



MONASH University

Science

# A quick introduction to the Coalescent

Anders Gonçalves da Silva

Population and landscape genomics workshop – Day 2

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CBA – ANU

# Outline

- Define the coalescent
- Introduce a reproductive model
- Use the reproductive model to turn-back time
- Conceptualize a simple coalescent model
- Try it out in *R*
- Introduce the idea of a *skyline plot*
- Modify our model to accommodate population growth
- Test it with *skyline plots*

## Question?

- What is the likelihood that at least one pair of genes in this room shares a common ancestor one generation in the past?
- How many generations do we need to go back before the probability is larger than 50%?



# What is the Coalescent?

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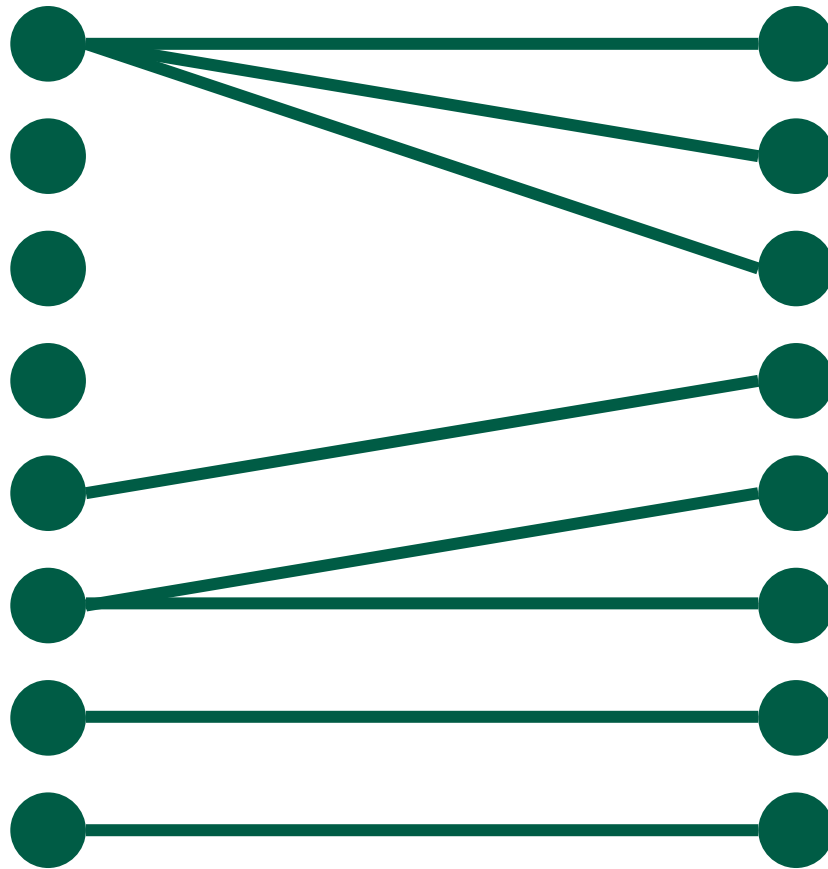
- A statistical model:

$$P(\text{Genealogy}|\theta)$$

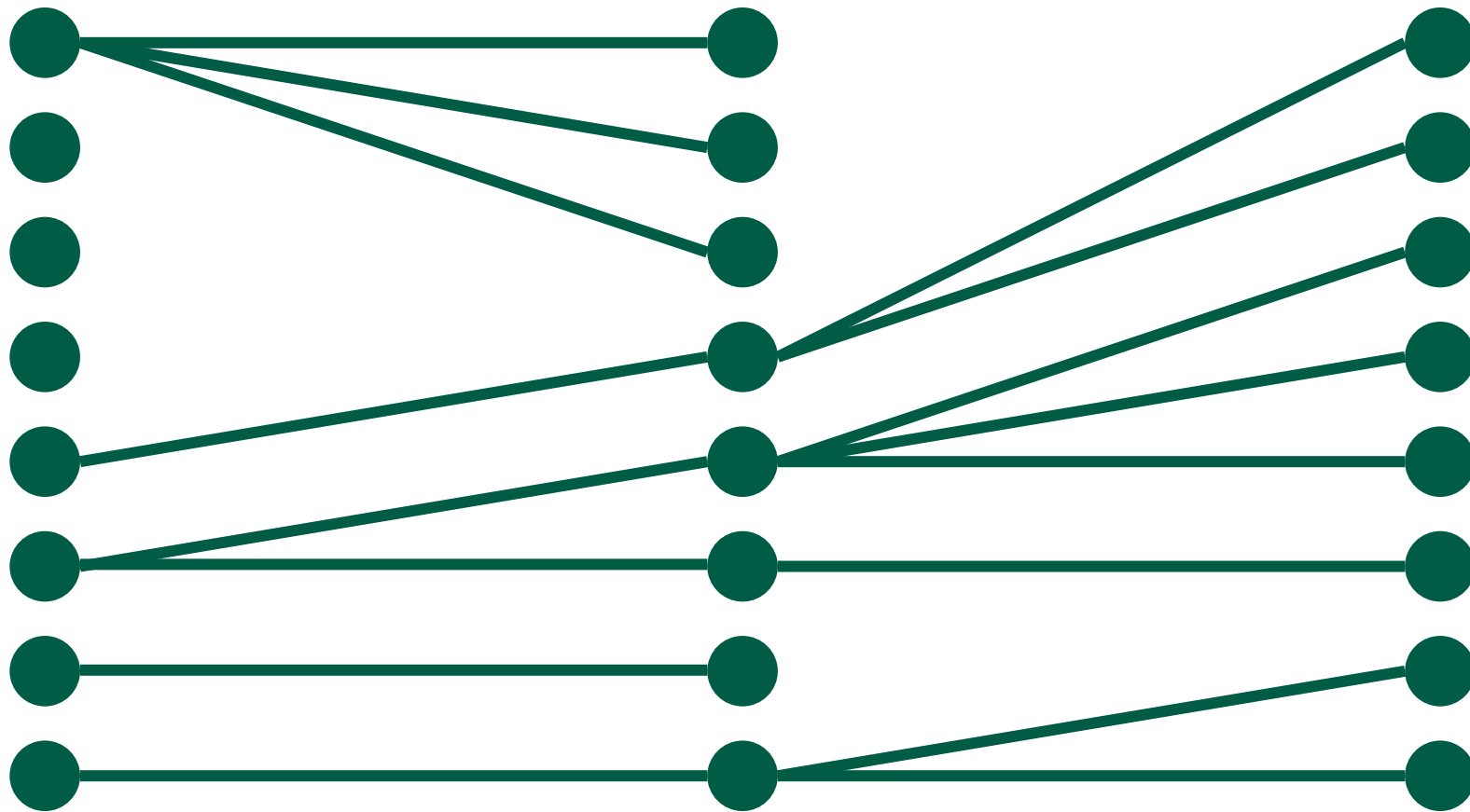
## Reproductive model: Wright-Fisher

- $2N = 8$  – number of haploid genomes
- 
- Each genes is equally represented
- in the gamete pool.
- 
- $2N$  gametes are chosen at random
- to form the next generation.
-

