Evolutionary Thinking 2022
TA session
week 5 – Basis of Population Genetics

Jilong Ma
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Outline

1. Learning outcome of today

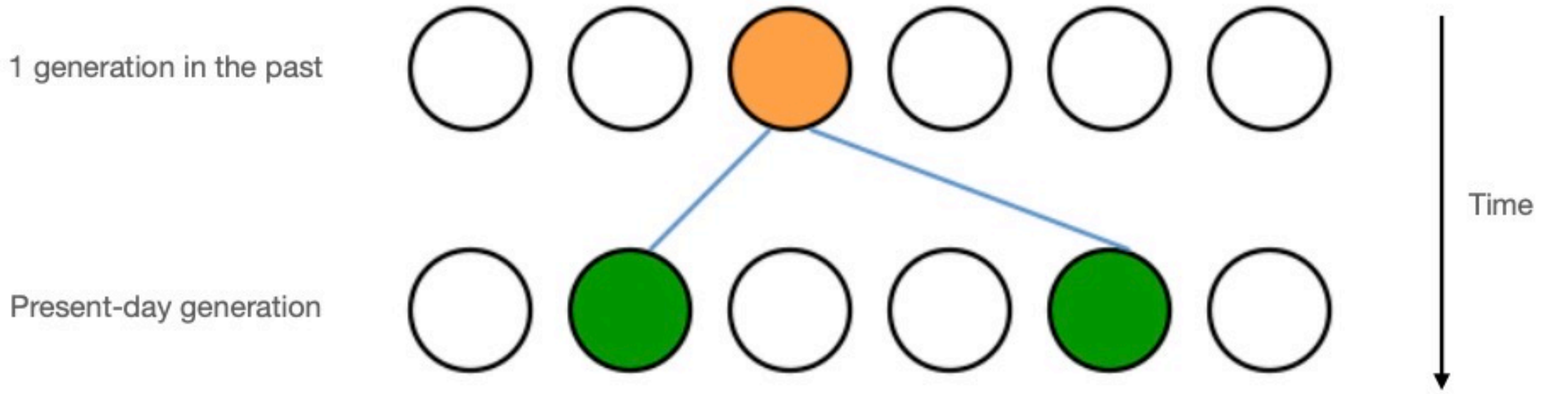
Coalescence Theory (Fri)

– Process, Tree and Tree length, Site Frequency Spectrum

2. R Exercises

Coalescence in a sample of two sequences

P[2 samples have the same parent in the previous generation] =

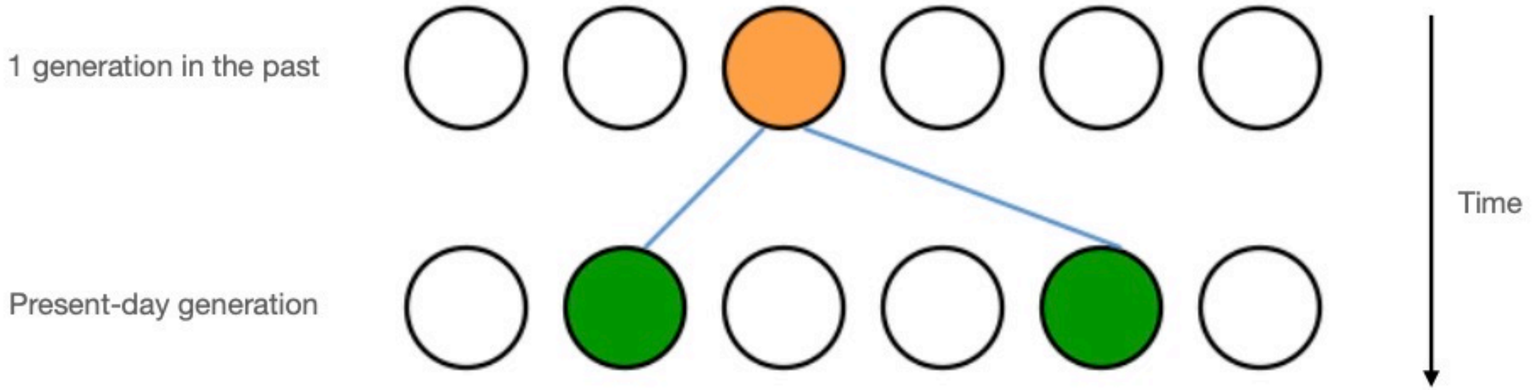


Slides from Fernando Racimo
“Intro to popgen”

Coalescence in a sample of two sequences

P[2 samples have the same parent in the previous generation] =

$$2N\frac{1}{2N}\frac{1}{2N}=\frac{1}{2N}$$



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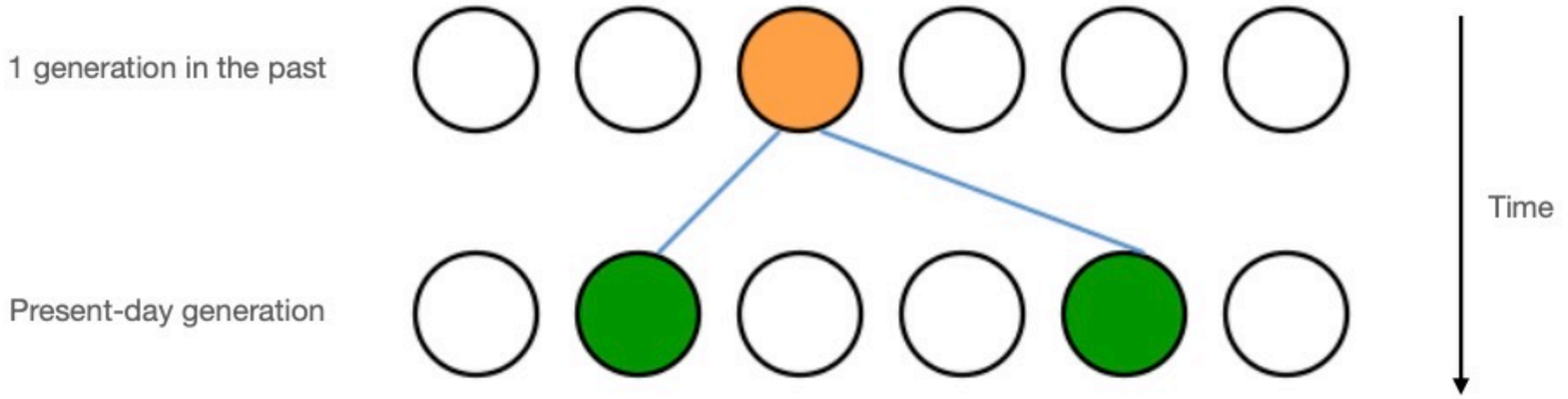

