Introgression & Deriving Patterson's D

BIOL 1435

March 7, 2023

Overview

1. Motivation

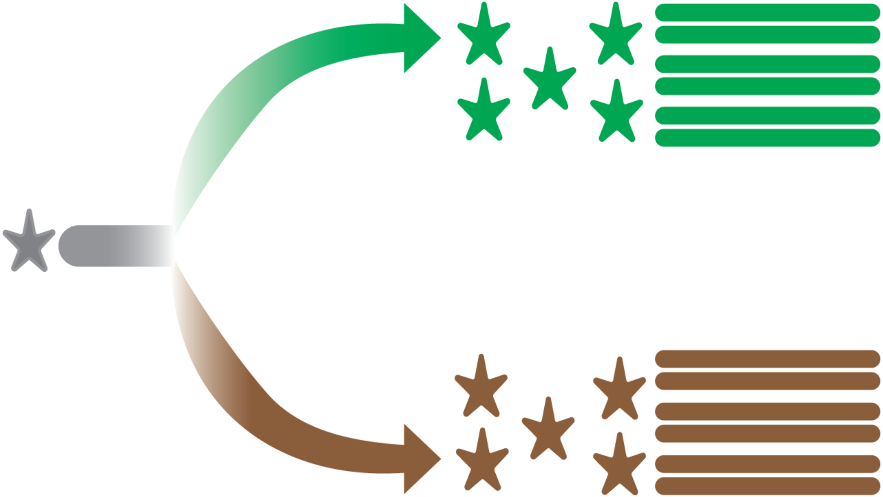
2. Preliminaries

3. Derivations

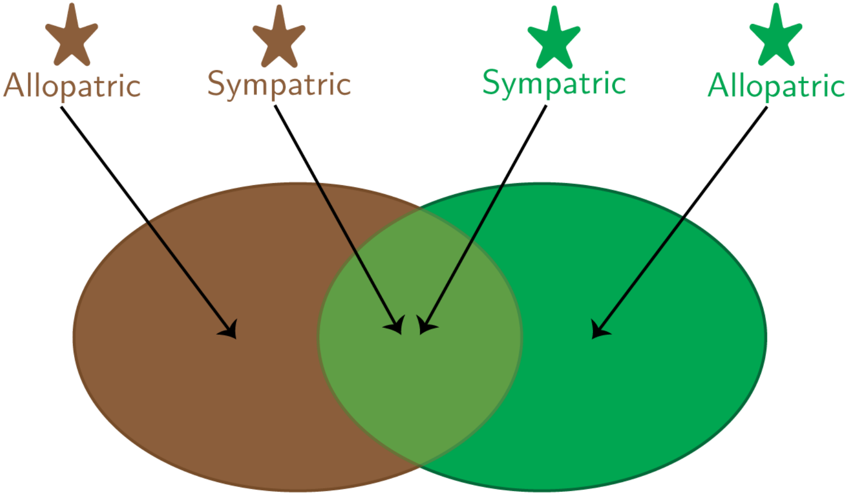
Overview

1. **Motivation**
2. Preliminaries
3. Derivations

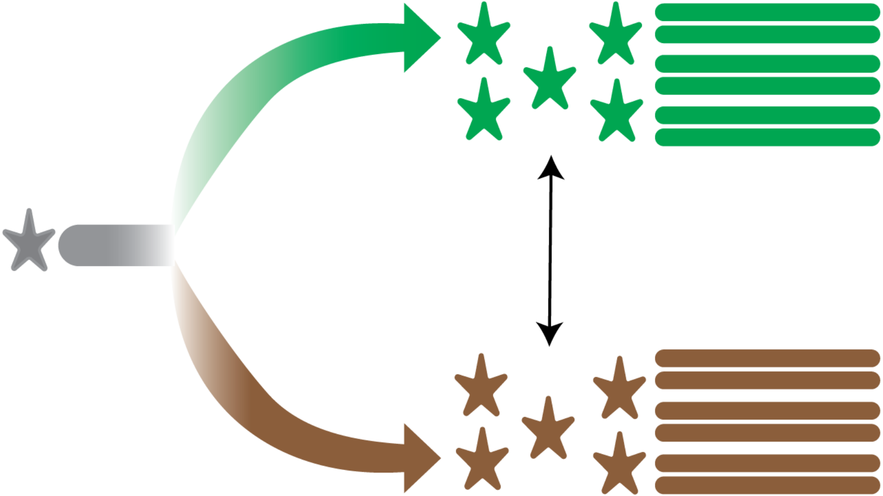
Evolution as a bifurcating process



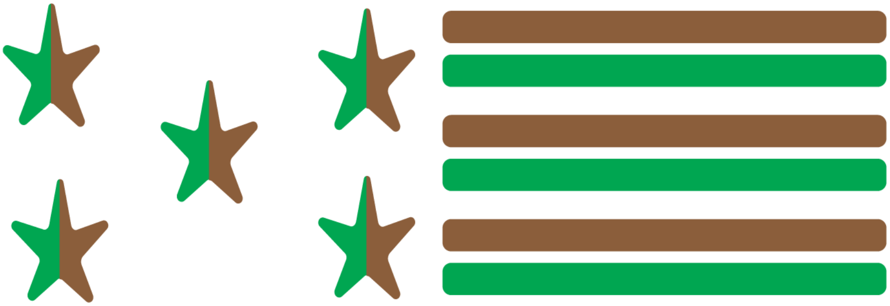