# Reproductive model: Wright-Fisher

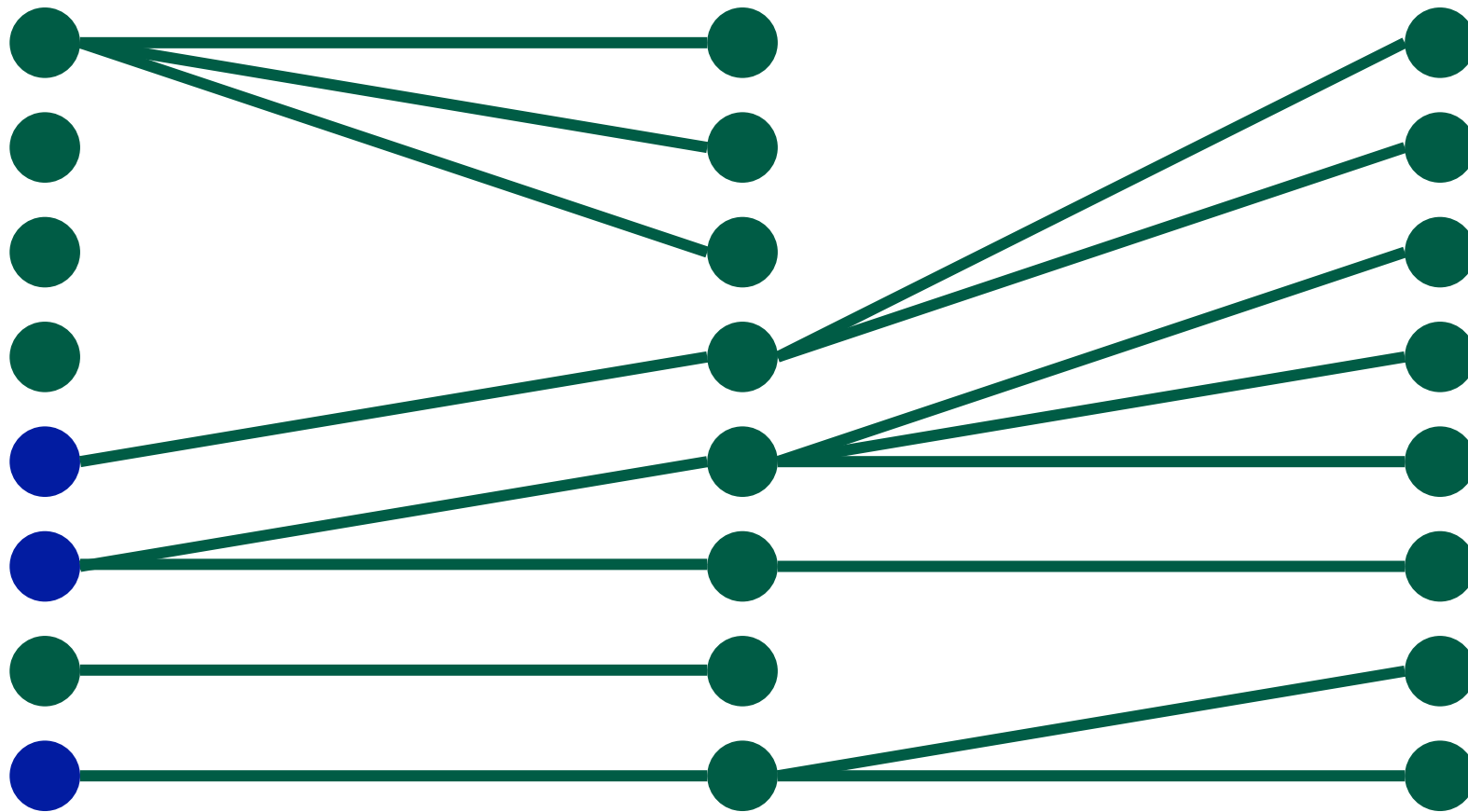


# Reproductive model: Wright-Fisher





# Reproductive model: Wright-Fisher



# The coalescent

How long before two lineages merge?

How long before all lineages merge?

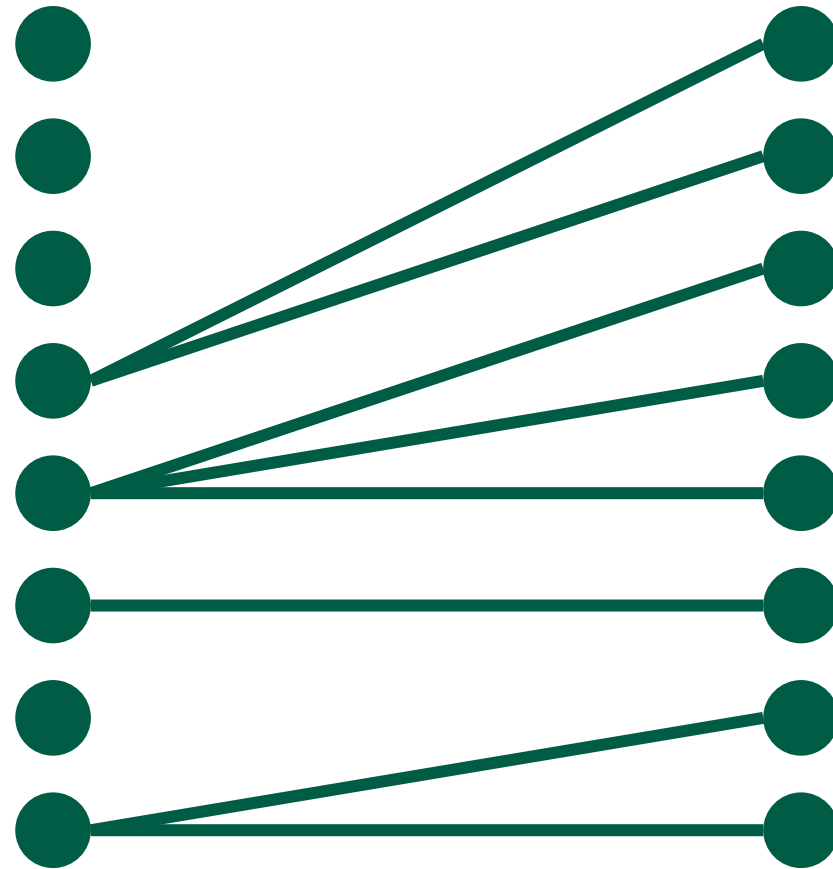
How long is measured in generations!

Merging is a coalescent event!



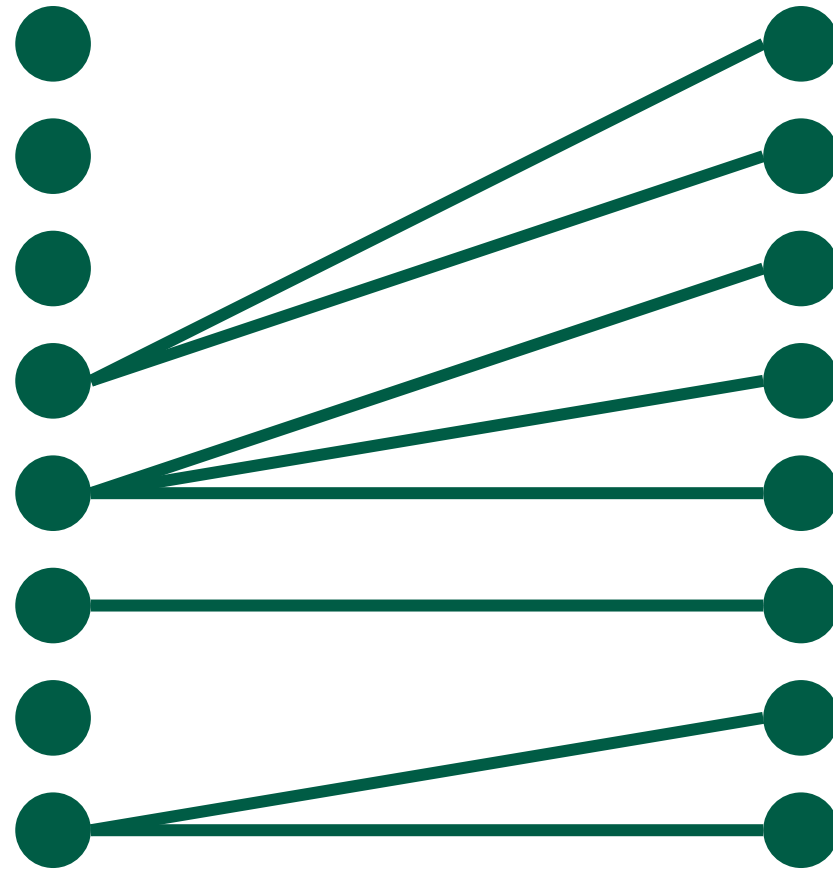
# The $n$ -coalescent

What is the probability  
that two lineages  
coalesce in the previous  
generation?



