Population Structure

BIOL 1435

February 23, 2023

Overview

1. Quiz Review
2. Population Differentiation & Structure
3. Relative Differentiation
4. Absolute Differentiation
5. Population Structure

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Q: What is a population?

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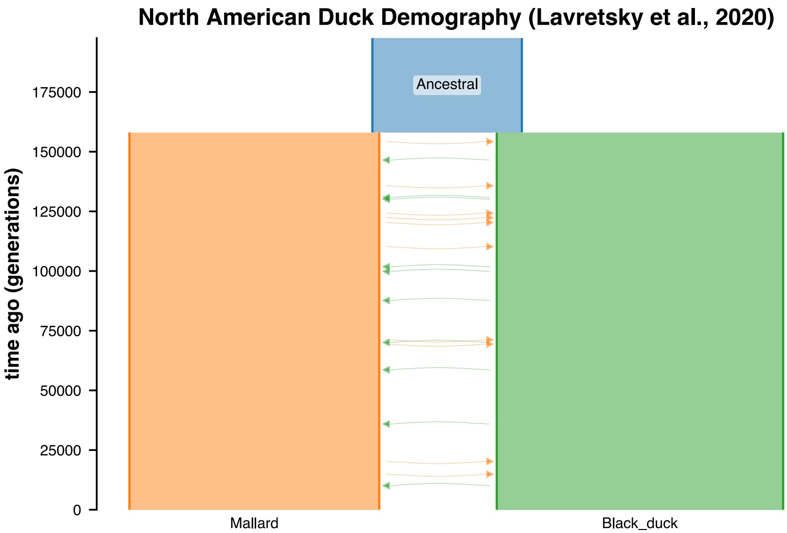
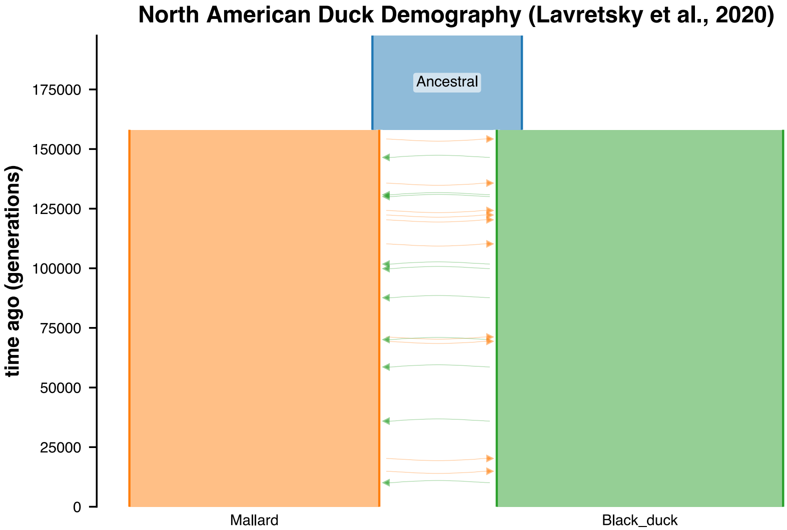
A: A group of freely interbreeding individuals.

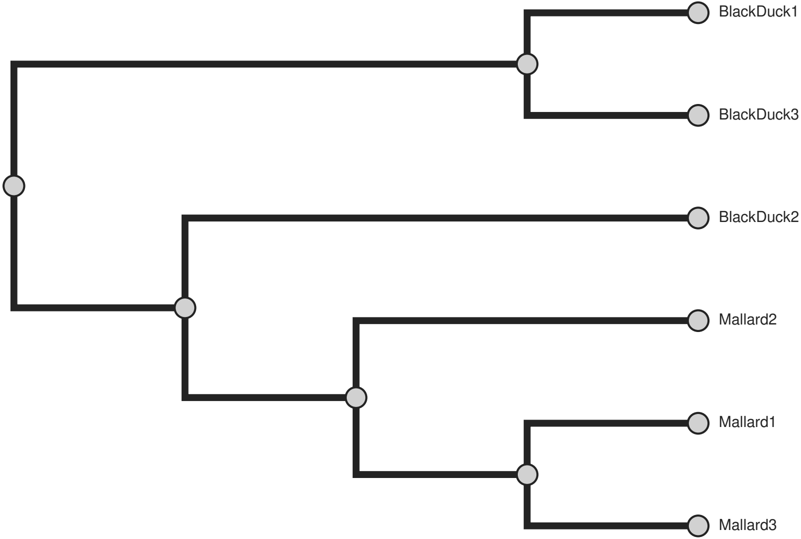
Populations



Differentiated Populations



Population Structure



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F_{ST}

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Definition

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OR

F_{ST} quantifies genetic drift between two populations relative to the average drift between the two populations.