Sympatry is necessary for gene flow



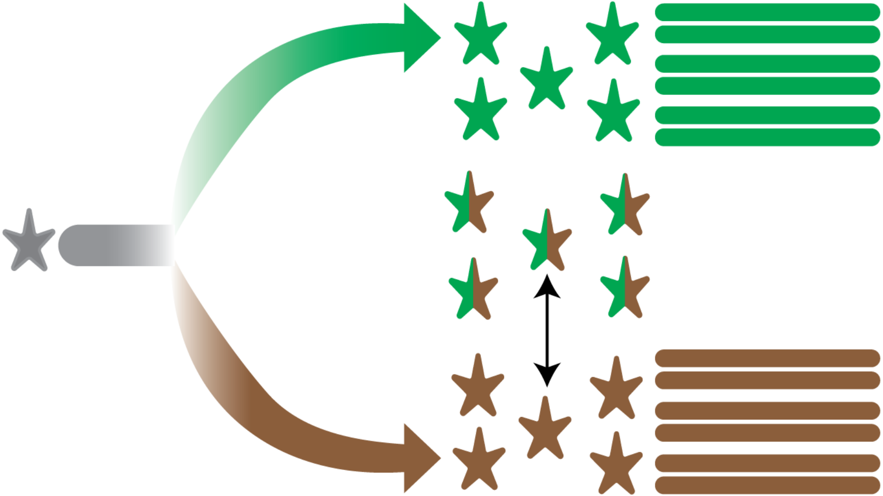
Incomplete RI leads to hybridization



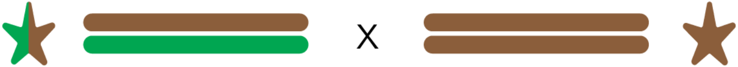
F1 hybrids = uniform mixture of parental ancestry



Hybridization is NOT sufficient for introgression



Incorporation of heterospecific loci via backcrossing



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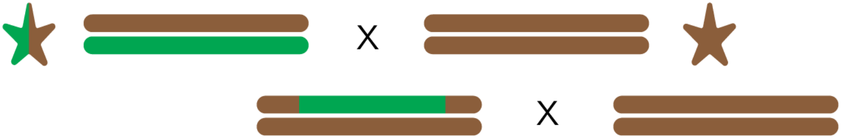

