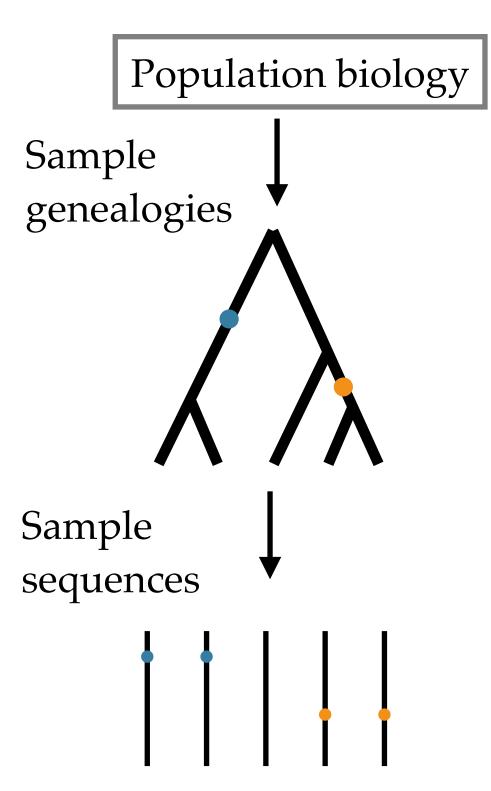
Distinguishing among coalescent models using two-site joint allele frequency spectra

Daniel P. Rice^{1*}, John Novembre¹, Michael M. Desai²

¹University of Chicago, ² Harvard University, *dpr@uchicago.edu

Genetic diversity depends on history through a genealogical process.



A population's demography, natural history, and the effects of selection determines how individuals are related to one another.

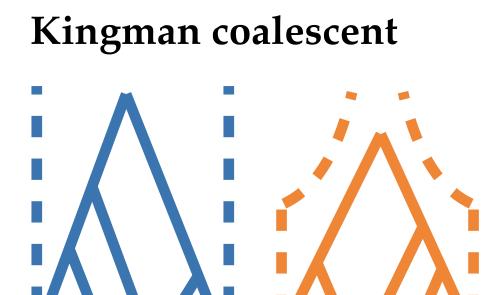
Relationships between individuals can be represented as genealogies.

The pattern of genetic diversity in a modern sample reflect statistical

properties of genealogies.

Population genetics uses models of the genealogical process to infer the past from the present.

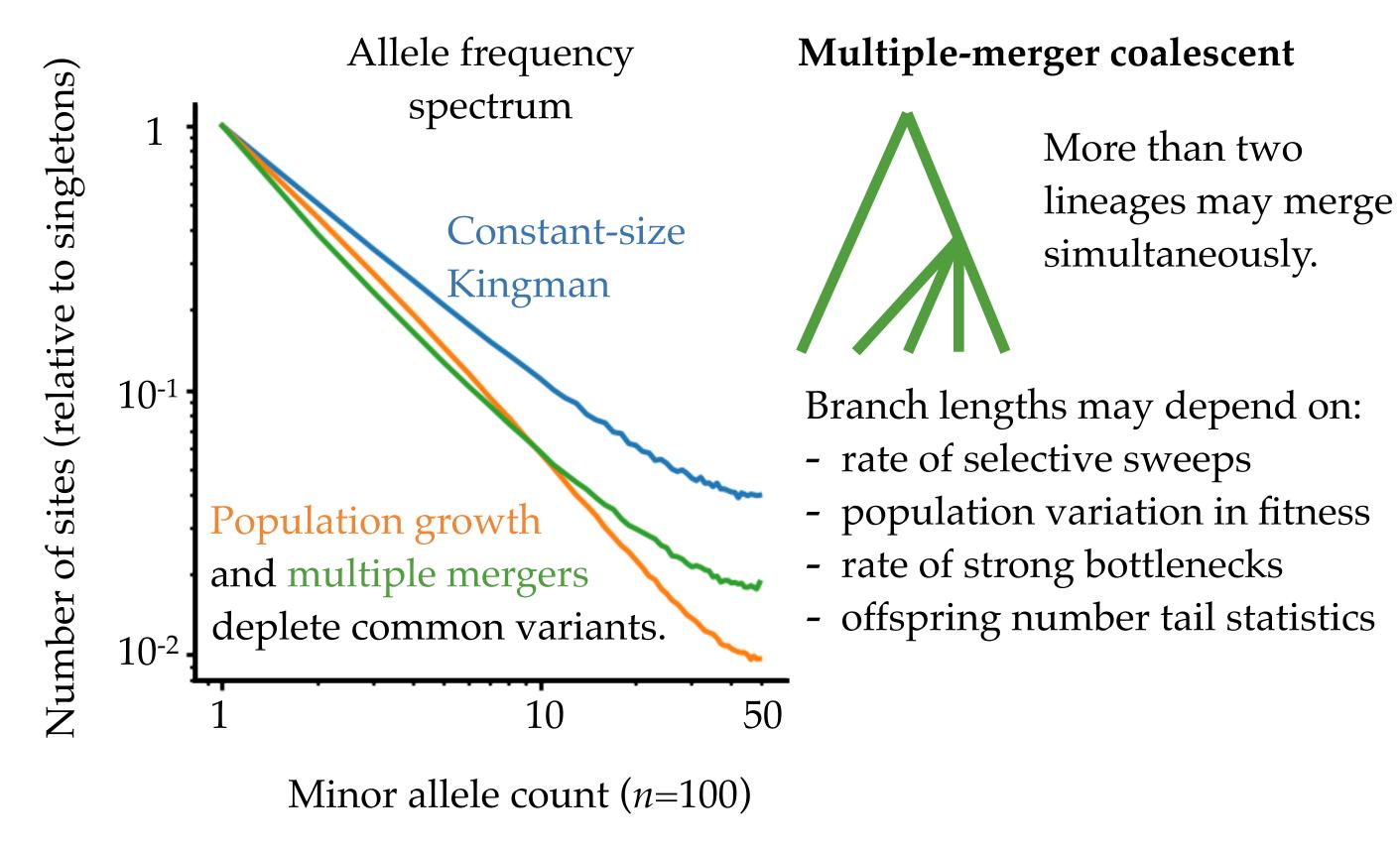
Signals of demographic and evolutionary history are confounded.