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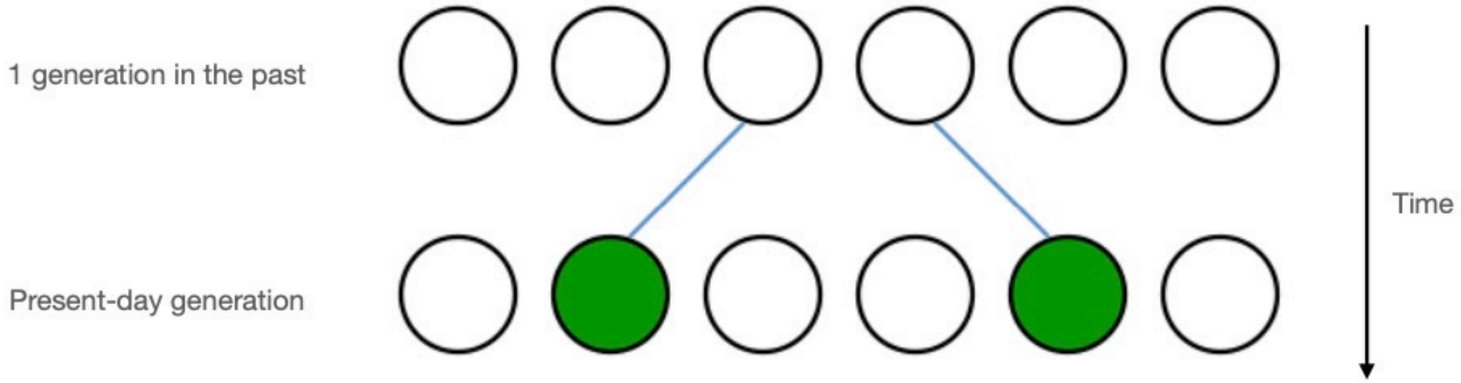
P[2 samples do not have the same parent in the previous generation] =



Slides from Fernando Racimo
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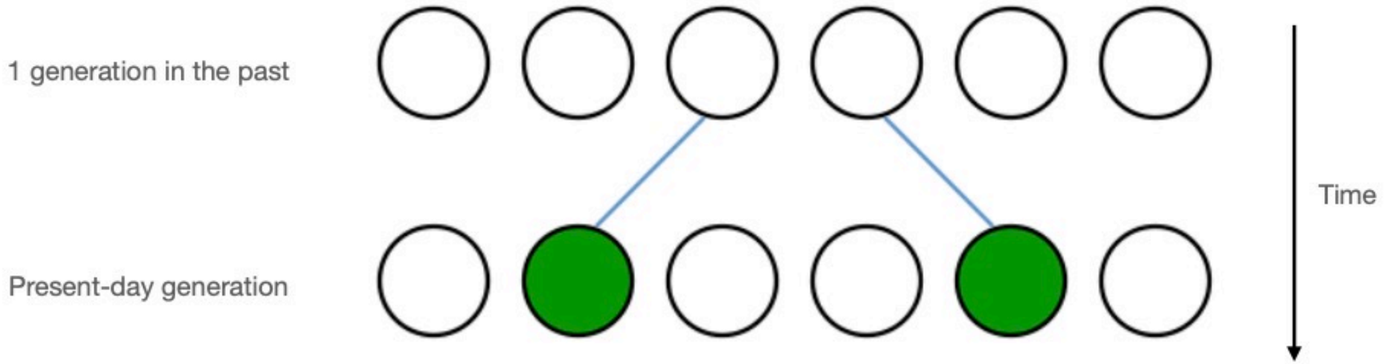
Coalescence in a sample of two sequences

P[2 samples do not have the same parent in the previous generation] =

$$1 - \frac{1}{2N}$$



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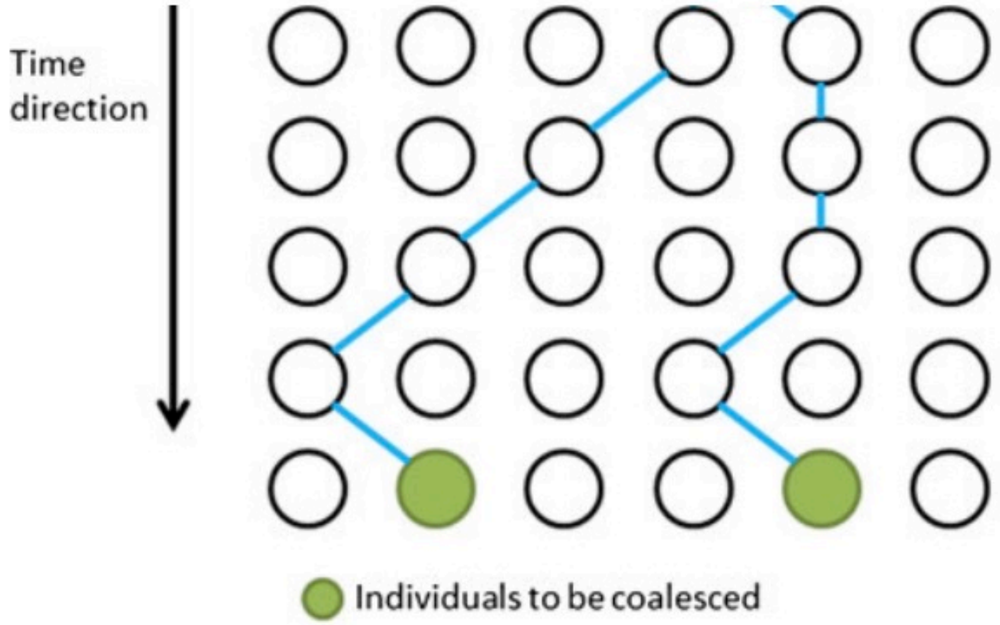
P[2 samples do not find a common ancestor in **r** generations] =