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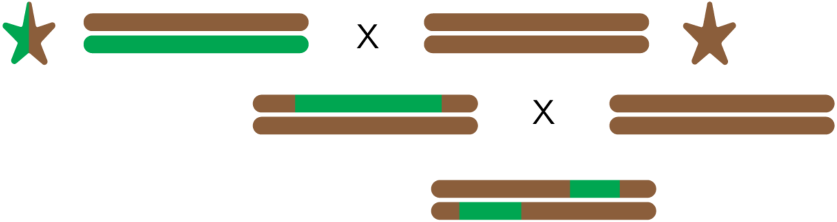
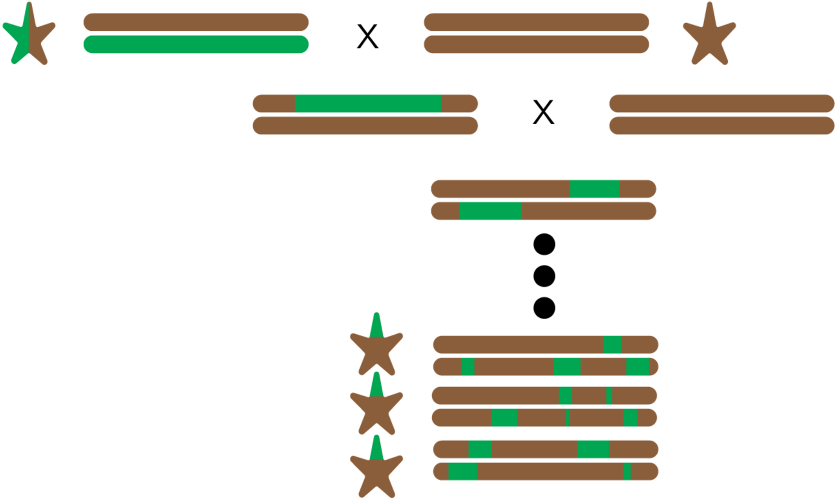
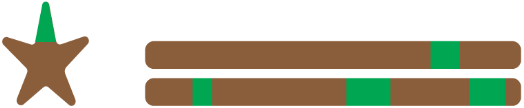


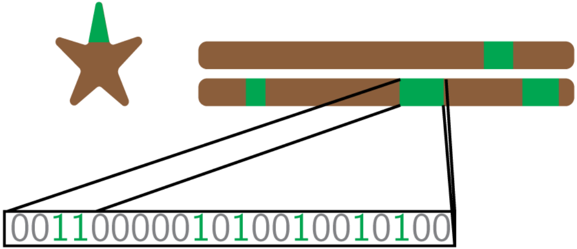
Incorporation of heterospecific loci via backcrossing



Genetic composition of an introgressed segment



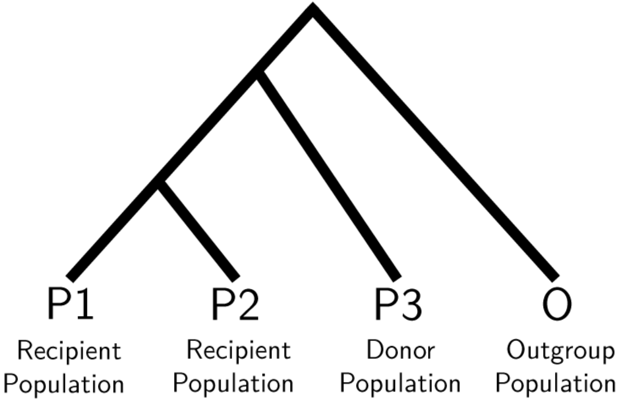
Introgression segments leaves a genomic footprint