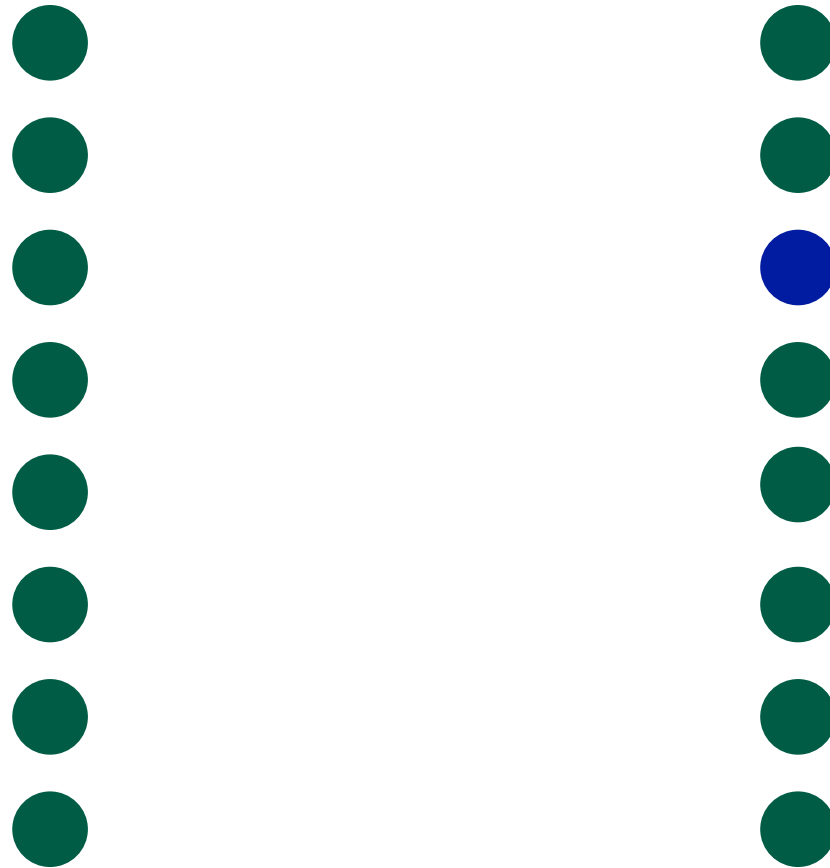
# The $n$ -coalescent

$$P(T_2=1)$$



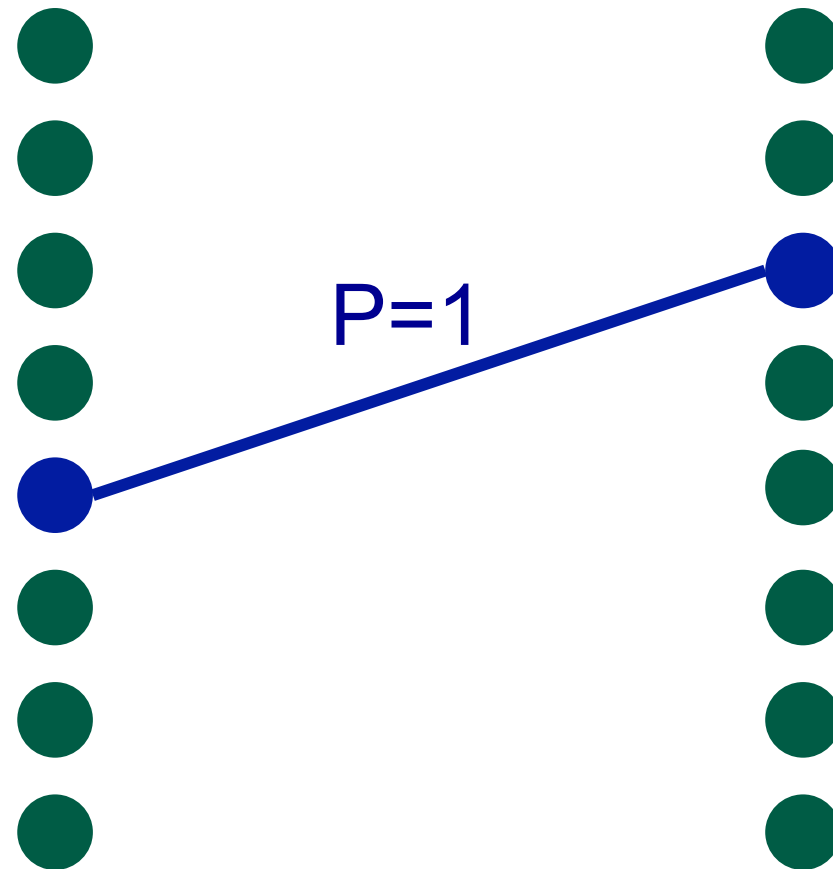
# The $n$ -coalescent: Wright's insight

What is the probability that a random individual has a parent in the previous generation?



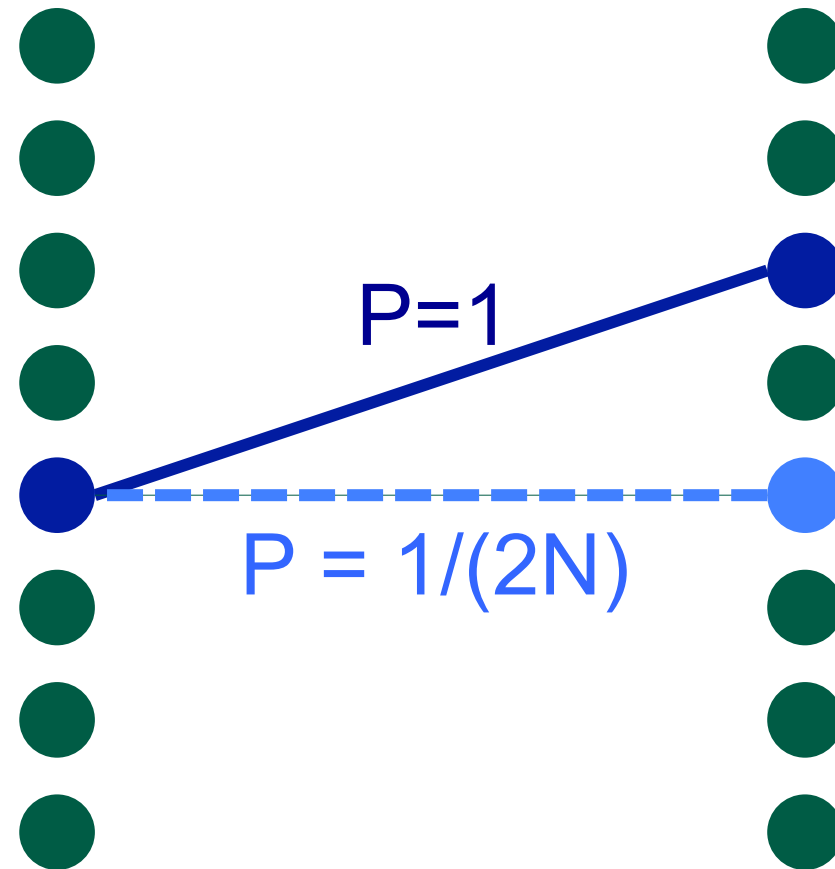
# The $n$ -coalescent: Wright's insight

What is the probability that a second individual shares the same parent?