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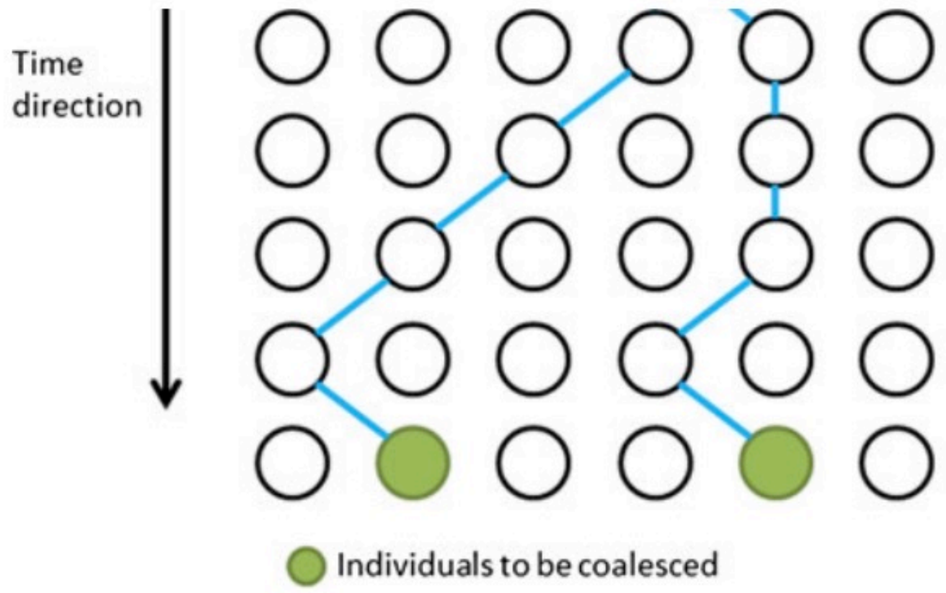
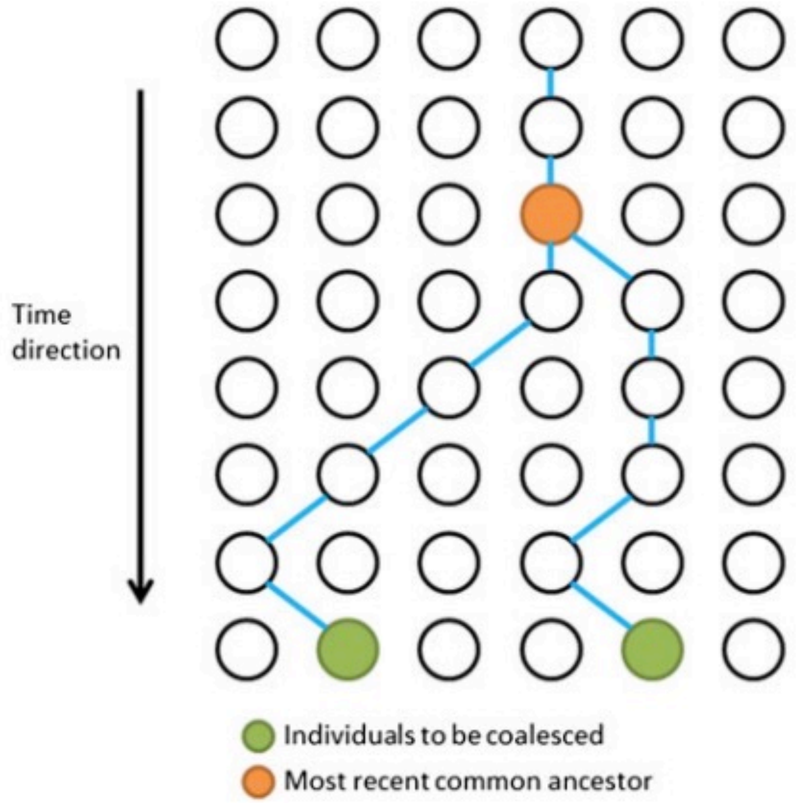


$$\left(1-\frac{1}{2N}\right)^{r}$$

Slides from Fernando Racimo
“Intro to popgen”

