{k} E[\rho^2] = \frac{1}{(\beta_1 - \beta_2)} (-2\theta) - h \left\{ \frac{1}{(\beta_1 - \beta_2)^3} \left[2(-2\theta) - (\beta_1 - \beta_2) 2 \left(-\frac{1}{h} \right) \right] \right\}$$

$$E[\rho^2] = \frac{k}{2} \left(2\theta \left[-\frac{1}{(\beta_1 - \beta_2)} + \frac{2h}{(\beta_1 - \beta_2)^3} \right] - \frac{2}{(\beta_1 - \beta_2)^2} \right) = -\frac{k}{\omega^2} + \frac{k\theta}{\omega} \left[1 - \frac{2h}{\omega^2} \right] = \frac{k}{\omega^2} \left[\theta \left(\omega - \frac{2h}{\omega} \right) - 1 \right]$$

b- Uniform distribution

$$E[\rho^2] = \int_0^1 \left(\frac{p(1-p)}{h+p(1-p)} \right)^2 dp = \int_0^1 \left(1 - \frac{2hp(1-p)}{(h+p(1-p))^2} + \frac{h^2}{(h+p(1-p))^2} \right) dp$$

From [1] and [2], $E[\rho^2] = 1 - 2h(A - hB) + h^2B = (1 + h)^2B - 2hA$

$$E[\rho^2] = (1 + h)^2 \left\{ \frac{1}{(\beta_1 - \beta_2)^3} \left[2 \log \left(\frac{p - \beta_2}{p - \beta_1} \right) - (\beta_1 - \beta_2) \left(\frac{1}{p - \beta_1} + \frac{1}{p - \beta_2} \right) \right]_0^1 \right\} - 2h \frac{1}{(\beta_1 - \beta_2)} \left[\log \left(\frac{p - \beta_2}{p - \beta_1} \right) \right]_0^1$$

We used $\left[\log \left(\frac{p - \beta_2}{p - \beta_1} \right) \right]_0^1 \sim -2\theta$ and $\left[\left(\frac{1}{p - \beta_1} \right) \right]_0^1 = -\frac{1}{h}$

$$E[\rho^2] = (1 + h)^2 \left\{ \frac{1}{(\beta_1 - \beta_2)^3} \left[-4\theta - (\beta_1 - \beta_2) \left(-\frac{2}{h} \right) \right] \right\} - 2h \frac{1}{(\beta_1 - \beta_2)} [-2\theta] = 4\theta \left(\frac{(1+h)^2}{\omega^3} - \frac{h}{\omega} \right) + 2 \left(\frac{(1+h)^2}{h\omega^2} \right) = \frac{4\theta}{\omega} \left(\left(\frac{1+h}{\omega} \right)^2 - h \right) + \frac{2}{h} \left(\frac{1+h}{\omega} \right)^2 = \left(\frac{4\theta}{\omega} + \frac{2}{h} \right) \left(\frac{1+h}{\omega} \right)^2 - \frac{4\theta h}{\omega}$$

5) $E[\rho^2/\sigma^2]$

a- U shaped distribution

$$E[\rho^2/\sigma^2] = \int_{1/N_e}^{1-1/N_e} \frac{\left(\frac{p(1-p)}{h+p(1-p)} \right)^2}{2p(1-p)} f(p) dp = \frac{k}{4} \int_{1/N_e}^{1-1/N_e} \left(\frac{1}{h+p(1-p)} \right)^2 dp$$

$$\text{From [2]} \quad E[\rho^2/\sigma^2] = \frac{k}{4} \frac{1}{(\beta_1 - \beta_2)^3} \left[2 \log \left(\frac{p - \beta_2}{p - \beta_1} \right) - (\beta_1 - \beta_2) \left(\frac{1}{p - \beta_1} + \frac{1}{p - \beta_2} \right) \right]_{1/N_e}^{1-1/N_e}$$

When N_e is large and using $\left[\log \left(\frac{p - \beta_2}{p - \beta_1} \right) \right]_0^1 \sim -2\theta$ and $\left[\left(\frac{1}{p - \beta_1} \right) \right]_0^1 = -\frac{1}{h}$

$$E[\rho^2/\sigma^2] = \frac{k}{4} \frac{1}{(\beta_1 - \beta_2)^3} \left[-4\theta - (\beta_1 - \beta_2) \left(-\frac{2}{h} \right) \right] = \frac{k}{2h\omega^2} + \frac{k\theta}{\omega^3} = \frac{k}{2\omega^3} \left[\frac{\omega}{h} + 2\theta \right]$$

b- Uniform distribution

$$E[\rho^2/\sigma^2] = \int_0^1 \frac{\left(\frac{p(1-p)}{h+p(1-p)} \right)^2}{2p(1-p)} dp = \frac{1}{2} \int_0^1 \frac{p(1-p)}{(h+p(1-p))^2} dp \quad \text{which was found in the } E[\rho^2] \text{ for the U-shaped distribution.}$$

$$\text{Thus } E[\rho^2/\sigma^2] = \frac{1}{\omega^2} \left[\theta \left(\omega - \frac{2h}{\omega} \right) - 1 \right]$$

6) $E[1/\sigma^2]$

a- U shaped distribution

$$E[1/\sigma^2] = \int_{1/N_e}^{1-1/N_e} \frac{1}{2p(1-p)} f(p) dp = \frac{k}{4} \int_{1/N_e}^{1-1/N_e} \left(\frac{1}{p(1-p)} \right)^2 dp = \frac{k}{4} \int_{1/N_e}^{1-1/N_e} \left(\frac{1}{p^2} + \frac{1}{(1-p)^2} + \frac{2}{p} + \frac{2}{1-p} \right) dp$$

$$\text{Thus } E[1/\sigma^2] = \frac{k}{4} \left[2 \log \left(\frac{p}{1-p} \right) - \frac{1}{p} + \frac{1}{1-p} \right]_{1/N_e}^{1-1/N_e} = \frac{k}{4} \left[2 \log(N_e - 1) + 2 \frac{N_e(N_e - 2)}{N_e - 1} \right]$$

b- Uniform distribution

$$\text{Keeping the } 1/N_e, 1 - 1/N_e \text{ bounds, } E[1/\sigma^2] = \int_{1/N_e}^{1-1/N_e} \frac{1}{2p(1-p)} dp = \log(N_e - 1)$$