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# The $n$ -coalescent: Wright's insight

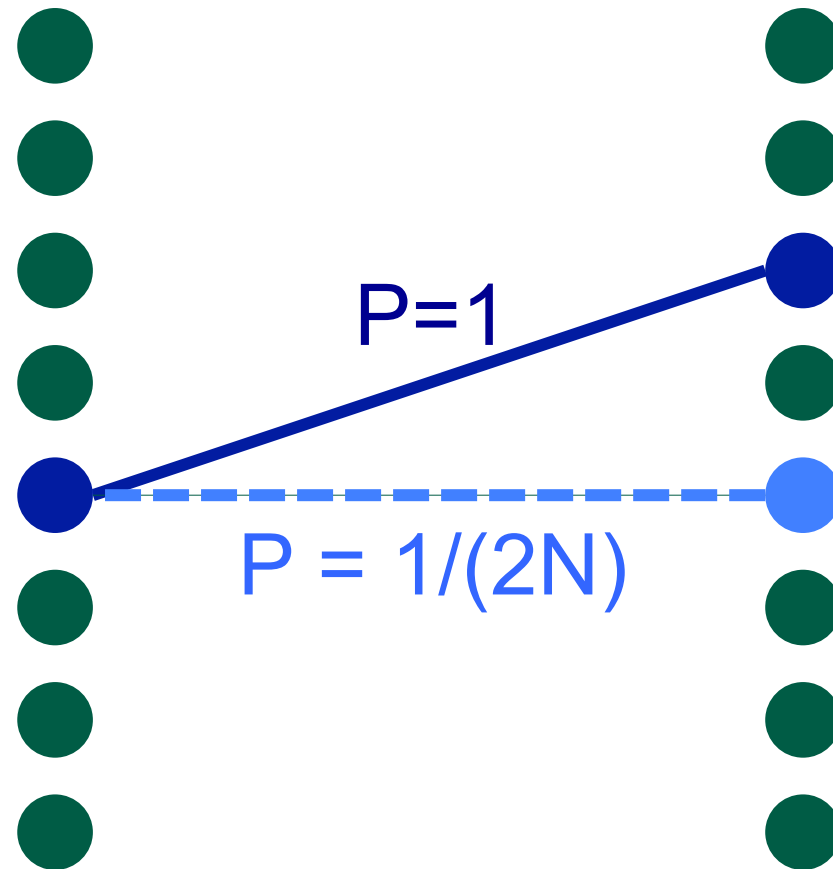
$$P(T_2=1)$$

Pr(Individual has parent)

$\times$

Pr(Individual

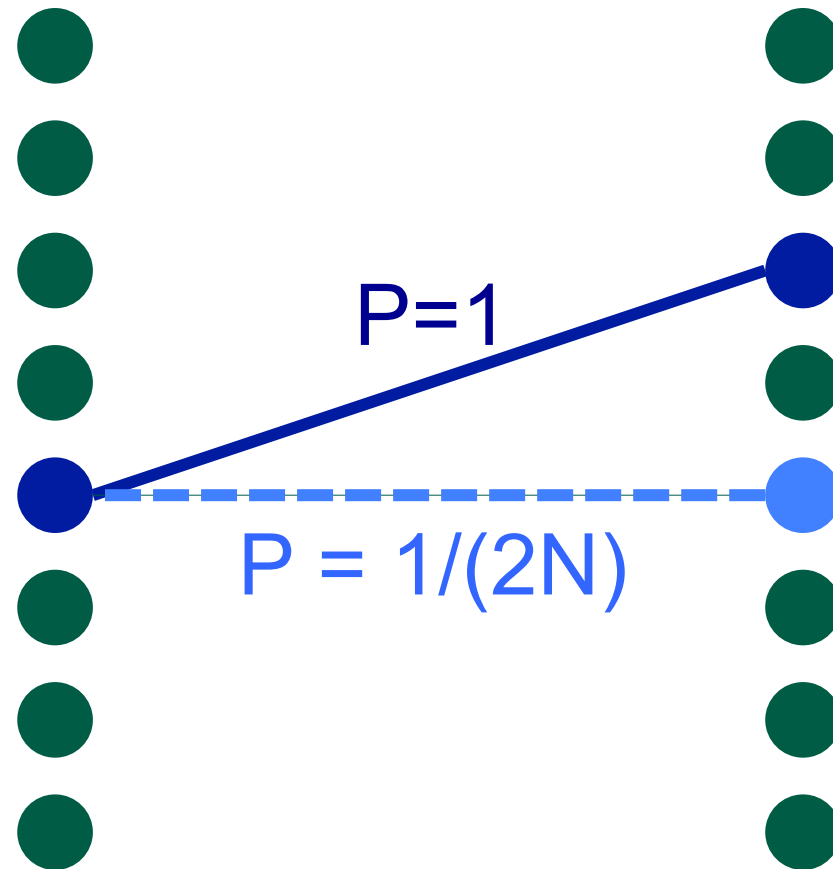
shares parent)