Coalescence in a sample of two sequences

P[2 samples find a common ancestor in **exactly r** generations] =



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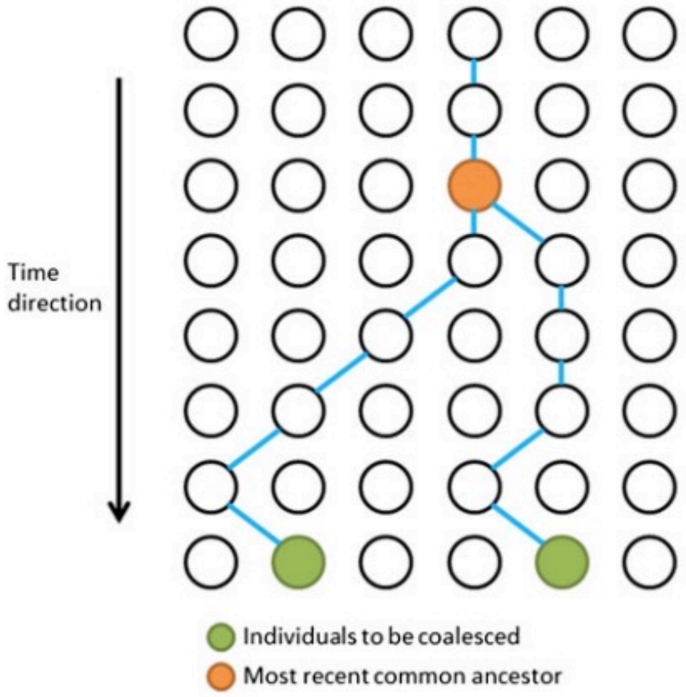

$$\left(1-\frac{1}{2N}\right)^{r-1}\frac{1}{2N}$$

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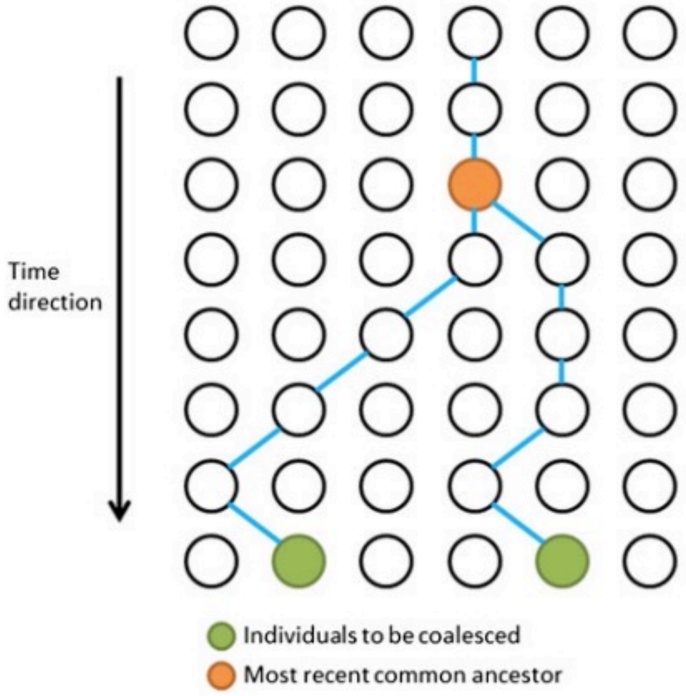

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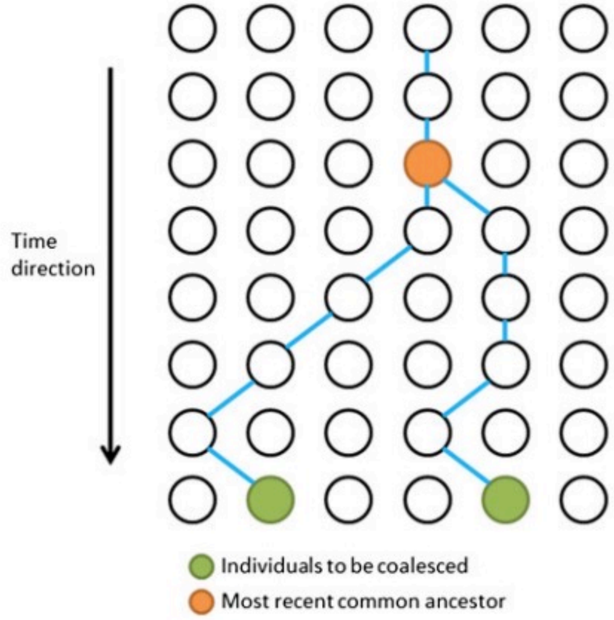

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$$\approx \frac{1}{2N}e^{-r/(2N)}$$

This approximation is very good when the population size is large (2N > ~1000)

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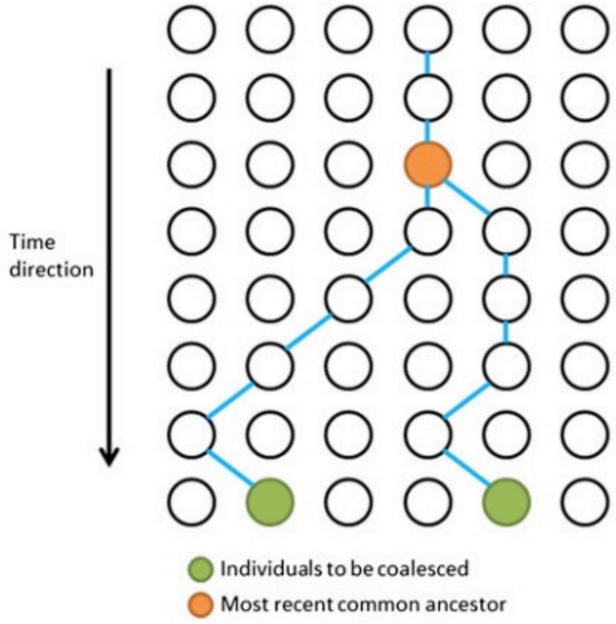

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If we measure time in units of 2N generations (1 t = 2N r)

