

Lecture 10. RGD-mutation equilibrium. RGD-migration equilibrium.

3.5 RGD-mutation equilibrium

Population heterozygosity in a dynamic equilibrium

non-reversible mutation generates new alleles

RGD eliminates alleles due to genetic sampling

IBD in the presence of mutation = no mutation

since MRCA (most recent common ancestor)

Neutral mutation rate μ per gene per generation

$$F_t = (1 - \mu)^2 \left(\frac{1}{2N_e} + \left(1 - \frac{1}{2N_e}\right) F_{t-1} \right)$$

equilibrium solution $\hat{F} = \frac{1}{1+\theta}$, where $\theta = 4N_e\mu$

Infinite-alleles mutation model (IAM)

each mutation produces a novel allele

Identity by descent = identity by state = homozygosity

$$\text{average heterozygosity } \hat{H} = 1 - \hat{F} = \frac{\theta}{1+\theta}$$

Effective number of alleles

Number k_e of hypothetical alleles with equal frequencies

resulting in the same as observed $H = 1 - p_1^2 + \dots + p_k^2$

$$1 - H = \left(\frac{1}{k_e}\right)^2 + \dots + \left(\frac{1}{k_e}\right)^2 = \frac{1}{k_e}$$

neutral mutation equilibrium $k_e = \theta + 1$ under IAM

Ex 9: mutation rate estimation

Fig 2.9, p.55 allozyme alleles in *Drosophila*

$$N_e = 10^6, \hat{H} = 0.14, \hat{\theta} = \frac{\hat{H}}{1-\hat{H}} = 0.163$$

$$k_e = 1.163, \hat{\mu} = 4 \cdot 10^{-8}$$

IAM underestimates μ if based on electrophoresis H

usually $\mu = 10^{-4} - 10^{-6}$ mut. per gene per generation

Ewens sampling formula

gives a rough estimate of θ based on the sample size

and the observed number of alleles

Average number of IAM alleles in a sample of size n

$$E(k) = 1 + \frac{\theta}{\theta+1} + \frac{\theta}{\theta+2} + \dots + \frac{\theta}{\theta+n-1}$$

diminishing return in new alleles when n increases

$E(k) \approx 1$ for small θ and $E(k) \approx n$ for large θ

3.6 RGD-migration equilibrium

RGD reduces H due to random allele fixation

migration increases H due to gene inflow

Assumptions

subpopulation $EPS = N_e$

immigration rate m , no mutation

$$\boxed{\text{Stable equilibrium: } \hat{F} = \frac{1}{1+4Nm}, \hat{H} = \frac{4Nm}{1+4Nm}}$$

Similar to RGD-mutation equilibrium only $m \gg \mu$

	Nm	\hat{F}	\hat{H}
complete genetic isolation	0	1	0
one migrant every fourth generation	0.25	0.50	0.50
one migrant every second generation	0.5	0.33	0.67
one migrant every generation	1	0.20	0.80
two migrants every generation	2	0.11	0.89

Ex 10: estimation of $N_e m$

Fig 5.18, p. 197: \hat{F}_{ST} for 61 genes (61 scenarios)

in natural populations of *D.melanogaster*

average $\hat{F}_{ST} = 0.16$, leading to $Nm = 1.3$

$$\boxed{\text{If both mutation and migration occur } \hat{F} = \frac{1}{1+4N(\mu+m)}}$$

Ex 11: melanic forms in moth

Two moth species in the Liverpool-Manchester area

Fig 5.19, p. 199: frequency of melanic forms

A: high migration rate, B: low mobility

Literature:

1. D.L.Hartl, A.G.Clarc. Principle of population genetics. Sinauer Associates, 2007.
2. R.Nielson, M. Statkin. An introduction to population genetics: theory and applications, Sinauer Associates. 2013.