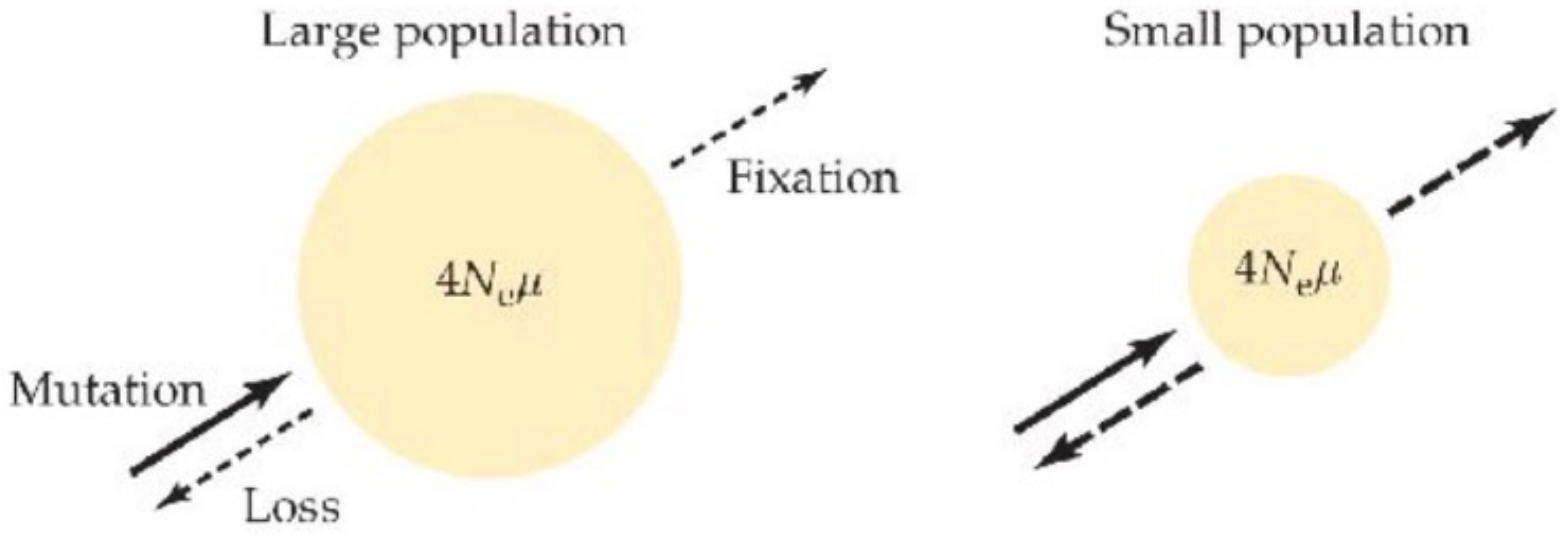
Population Genomics

Week 3 PSMC

Overview

- Wrap up of Coalescense and Migration ~5 min
- Wrap up to Tajimas D ~5 min
- Introduction to PSMC (again) ~5 min
- Introduction to the Exercise ~5 min
- Exercise ~40 min
- BREAK = 15min
- Exercise ~30 min
- Class Wrap up ~ 30 min

Drift and mutation equilibrium



Drift and mutation equilibrium

$$\mathbb{P}(\text{coal. in t+1 \& no mutations}) \approx \frac{1}{2N}\left(1-\frac{1}{2N}\right)^{t}(1-\mu)^{2t} \quad (4.9)$$

$$\approx \frac{1}{2N}e^{-t/(2N)}e^{-2\mu t} \quad (4.10)$$

$$= \frac{1}{2N}e^{-t(2\mu+1/(2N))} \quad (4.11)$$

Then we can approximate the summation by an integral, giving us:

$$\frac{1}{2N}\int_0^\infty e^{-t(2\mu+1/(2N))}dt = \frac{1/(2N)}{1/(2N)+2\mu} \quad (4.12)$$