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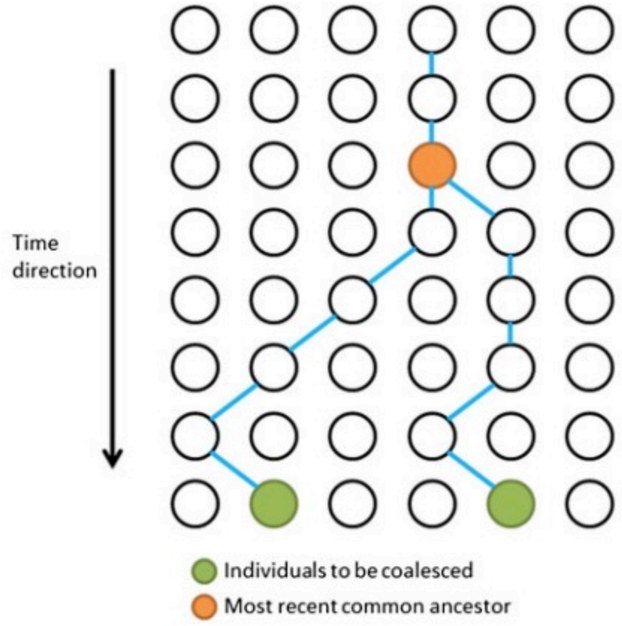

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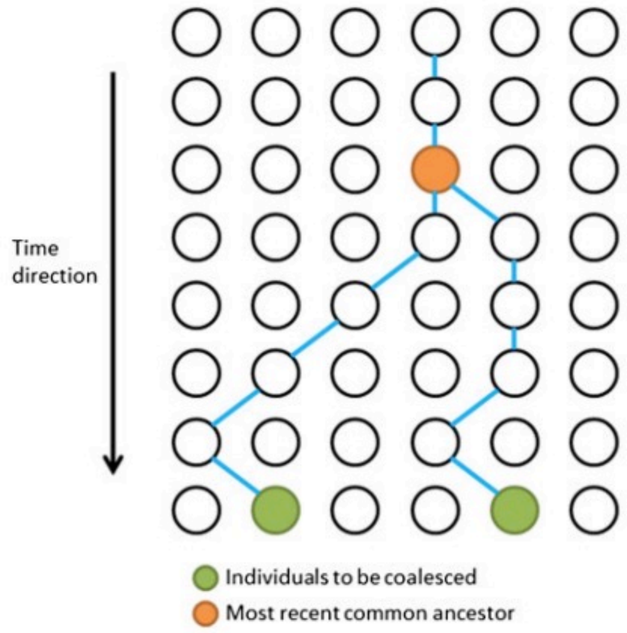

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This approximation is very good when the population size is large (2N > ~1000)

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If we measure time in units of 2N generations (1 t = 2N r)

This is an **exponential distribution** with rate 1

Slides from Fernando Racimo
“Intro to popgen”

The expected waiting time

One parameter: λ

$$E[T] = \frac{1}{\lambda}$$

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The expected waiting time

One parameter: λ

$$E[T] = \frac{1}{\lambda}$$

The expected waiting time is the inverse of the rate

If buses arrive at an average rate of λ=4 per hour,
I expect to wait 1/4 of an hour for the next bus, on average

Slides from Fernando Racimo
“Intro to popgen”

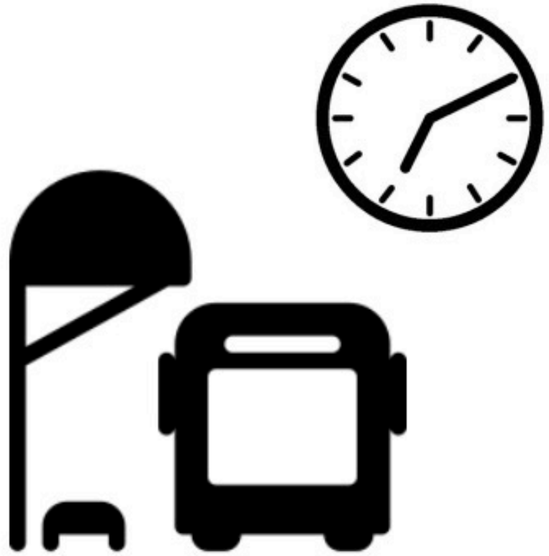
The expected waiting time

"Buses arrive at a rate of λ per hour"

$$f(t) = \lambda e^{-\lambda t}$$

"The expected waiting time for the next bus is $1/\lambda$ hours"

$$E[T] = \frac{1}{\lambda}$$

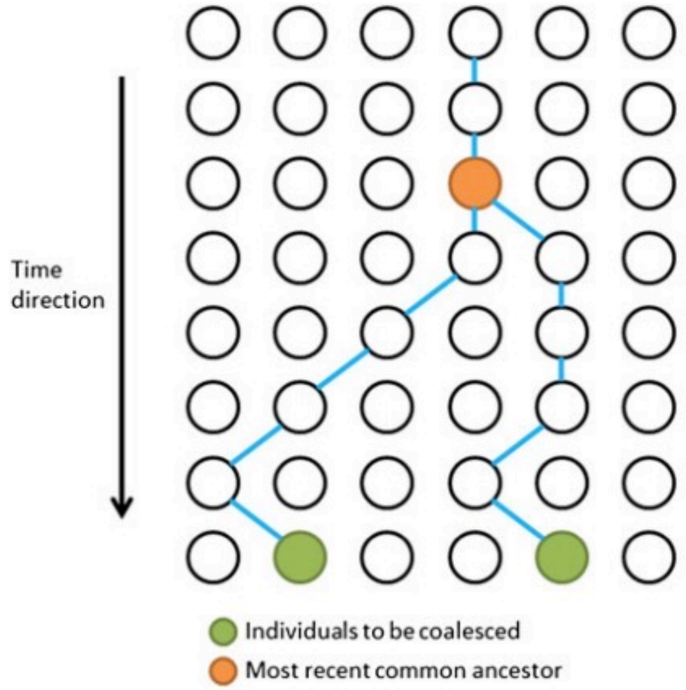


Slides from Fernando Racimo
"Intro to popgen"