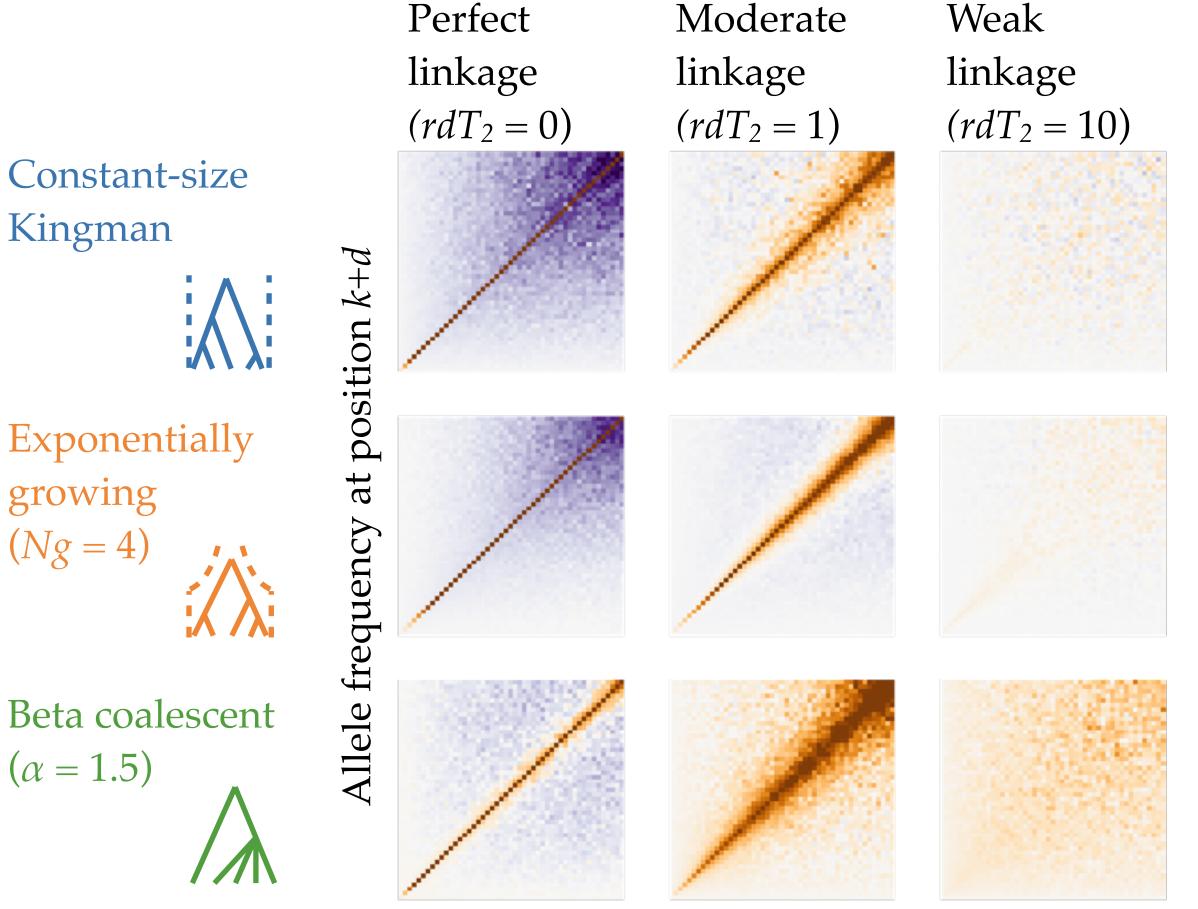
Only two lineages may merge at the same time.