$$H = \frac{2\mu}{1/(2N)+2\mu} = \frac{4N\mu}{1+4N\mu} \quad (4.13)$$

$$\theta = 4N\mu \quad (4.14)$$

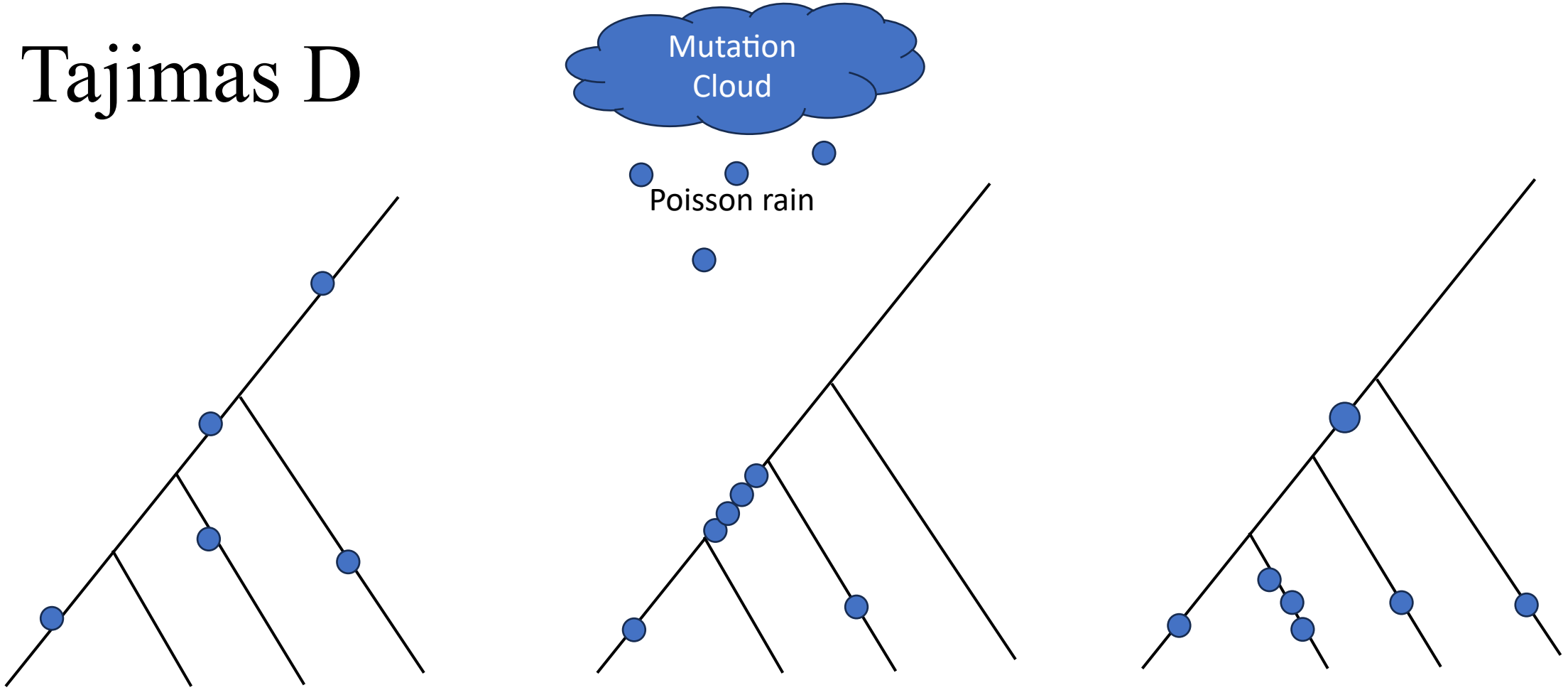
Probability of homozygosity (IBD):

$$\frac{1/2N}{1/2N+2\mu} = \frac{1}{1+4N\mu} = \frac{1}{1+\theta}$$

Probability of heterozygosity:

$$\frac{2\mu}{1/2N+2\mu} = \frac{4N\mu}{1+4N\mu} = \frac{\theta}{1+\theta}$$

Tajimas D



Tajimas D ~ 0

Neutral

Tajimas D < 0

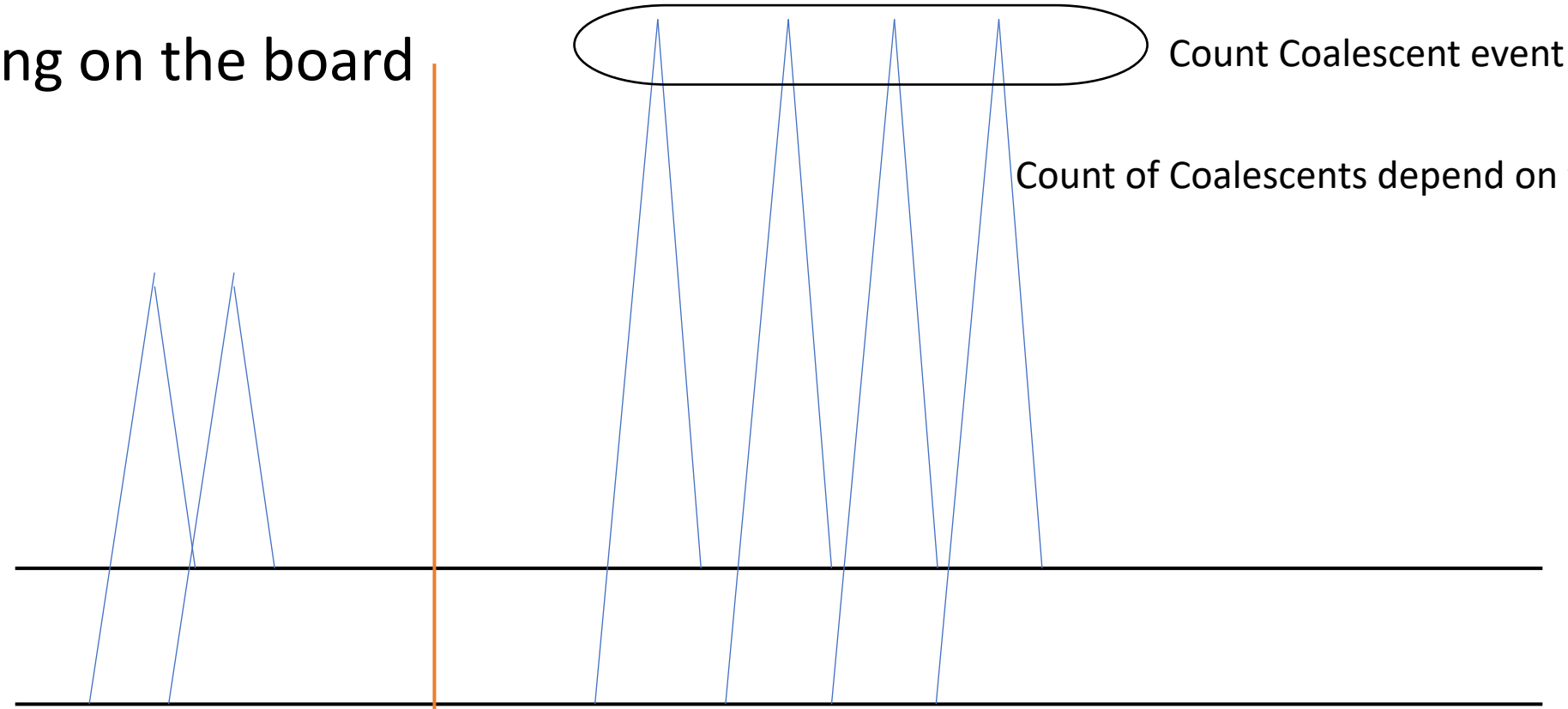
After a Sweep

Tajimas D > 0

Balancing Selection

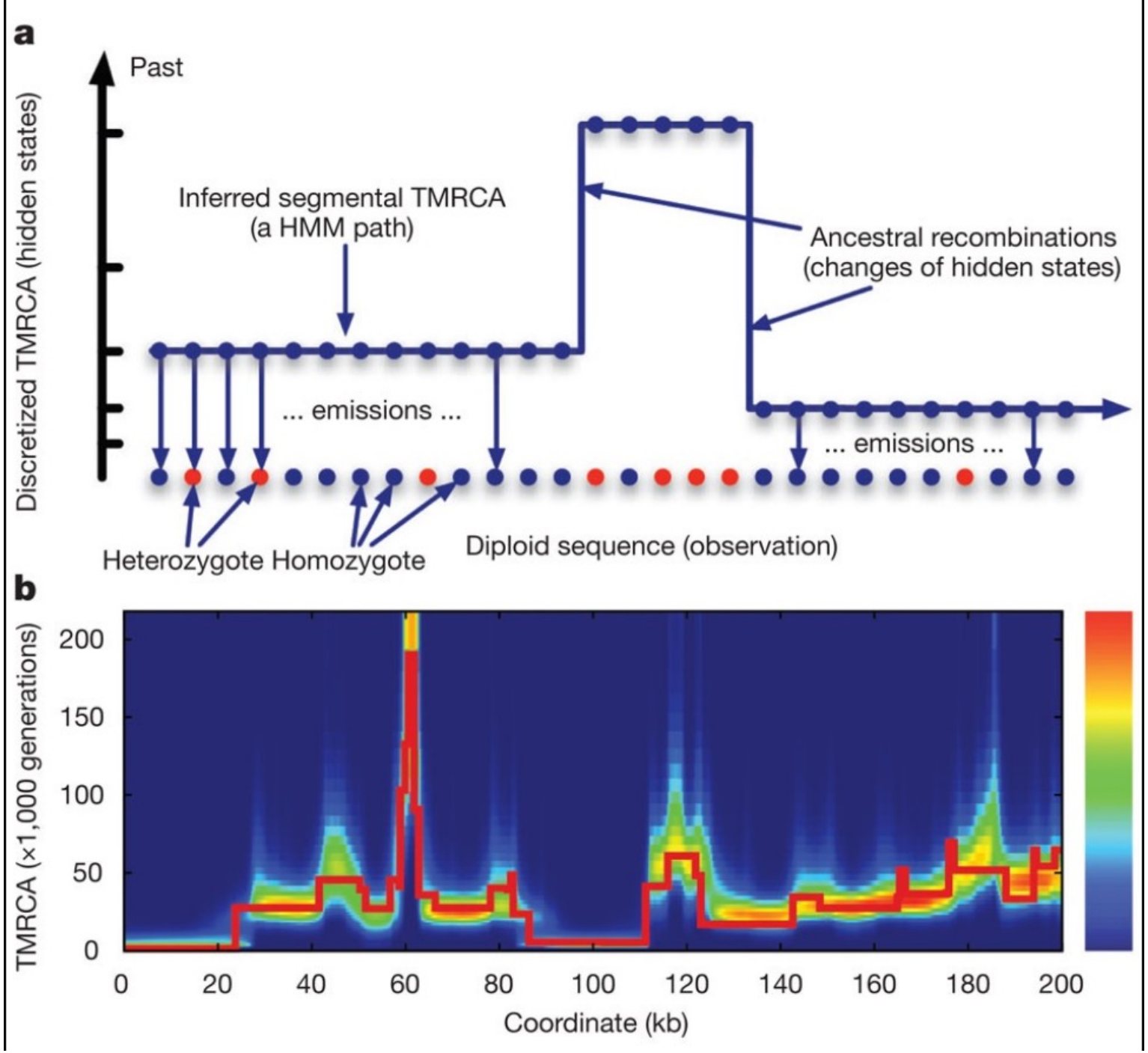
PSMC

Drawing on the board



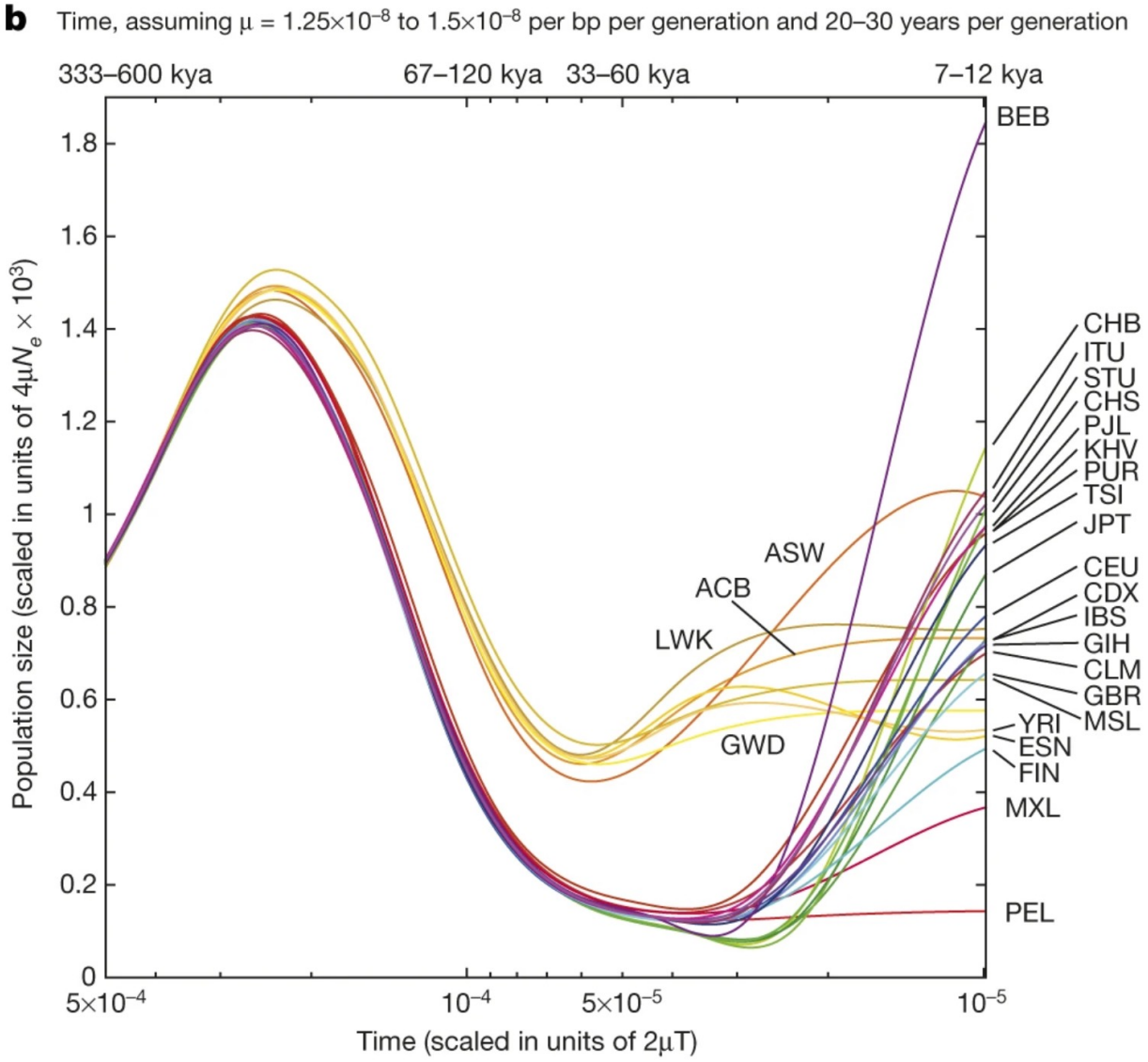
Coalescent time depends on mutation rate

Recombination


a
Past
Discretized TMRCA (hidden states)
Inferred segmental TMRCA
(a HMM path)
Ancestral recombinations
(changes of hidden states)
... emissions ...
... emissions ...
Heterozygote
Homozygote
Diploid sequence (observation)
b
TMRCA (×1,000 generations)
200
150
100
50
0
0
20
40
60
80
100
120
140
160
180
200
Coordinate (kb)

The Exercise

Focus: Understand how PSMC works, and think about the caveats with using it



The PSMC File

N: This character represents a homozygous site[1].
T: This character represents a heterozygous site[1].

Class Wrap Up

- What do you have to give PSMC?
  - How does PSMC work?
- Which assumptions should we have for PSMC to work?

Questions for next week

Weird Perl Error

- Run
- export LC_ALL="en_US.utf-8"