The expected waiting time

"A coalescent event between 2 lineages occurs at a rate of 1/2N per generation"

$$f(t) = \frac{1}{2N} e^{-t/(2N)}$$



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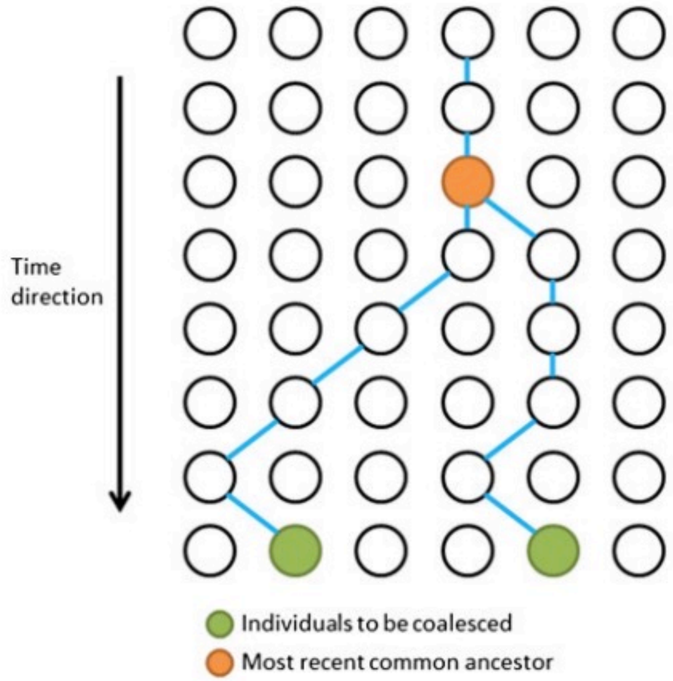
The expected waiting time

"A coalescent event between 2 lineages occurs at a rate of 1/2N per generation"

$$f(t) = \frac{1}{2N} e^{-t/(2N)}$$

"The expected waiting time for a coalescent event is 2N generations"

$$E[T] = 2N$$



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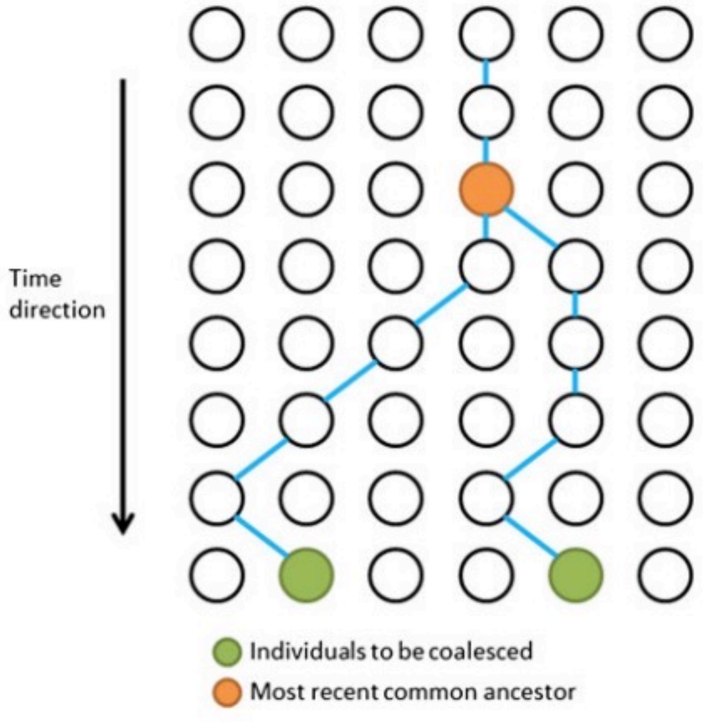
The expected waiting time

"A coalescent event between 2 lineages occurs at a rate of 1 per coalescent unit (1 unit = 2N generations)."

$$f(t) = e^{-t}$$



slides from Fernando Racimo "Intro to popgen"

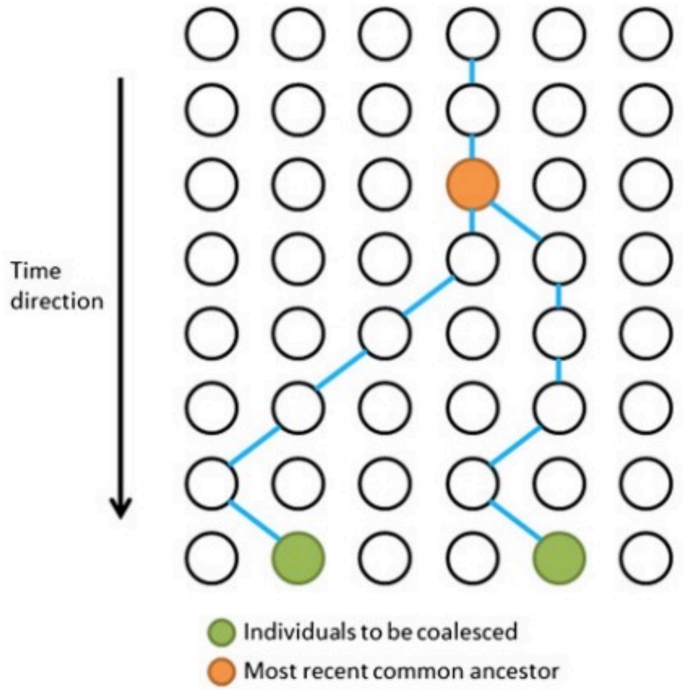
The expected waiting time

"A coalescent event between 2 lineages occurs at a rate of 1 per coalescent unit (1 unit = 2N generations)."