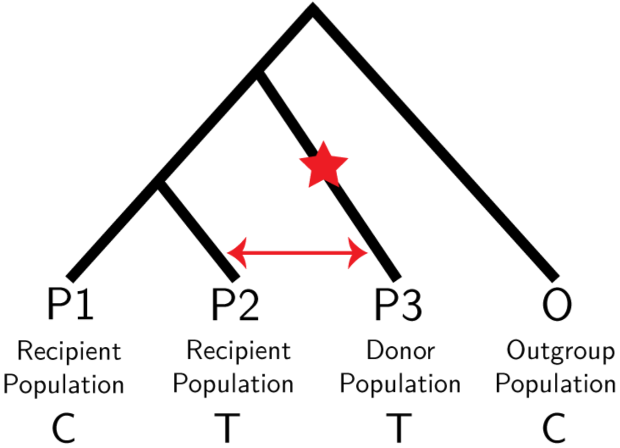
0 = reintroduced ancestral allele

1 = newly introduced derived allele

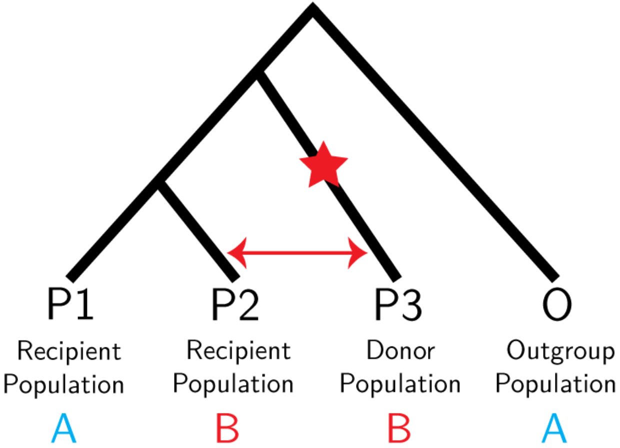
Site pattern tests of introgression



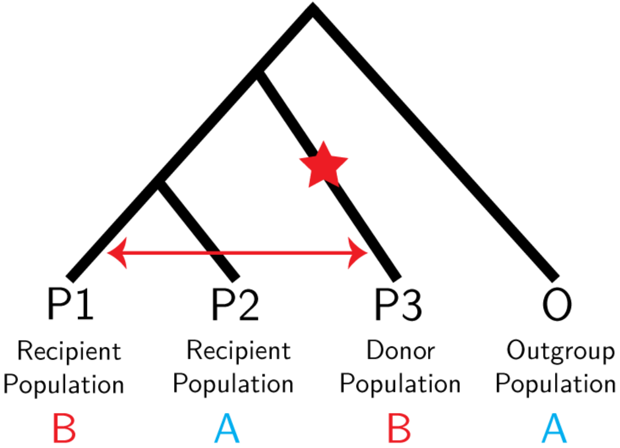
Derived allele sharing (P2 $\longleftrightarrow$ P3)



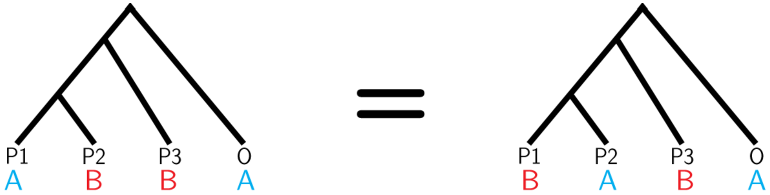
Derived allele sharing (P2 ⟷ P3)



Derived allele sharing (P1 ⟷ P3)



Patterson's D



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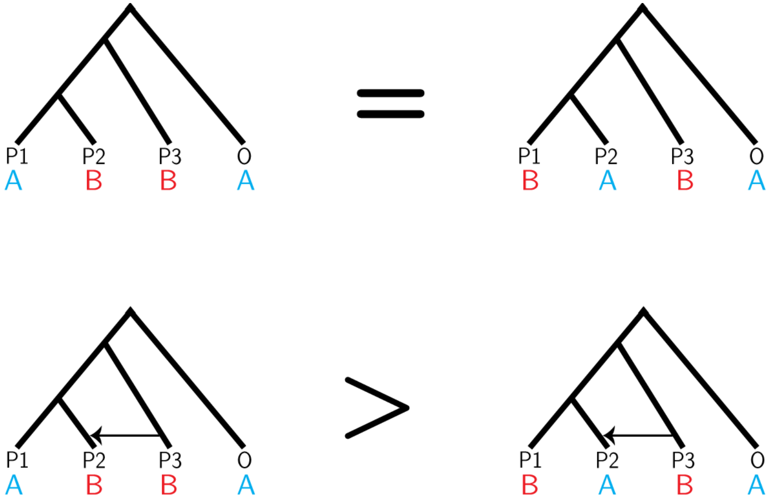