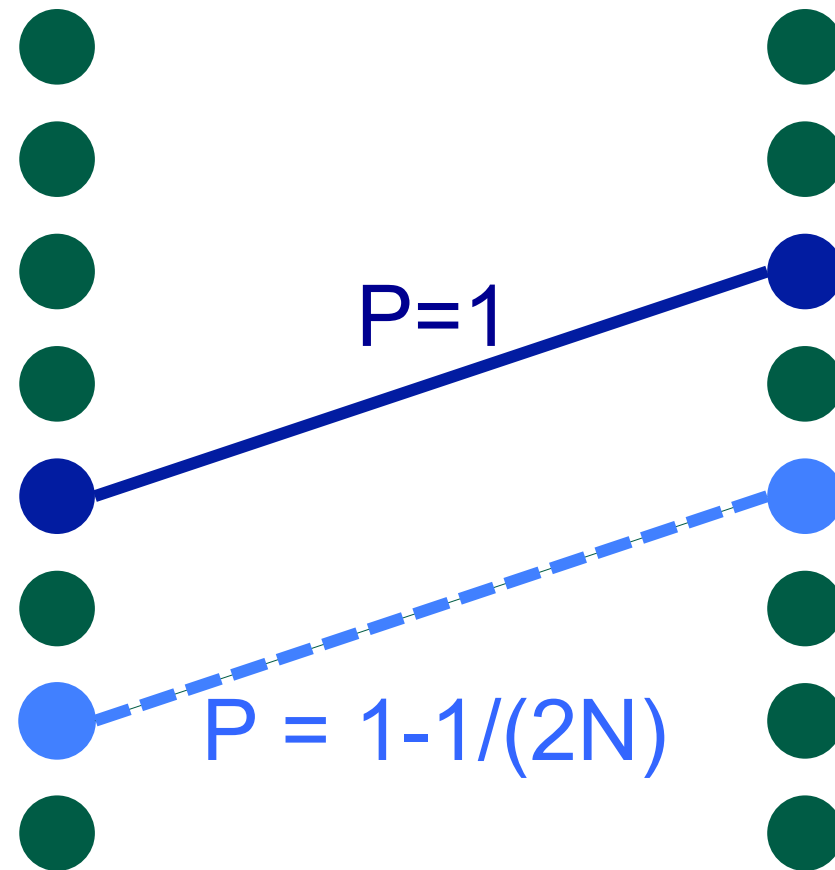
# The $n$ -coalescent: Wright's insight

$$P(T_2=1) = 1/(2N)$$



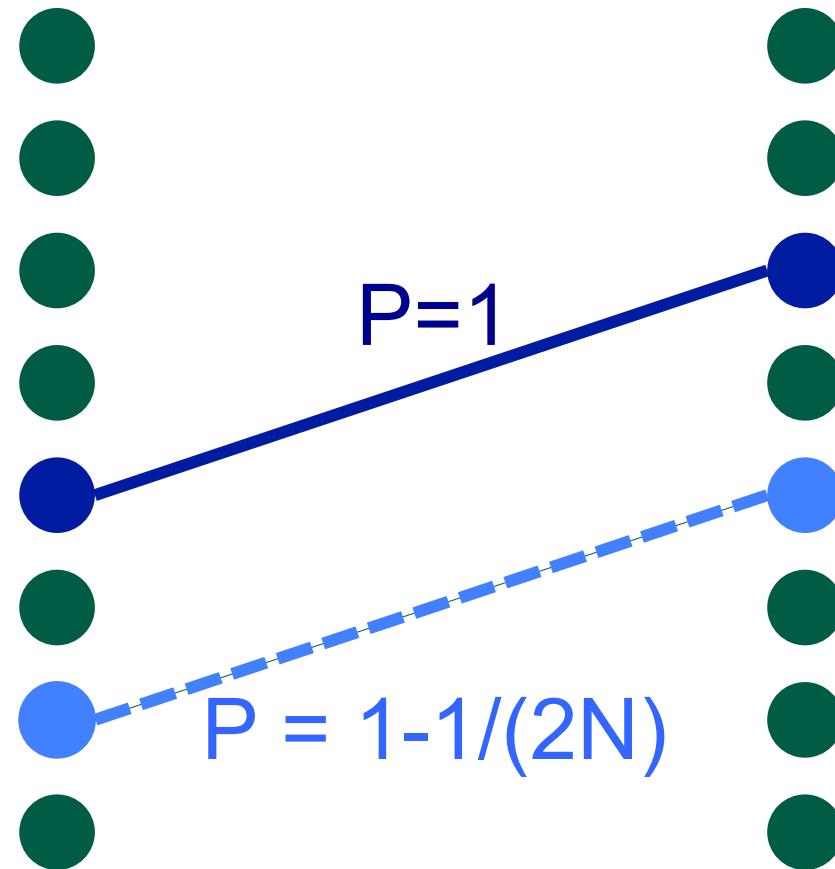
# The $n$ -coalescent: Wright's insight

$P(T_2 \neq 1)$

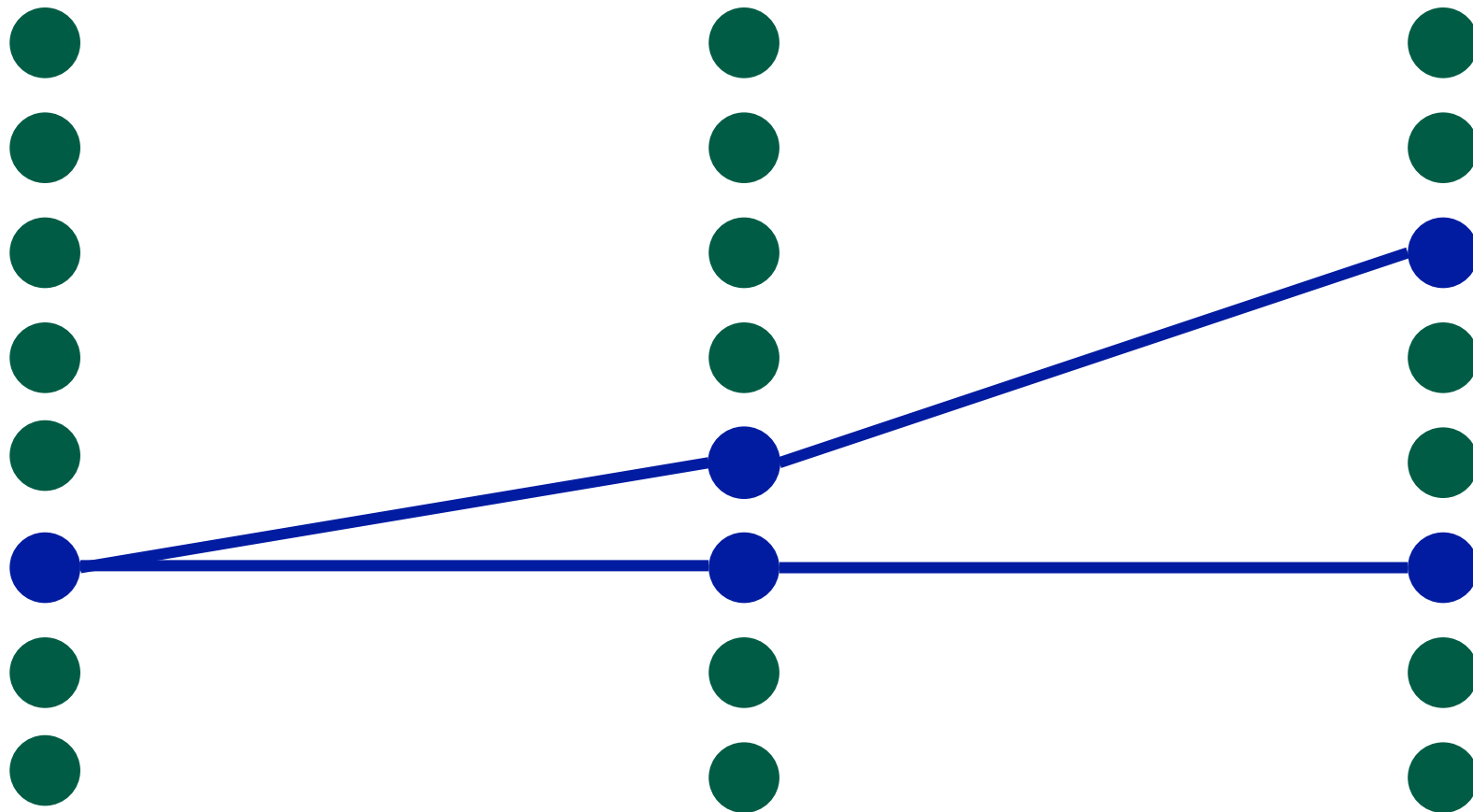