$$f(t) = e^{-t}$$

"The expected waiting time for a coalescent event is 1 coalescent unit"

$$E[T] = 1$$



Slides from Fernando Racimo
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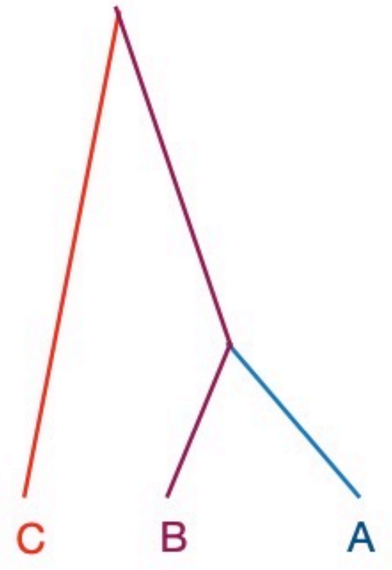
Tree with 3 samples



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Tree with 3 samples

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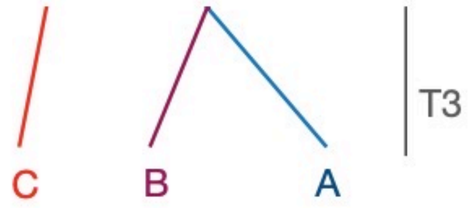
Tree with 3 samples

T3 = time while there are 3 lineages

Rate: $\lambda_{T3} = 1 * \binom{3}{2} = 3$



Slides from Fernando Racimo
“Intro to popgen”

Tree with 3 samples

T3 = time while there are 3 lineages

Rate: $\lambda_{T3} = 1 * \binom{3}{2} = 3$

…because there are 3 "competing pairs" of lineages fighting for the opportunity to coalesce (each at rate 1): A+B, B+C, A+C

Expected time: $E[T3] = \frac{1}{\binom{3}{2}} = \frac{1}{3}$

Slides from Fernando Racimo "Intro to popgen"

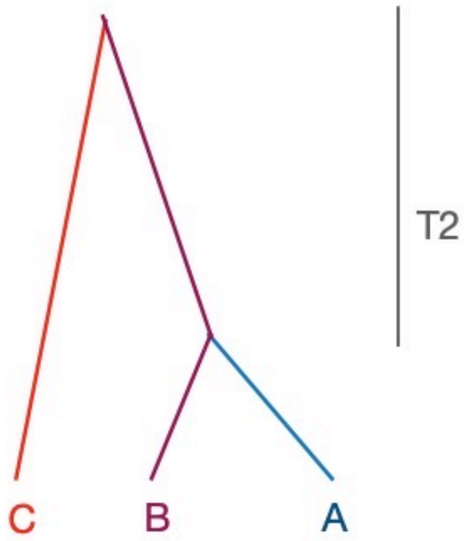
Tree with 3 samples

T2 = time while there are 2 lineages

Rate: $\lambda_{T2} = \frac{1}{\binom{2}{2}} = 1$

…because there is just one possible pair that can coalesce (at rate 1)

Expected time: $E[T2] = 1$



Slides from Fernando Racimo
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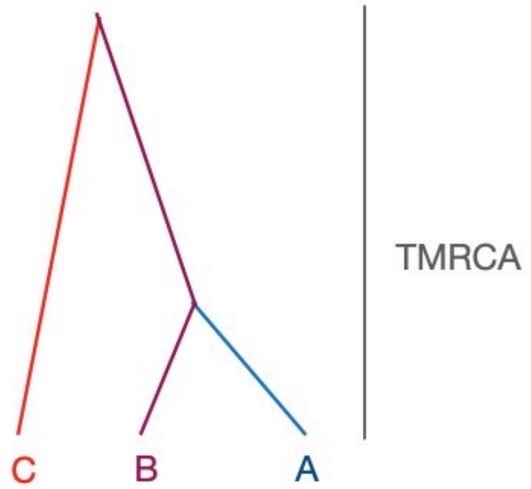
Tree with 3 samples

$$E[T_{MRCA}] = T_2 + T_3 = 1 + 1/3 \text{ coalescent units}$$

$$E[T_{MRCA}] = (1 + 1/3) * 2N \text{ generations}$$



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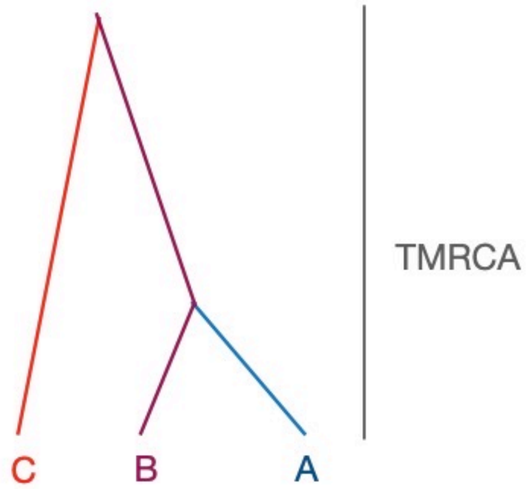

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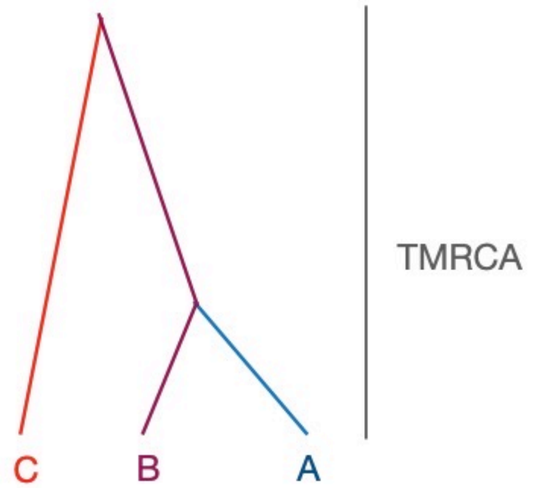

