

§ 2.3 Mutation and Drift

- Drift removes variation
- Mutation adds variation in terms of selection
- Let $u =$ mutation rate to neutral alleles
 \downarrow $P_r(\text{mutation at a single gamete at a given locus})$
- A mutation that occurs is assumed to produce a unique allele.

Gen-1	A, A, A, A, A,
Gen-2	A, A ₂ , A, A, A,
Gen-3	A, A ₂ , A ₂ , A ₃ , A,
⋮	

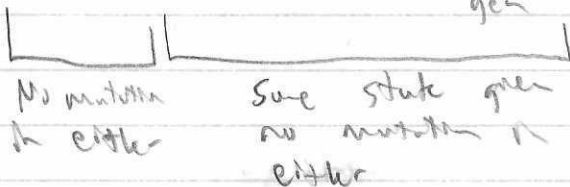
\uparrow This assumption is called "infinite alleles model"

2.1) Size = 3000, # 1-step away is $3000 \cdot 3$ (change of nucleotide)
 # 2-steps away is $3000 \cdot 2 \cdot 4 \cdot 4 \cdot 4$ (change 2 nucleotides)
 = 80,973,000

$$P' = (1-u)^2 \left[\frac{1}{2N} + \left(1 - \frac{1}{2N}\right) P \right] \quad] \quad P_r(\text{same state | different origin})$$

Next
gen

Current
gen



• Goal: Approximation to change a heterozygosity

Note, $u \approx 10^{-5}$, N is very large

so $(1-u) \approx 1 - 2u$ and $\frac{u}{2} \approx 0$

↑
Taylor
series

$$\begin{aligned}
 A' &\approx (1-2u) \left[\frac{1}{2N} + \left(1 - \frac{1}{2N}\right) A \right] \\
 &= \frac{1}{2N} - \underbrace{\frac{2u}{2N}}_0 + \left(1 - 2u - \frac{1}{2N} + \frac{2u}{2N}\right) A \\
 &= \frac{1}{2N} + \left(1 - 2u - \frac{1}{2N}\right) A \\
 &= \frac{1}{2N} + \left(1 - \frac{1}{2N}\right) A - 2u A
 \end{aligned}$$

$$\begin{aligned}
 H' = 1 - A' &\approx 1 - \frac{1}{2N} - \left(1 - \frac{1}{2N}\right) A + 2u A \\
 &= 1 - \frac{1}{2N} - \left(1 - \frac{1}{2N}\right) (1-H) + 2u(1-H) \\
 &= \left(1 - \frac{1}{2N}\right) H + 2u(1-H)
 \end{aligned}$$

$$\Delta H = H' - H \approx \underbrace{-\frac{1}{2N} H}_{\text{effect of drift}} + \underbrace{2u(1-H)}_{\text{effect of mutation}}$$

When $\Delta H = 0$, mutation and drift offset, and we have

$$\begin{aligned}
 \frac{1}{2N} H &= 2u(1-H) \Rightarrow \left(\frac{1}{2N} + 2u\right) H = 2u \\
 \Rightarrow H &= \frac{2u}{\frac{1}{2N} + 2u} = \frac{4Nu}{1 + 4Nu}
 \end{aligned}$$

$$H + Ht = t$$

$$H = t(1-H)$$

$$t = \frac{H}{1-H}$$

• When mutation and drift offset,

$$H \approx \frac{4Nu}{1 + 4Nu} \quad (\text{in humans } 4Nu \approx 0.05)$$

• Back to interplay

$$\Delta H \approx -\frac{1}{2N}H + \underbrace{2u(1-H)}$$

↳ lets look at this

Suppose infinite population and we have mutation

$$H' = H + (1-H)[1 - (1-u)^2]$$

In infinite pop, every parent is unique, so

$$H' = P.(\text{parents differ by state}) +$$

$$P.(\text{parents same by state}) \cdot P.(\text{at least one mutation})$$

$$\approx H + (1-H)(1 - (1-2u))$$

$$\approx H + 2u(1-H)$$

$$\Rightarrow \Delta H = 2u(1-H)$$

• $4Nu \uparrow \Rightarrow H \uparrow$ so u dominating
 $4Nu \downarrow \Rightarrow H \downarrow$ so drift dominating