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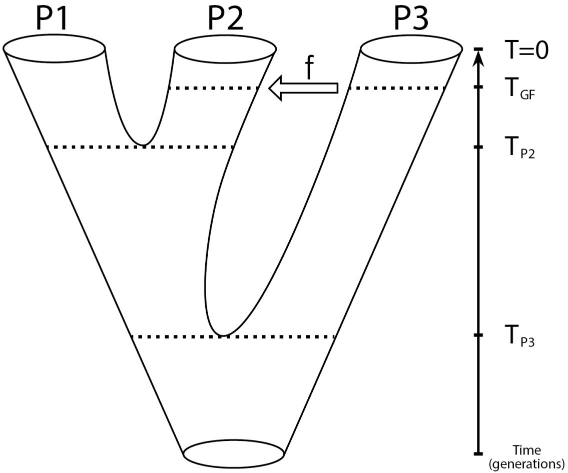
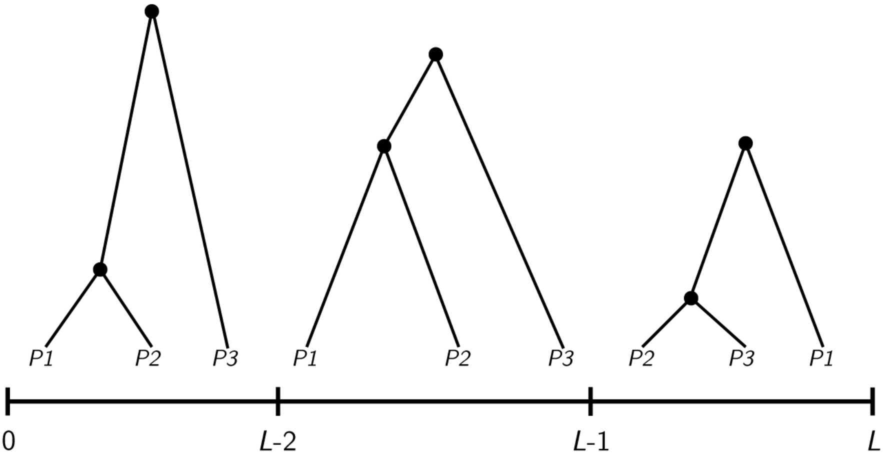
Species tree model



Each locus has its own coalescent history



Probability of gene flow

Equation

$$Pr\left(gene\ flow\right) = f \tag{1}$$

$$Pr\left(no\ gene\ flow\right) = \left(1 - f\right) \tag{2}$$

Where f represents the admixture proportion—the probability that any lineage from *P3* migrates to *P2*.

Probability of no coalescence during time interval t

Probability of no coalescence during time interval t

Equation

$$Pr\left(no\ coalescences\right) = \left(1 - \frac{1}{2N}\right)^{t} \tag{3}$$

Where t denotes the time interval where coalescence can occur.

Probability of coalescence during time interval t

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Equation

$$Pr\left(coalescences\right) = 1 - \left(1 - \frac{1}{2N}\right)^{t} \tag{4}$$

Where t denotes the time interval where coalescence can occur.

Expected time of coalescence T_2 & T_3

Expected time of coalescence T_2 & T_3

Equation

$$T_2 = 2N \tag{5}$$

$$T_3 = \frac{2N}{3} \tag{6}$$

Expected time of coalescence | coalescence during t

Expected time of coalescence | coalescence during t

Equation

$$\bar{t} \sim Tgeo\left(t|p, c\right) \tag{7}$$

Expected time of coalescence | coalescence during t

Equation

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$$f\left(\bar{t}\right) = \frac{\frac{1}{2N}\left(1 - \frac{1}{2N}\right)^{t-1}}{1 - \left(1 - \frac{1}{2N}\right)^{c}} \tag{8}$$

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$$\mathbb{E}\left(\bar{t}\right) = \sum_{t=1}^{c} t \frac{\frac{1}{2N}\left(1 - \frac{1}{2N}\right)^{t-1}}{1 - \left(1 - \frac{1}{2N}\right)^{c}} = \frac{2N - \left(\left(1 - \frac{1}{2N}\right)^{c}\left(c + 2N\right)\right)}{1 - \left(1 - \frac{1}{2N}\right)^{c}} \tag{9}$$

Where c denotes the time interval where coalescence must occur.