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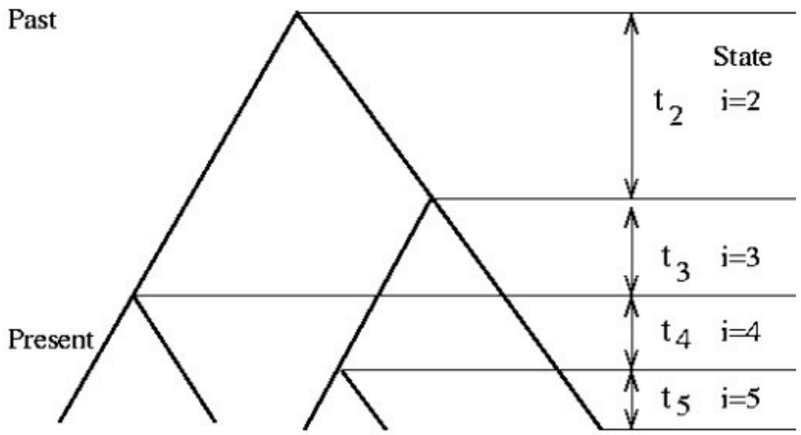
Tree with 3 samples

$$E[T_{MRCA}] = T_2 + T_3 + \ldots + T_n$$

$$E[T_{MRCA}] = \sum_{i=2}^{n} \frac{1}{\binom{i}{2}} = 2\left(1 - \frac{1}{n}\right)$$

where n is our sample size



Slides from Fernando Racimo
“Intro to popgen”

Variance of an exponential distribution

One parameter: λ

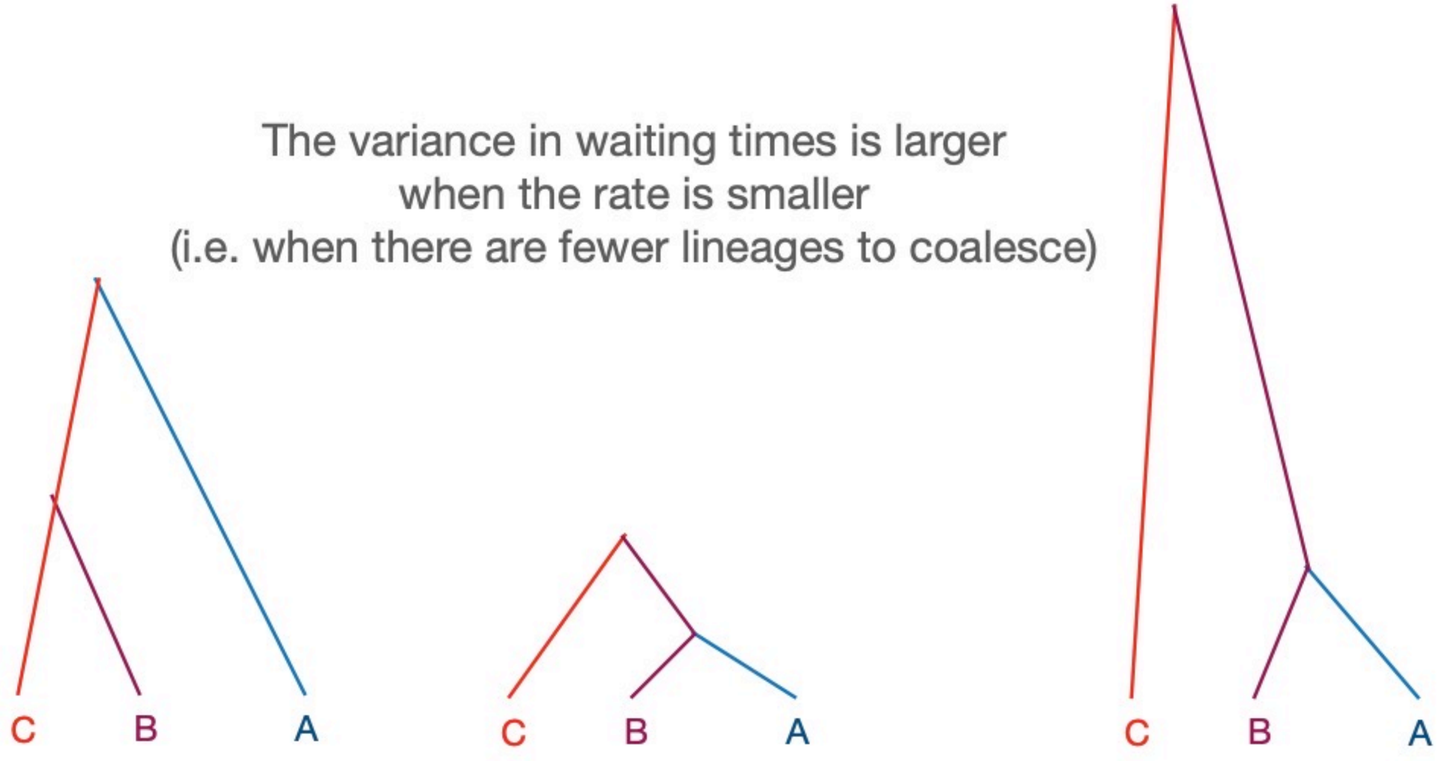
$$E[T] = \frac{1}{\lambda}$$

$$Var[T] = \frac{1}{\lambda^2} \longrightarrow \text{The variance in waiting times is larger when the rate is smaller}$$

Slides from Fernando Racimo
“Intro to popgen”

Variance in coalescence times

The variance in waiting times is larger
when the rate is smaller
(i.e. when there are fewer lineages to coalesce)



Slides from Fernando Racimo
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Exercises

R exercises

Left from Wednesday
Chapter 1:
1.1-1.4
Chapter 2:
2.1-2.3
2.6-2.9
Chapter 3:
3.1-3.8

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