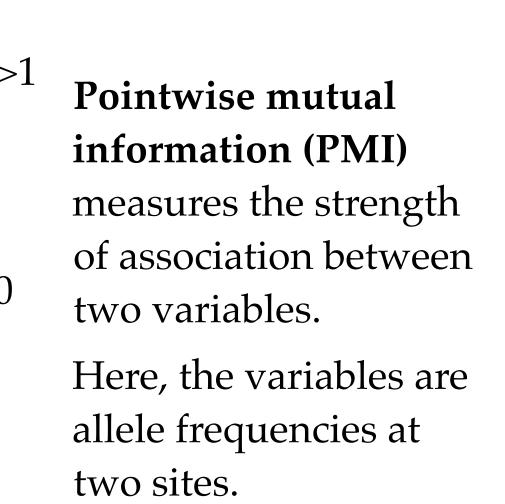
Branch lengths depend on slow variations in the population size.

Demographic inference is possible from genetic diversity data.



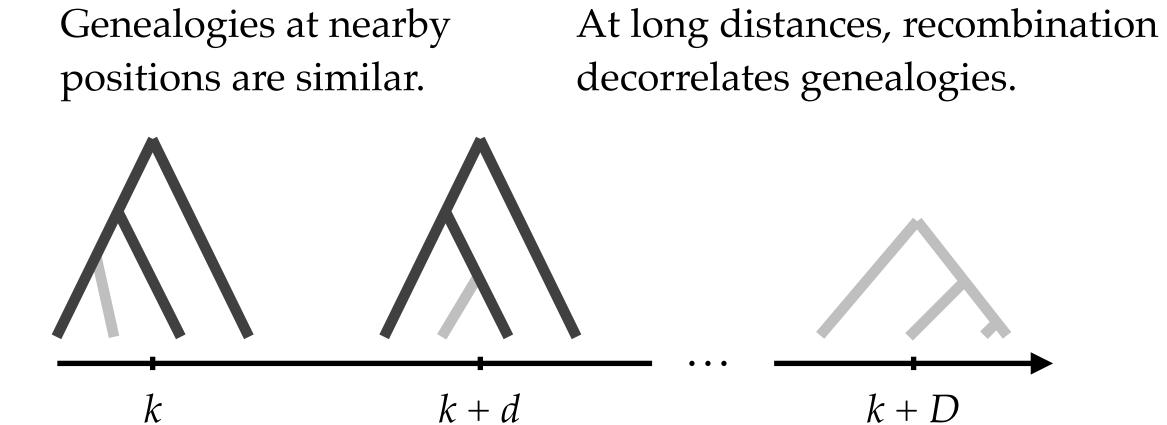
The joint allele frequency spectrum at pairs of linked sites distinguishes Kingman from multiple-merger coalescent models.





Multiple-merger coalescents have positive PMI between alleles at different frequencies.

The joint allele frequency spectrum depends on the distance between two sites.



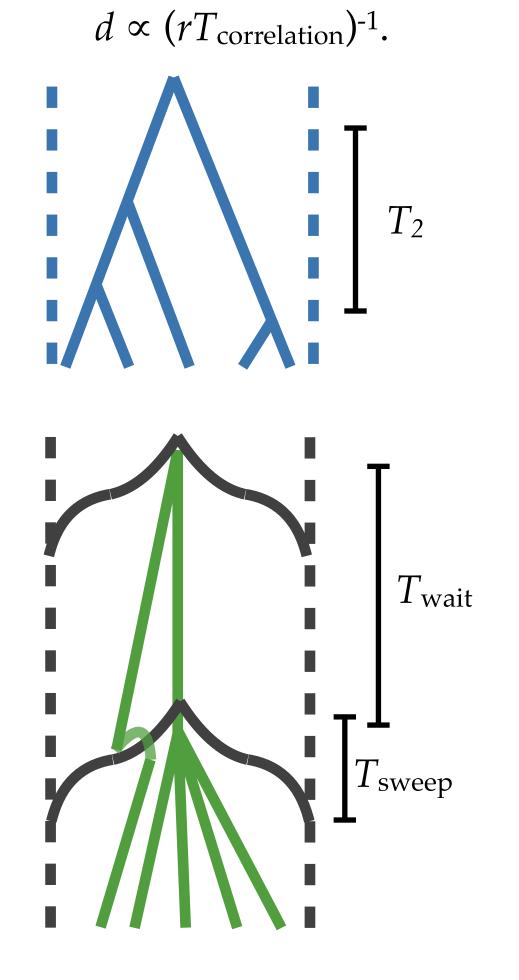
Genomic position

Multiple merger coalescents are generated by processes with multiple timescales.

The average pairwise diversity (π) depends on the mutation rate through:

 $\pi \propto \mu T_{\text{diveristy}}$.

The length scale of allele frequency correlations depends on the recombination rate through:



The Kingman coalescent is characterized by a single timescale:

 $T_{\text{diversity}} = T_{\text{correlation}} = T_2.$

So, $d \sim (rT_{\text{diversity}})^{-1}$.

Multiple mergers generated by selective sweeps at linked sites has a separation of timescales betwen:

 $T_{\rm diveristy} \propto T_{\rm wait}$

and

 $T_{\rm correlation} \propto T_{\rm sweep}$.

So, $d \gg (rT_{\text{diversity}})^{-1}$.

Mutual information between allele frequencies persists at long distances in multiple merger coalescents.

Allele frequency at position *k*