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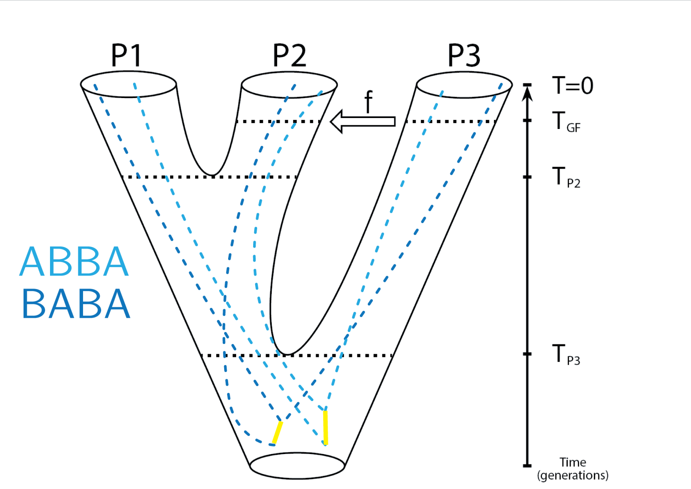
Coalescent history 1

No gene flow from *P3* → *P2*, *P1* & *P2* don't coalesce between T_{P2} & T_{P3}, and *P1* & *P2* don't coalesce.



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$$Pr\left(no\ gene\ flow\right) = (1 - f) \tag{10}$$

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$$Pr\left(P1\ \&\ P2\ don't\ coalesce\ between\ T_{P2}\ \&\ T_{P3}\right) = \left(1 - \frac{1}{2N}\right)^{T_{P3} - T_{P2}} \tag{11}$$

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$$Pr\left(\textit{P1 \& P2 don't coalesce}\right) = \frac{1}{3} \tag{12}$$

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$$\mathbb{E}\left(Branch\ length\ between\ the\ 1^{st}\ \&\ 2^{nd} coalescent\ event\right) = 2N \tag{13}$$

Coalescent history 1

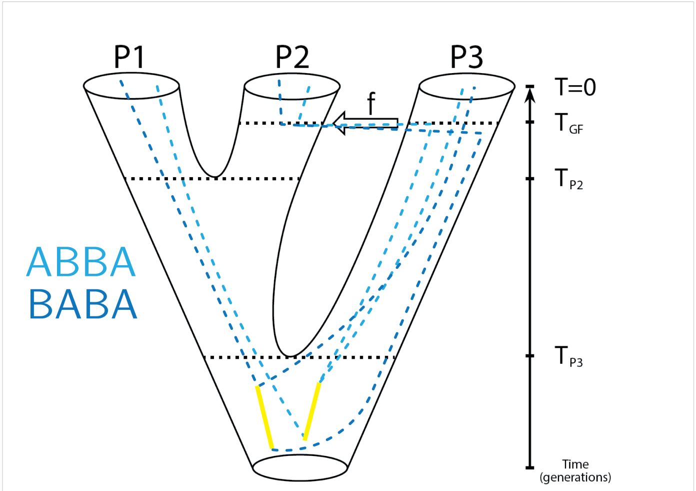
No gene flow from $P3 \rightarrow P2$, *P1* & *P2* don't coalesce between T_{P2} & T_{P3}, and *P1* & *P2* don't coalesce.

Derivation

$$\mathbb{E}\left(C_{ABBA_1}\right) = \mathbb{E}\left(C_{BABA_1}\right) = (1 - f)\left(1 - \frac{1}{2N}\right)^{T_{P3} - T_{P2}} \frac{2N}{3} \tag{14}$$

Coalescent history 2

Gene flow from *P3* → *P2*, *P2* & *P3* don't coalesce between T_{GF} & T_{P3}, and *P1* & *P2* don't coalesce.



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$$Pr\left(P1\ \&\ P2\ don't\ coalesce\right) = \frac{1}{3} \tag{17}$$

$$\mathbb{E}\left(Branch\ length\ between\ the\ 1^{st}\ \&\ 2^{nd} coalescent\ event\right) = 2N \tag{18}$$

Coalescent history 2

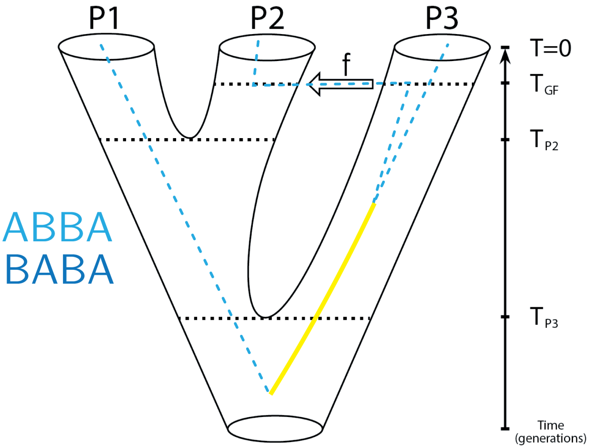
Gene flow from $P3 \rightarrow P2$, *P2* & *P3* don't coalesce between T_{GF} & T_{P3}, and *P1* & *P2* don't coalesce.