# The $n$ -coalescent: Wright's insight

$$P(T_2 \neq 1) = 1 - 1/(2N)$$

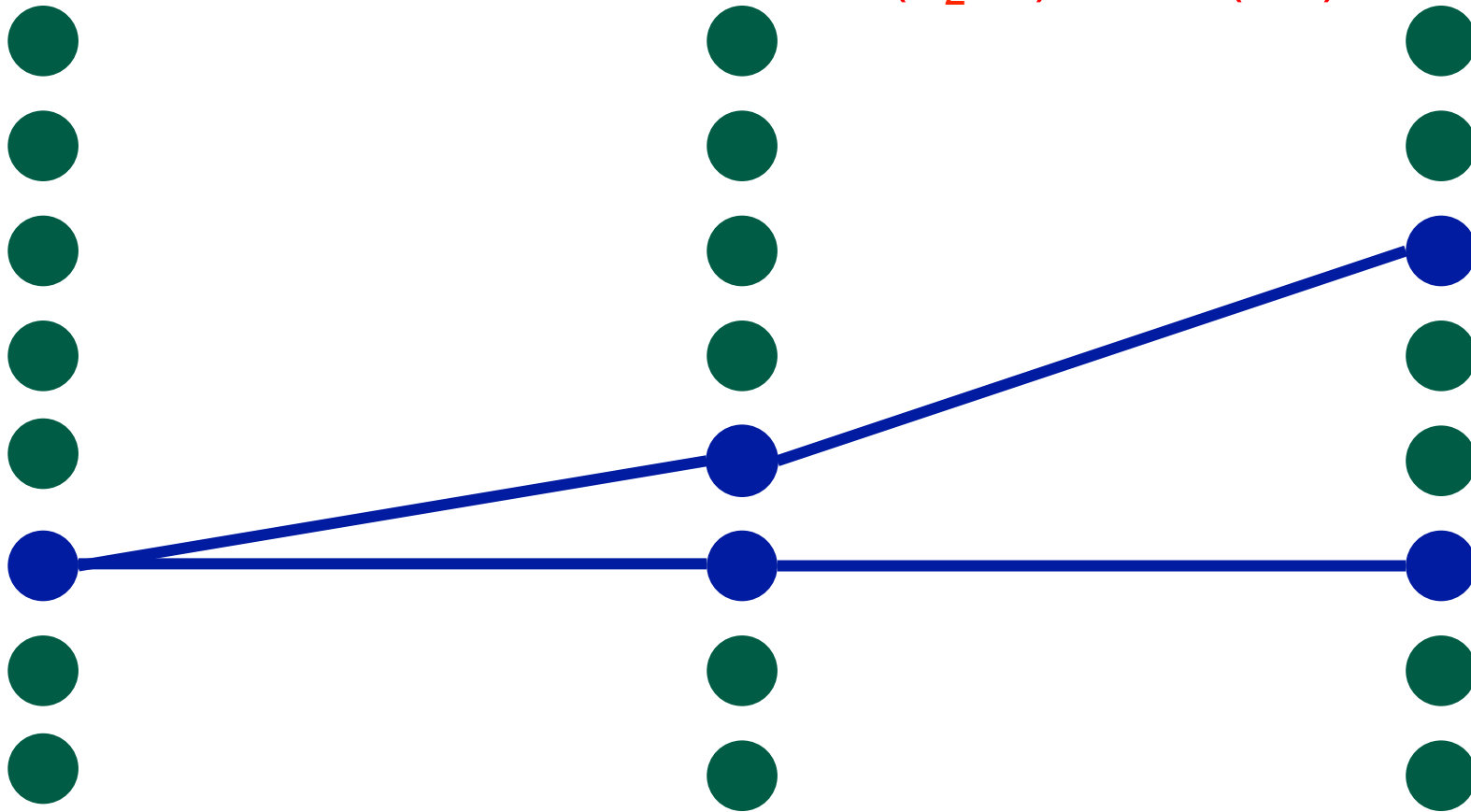


$P(T_2=2)?$



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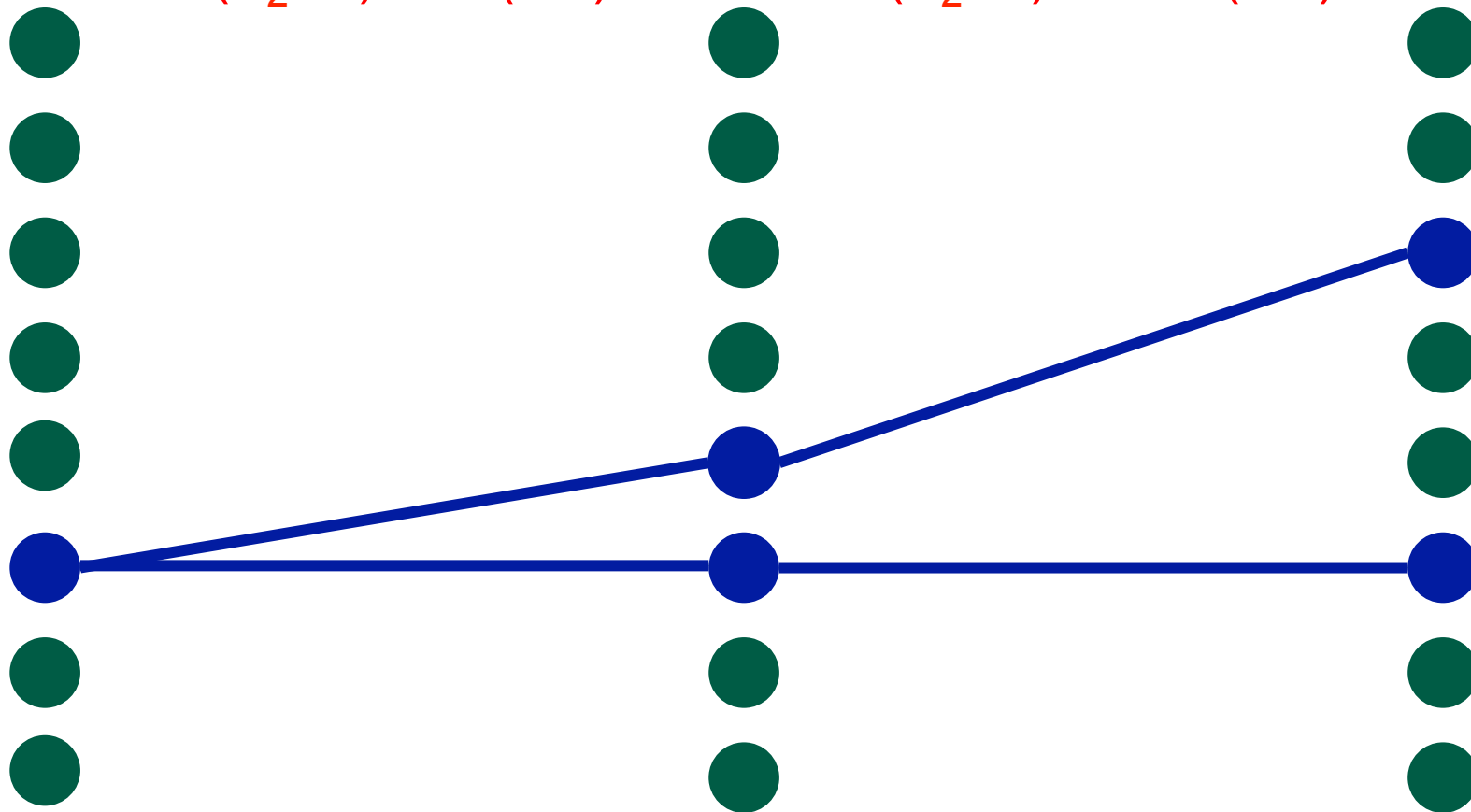
$$P(T_2 \neq 1) = 1 - 1/(2N)$$