Derivation

$$\mathbb{E}\left(C_{ABBA_2}\right) = \mathbb{E}\left(C_{BABA_2}\right) = f\left(1 - \frac{1}{2N}\right)^{T_{P3} - T_{GF}} \frac{2N}{3} \tag{19}$$

Coalescent history 3

Gene flow from $P3 \rightarrow P2$ and $P2$ & $P3$ coalesce between T_{GF} & T_{P3}.



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Derivation

$$Pr\left(gene\ flow\right) = f \tag{20}$$

$$Pr\left(P2\ \&\ P3\ coalesce\ between\ T_{GF}\ \&\ T_{P3}\right) = 1 - \left(1 - \frac{1}{2N}\right)^{T_{P3}-T_{GF}} \tag{21}$$

Coalescent history 3

Gene flow from $P3 \rightarrow P2$ and $P2$ & $P3$ coalesce between T_{GF} & T_{P3}.

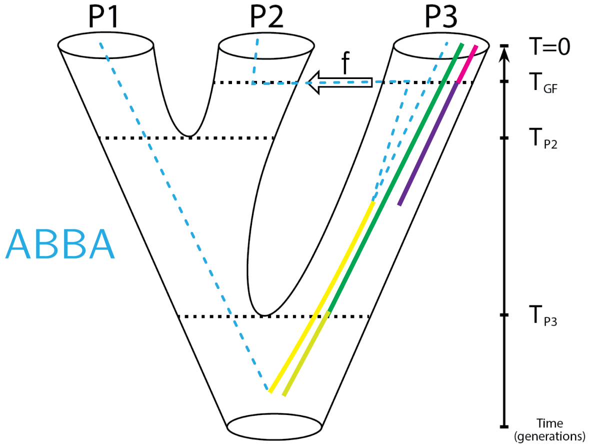
Derivation

$$Pr\left(gene\ flow\right) = f \tag{20}$$

$$Pr\left(P2\ \&\ P3\ coalesce\ between\ T_{GF}\ \&\ T_{P3}\right) = 1 - \left(1 - \frac{1}{2N}\right)^{T_{P3} - T_{GF}} \tag{21}$$

$$\mathbb{E}\left(Branch\ length\ between\ the\ 1^{st}\ \&\ 2^{nd} coalescent\ event\right) = \left(T_{P3} + 2N\right) - \left(T_{GF} + \overline{t}\right) \tag{22}$$

Coalescent history 3 ($(\bar{t})$)



$$\text{IBL} = T_{P3} + 2N - (T_{GF} + \bar{t})$$

Coalescent history 3

Gene flow from $P3 \rightarrow P2$ and $P2$ & $P3$ coalesce between T_{GF} & T_{P3}.

Derivation

$$\mathbb{E}\left(C_{ABBA_3}\right) = f\left(1 - \left(1 - \frac{1}{2N}\right)^{T_{P3} - T_{GF}}\right)\left(\left(T_{P3} + 2N\right) - \left(T_{GF} + \overline{t}\right)\right) \tag{23}$$

Coalescent history 3

Gene flow from $P3 \rightarrow P2$ and $P2$ & $P3$ coalesce between T_{GF} & T_{P3}.

Derivation

$$\mathbb{E}\left(C_{ABBA_3}\right) = f\left(1 - \left(1 - \frac{1}{2N}\right)^{T_{P3} - T_{GF}}\right)\left(\left(T_{P3} + 2N\right) - \left(T_{GF} + \bar{t}\right)\right) \tag{23}$$

$$\mathbb{E}\left(C_{BABA_3}\right) = 0 \tag{24}$$

$\mathbb{E}\left(ABBA\right)$

Derivation

$$\mathbb{E}\left(\tau_{ABBA}\right) = C_{ABBA_1} + C_{ABBA_2} + C_{ABBA_3} \tag{25}$$