$P(T_2=2)?$

$$P(T_2=1) = 1/(2N)$$

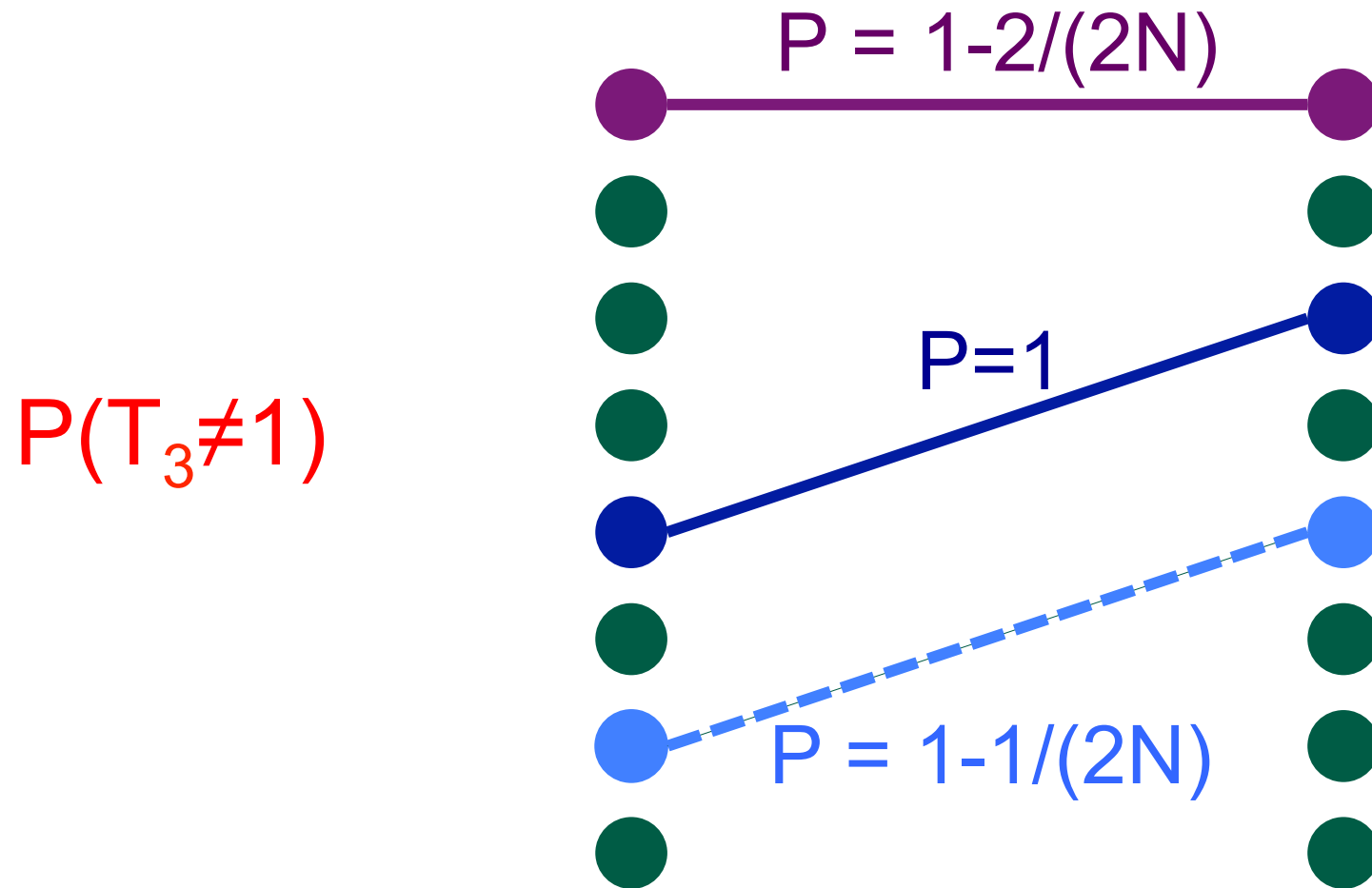
$$P(T_2 \neq 1) = 1 - 1/(2N)$$



**$P(T_2=j)?$**

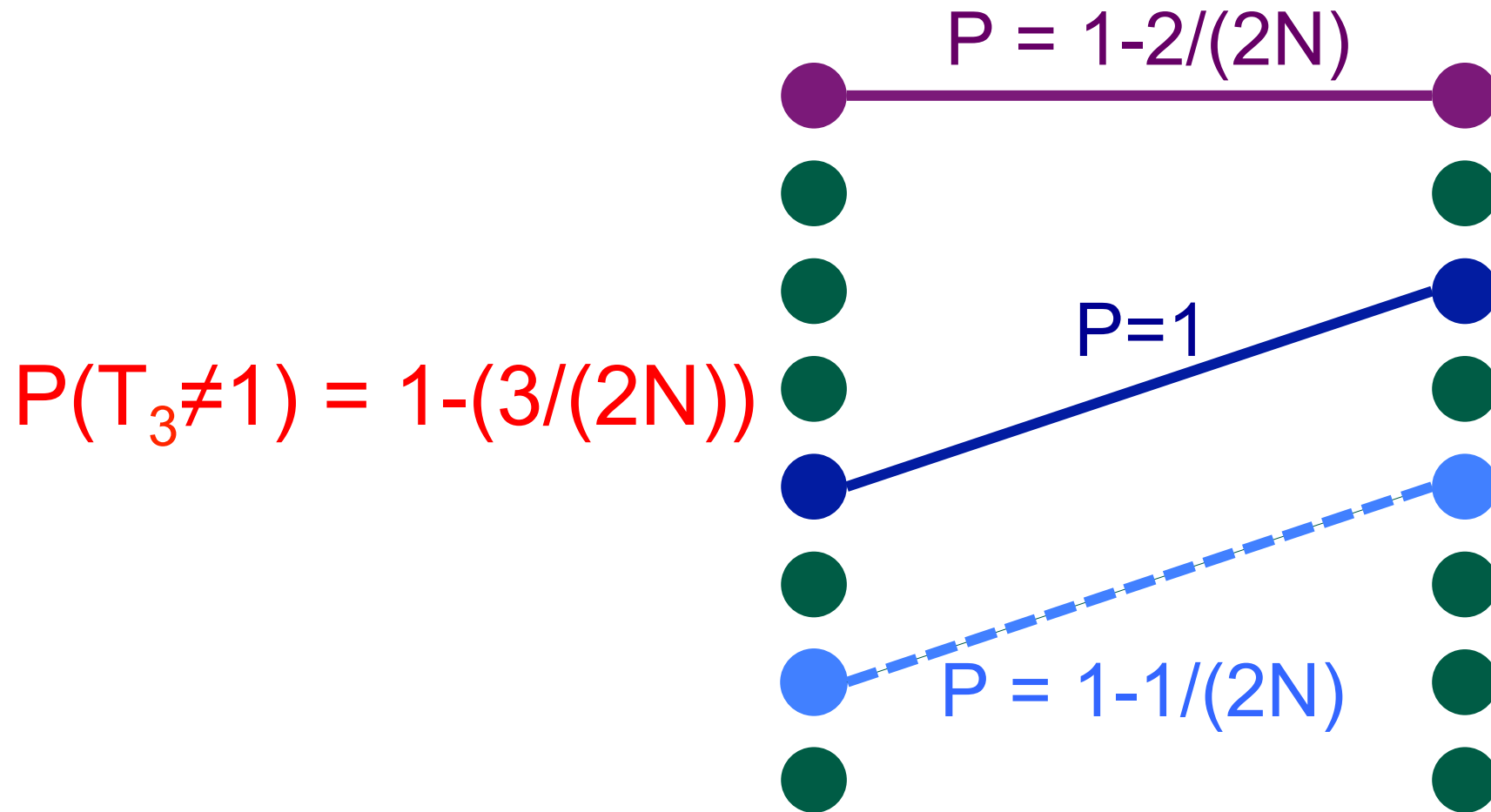
- $P(T_2=2) = P(T_2 \neq 1) \times P(T_2=1)$
- $P(T_2=3) = P(T_2 \neq 1) \times P(T_2 \neq 1) \times P(T_2=1)$
- $P(T_2=4) = P(T_2 \neq 1) \times P(T_2 \neq 1) \times P(T_2 \neq 1) \times P(T_2=1)$
- $P(T_2=j) = P(T_2 \neq 1)^{(j-1)} \times P(T_2=1)$
- $P(T_2=j) = (1-(1/(2N)))^{(j-1)} \times (1/(2N))$

## The $n$ -coalescent: $k > 2$

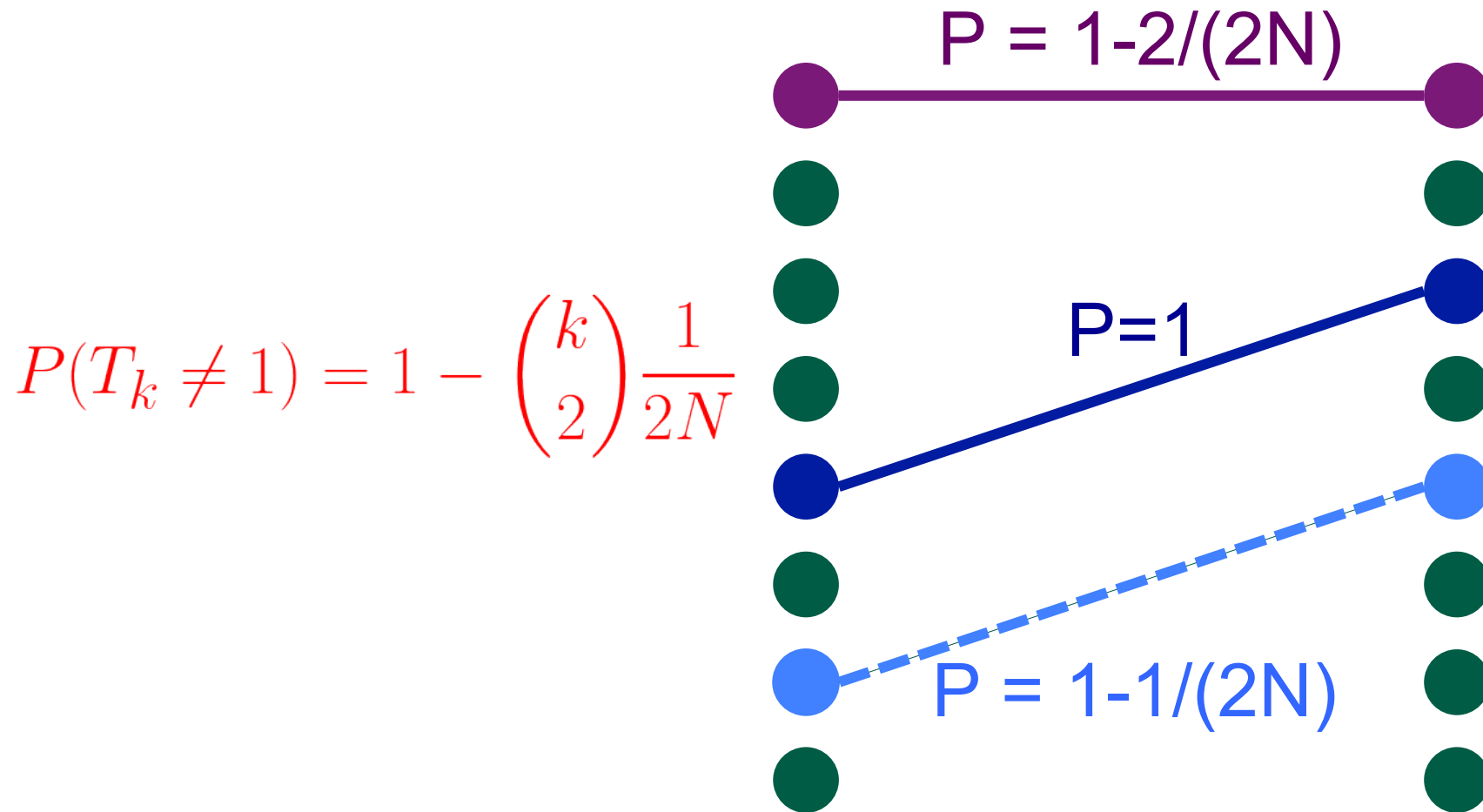




## The $n$ -coalescent: $k > 2$



## The $n$ -coalescent: $k > 2$




$$P(T_k = j)$$

$$P(T_k = j) = \left\{ 1 - \binom{k}{2} \frac{1}{2N} \right\}^{j-1} \binom{k}{2} \frac{1}{2N}$$

Thus...

$$T_k \sim \text{Geom}(p)$$

$$P(T_k = j) = (1 - p)^{j-1} p$$

$$p = \binom{k}{2} \frac{1}{2N}$$



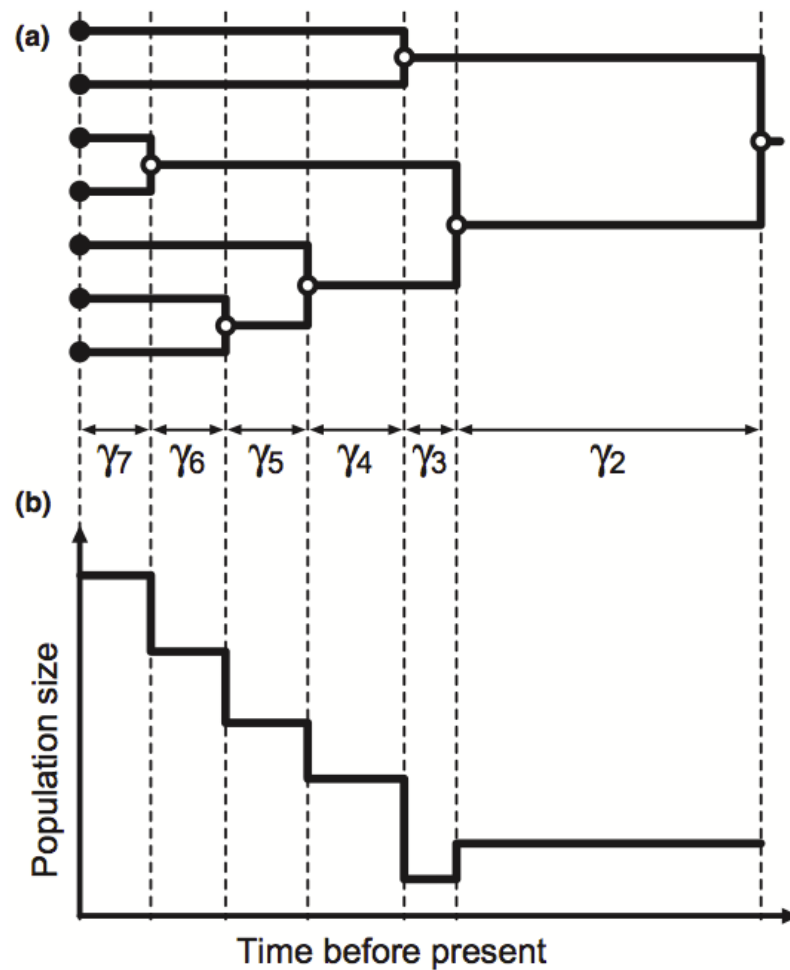
**LET'S TRY IT IN *R*...**

## The point of the coalescent...

- **Given a topology (genealogy)**, we can estimate:
  - Effective population size
  - Migration rates
  - Population size changes
  - Mutation rates
  - Age of mutations
  - Recombination rates
  - Time of divergence



# Estimating population growth...



$$N_i = \gamma_i \frac{i(i-1)}{2}$$

Ho & Shapiro (2011) *MER* 11: 423





# LET'S ESTIMATE POPULATION GROWTH IN *R*...

## Further reading

- Hein, Schierup and Wiuf (2010). Gene genealogies, variation and evolution: a primer in coalescent theory. Oxford University Press.
- Wakeley (2009). Coalescent theory: an introduction. Roberts & Company.