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Derivation

$$\mathbb{E}\left(\tau_{ABBA}\right) = C_{ABBA_1} + C_{ABBA_2} + C_{ABBA_3} \tag{25}$$

$$\begin{aligned}\mathbb{E}\left(\tau_{ABBA}\right) = {} & (1 - f)\left({}^{2N}\!/_{3}\left(1 - {}^{1}\!/_{2N}\right)^{T_{P3} - T_{P2}}\right) \\ & + (f)\left(\left({}^{2N}\!/_{3}\left(1 - {}^{1}\!/_{2N}\right)^{T_{P3} - T_{GF}}\right) + \left(T_{P3} - T_{GF}\right)\right)\end{aligned} \tag{26}$$

$\mathbb{E}\left(ABBA\right)$

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$$\mathbb{E}\left(\tau_{ABBA}\right) = C_{ABBA_1} + C_{ABBA_2} + C_{ABBA_3} \tag{25}$$

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$$\mathbb{E}\left(ABBA_{sites}\right) = \mathbb{E}\left(\tau_{ABBA}\right) \times \mu \times L \tag{27}$$

Where μ represents the mutation rate and L represents the sequence length.

$\mathbb{E}\left(BABA\right)$

Derivation

$$\mathbb{E}\left(\tau_{BABA}\right) = C_{BABA_1} + C_{BABA_2} \tag{28}$$

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Derivation

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$$\begin{aligned}\mathbb{E}\left(\tau_{BABA}\right) = (1-f)&\left({}^{2N}\!/_{3}\left(1-{}^{1}\!/_{2N}\right)^{T_{P3}-T_{P2}}\right)\\ +(f)&\left({}^{2N}\!/_{3}\left(1-{}^{1}\!/_{2N}\right)^{T_{P3}-T_{GF}}\right)\end{aligned} \tag{29}$$

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Derivation

$$\mathbb{E}\left(\tau_{BABA}\right) = C_{BABA_1} + C_{BABA_2} \tag{28}$$

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$$\mathbb{E}\left(BABA_{sites}\right) = \mathbb{E}\left(\tau_{BABA}\right) \times \mu \times L \tag{30}$$

Where μ represents the mutation rate and L represents the sequence length.

Patterson's D

Derivation

$$\mathbb{E}\left(D\right) = \frac{\mathbb{E}\left(\tau_{ABBA}\right) - \mathbb{E}\left(\tau_{BABA}\right)}{\mathbb{E}\left(\tau_{ABBA}\right) + \mathbb{E}\left(\tau_{BABA}\right)} \tag{31}$$

Patterson's D

Derivation

$$\mathbb{E}\left(D\right) = \frac{\mathbb{E}\left(\tau_{ABBA}\right) - \mathbb{E}\left(\tau_{BABA}\right)}{\mathbb{E}\left(\tau_{ABBA}\right) + \mathbb{E}\left(\tau_{BABA}\right)} \tag{31}$$

$$\mathbb{E}\left(D\right) = \frac{\left(f\right)\left(T_{P3} - T_{GF}\right)}{\left(1-f\right)\left[{}^{4N}\!/_{3}\left(1 - {}^{1}\!/_{2N}\right)^{T_{P3}-T_{P2}}\right] + \left(f\right)\left[\left({}^{4N}\!/_{3}\left(1 - {}^{1}\!/_{2N}\right)^{T_{P3}-T_{GF}}\right) + \left(T_{P3} - T_{GF}\right)\right]} \tag{32}$$

Patterson's D

Derivation

$$\mathbb{E}\left(D\right) = \frac{\mathbb{E}\left(\tau_{ABBA}\right) - \mathbb{E}\left(\tau_{BABA}\right)}{\mathbb{E}\left(\tau_{ABBA}\right) + \mathbb{E}\left(\tau_{BABA}\right)} \tag{31}$$

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$$\mathbb{E}\left(D\right) = \frac{\sum_{i=1}^{L}(1-p_{i1})p_{i2}p_{i3}(1-p_{iO}) - p_{i1}(1-p_{i2})p_{i3}(1-p_{iO})}{\sum_{i=1}^{L}(1-p_{i1})p_{i2}p_{i3}(1-p_{iO}) + p_{i1}(1-p_{i2})p_{i3}(1-p_{iO})} \tag{33}$$

Where $p_{i\#}$ represents the derived allele frequency at site i and L represents the sequence length.