Hudon's Estimator of F_{ST}

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Equation

$$F_{ST} = \frac{N}{D} \tag{1}$$

$$N = \left(p_1 - p_2\right)^2 \tag{2}$$

$$D = p_1\left(1 - p_2\right) + p_2\left(1 - p_1\right) \tag{3}$$

Where p_i is the frequency of the derived/alternative allele at a given site from the i^{th} population.

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$$F_{ST} = \frac{\sum_{j=1}^{L} N_j}{\sum_{j=1}^{L} D_j} \tag{4}$$

Where L is to the total number of sites.

Understanding F_{ST}

Understanding F$_{ST}$

Interpretation

$$F_{ST} = 0 : \text{ no differentiation}$$

$$F_{ST} = 1 : \text{ maximum differentiation}$$

Limitations of F_{ST}

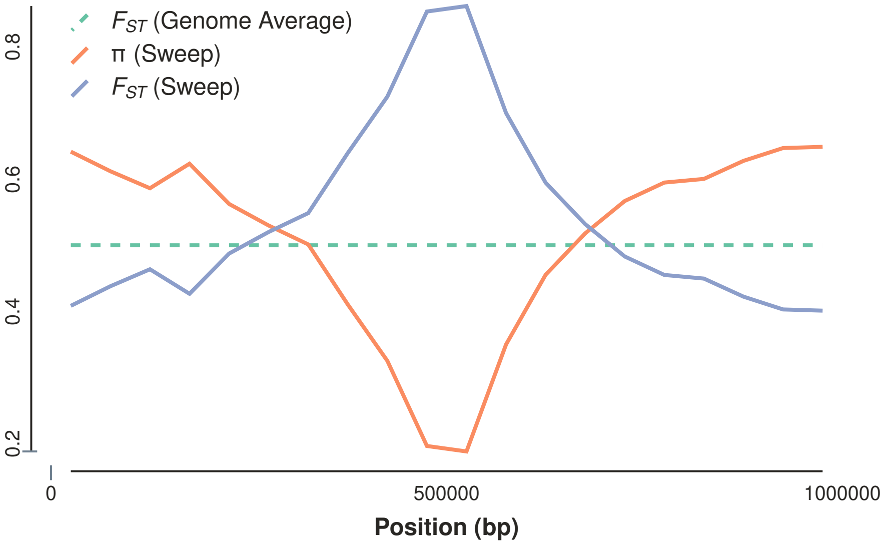
Q: Why is F_{ST} a relative measure of population differentiation?

Limitations of F_{ST}

Q: Why is F_{ST} a relative measure of population differentiation?

A: F_{ST} is strongly influenced by within-subpopulation levels of diversity!

Linked-Selection & F_{ST}



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d_{XY}

$\mathbf{d_{XY}}$

Definition

d_{XY} average number of pairwise differences between chromosomes from two populations.

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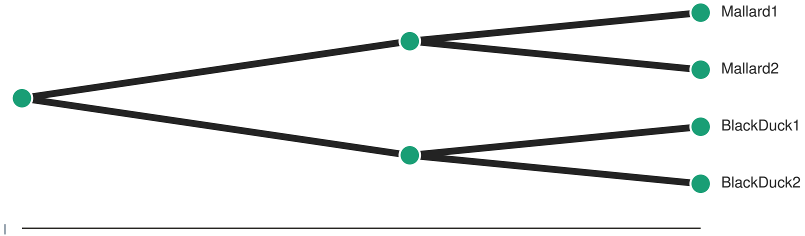
Equation

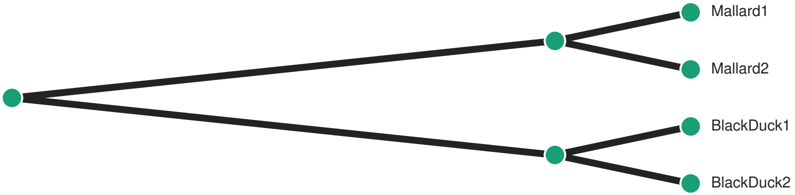
$$d_{XY} = \frac{1}{n_X n_Y} \sum_{i=1}^{n_X} \sum_{j=1}^{n_y} k_{ij} \tag{5}$$

Where n_X and n_Y correspond to the number of chromosomes in populations X and Y, , and k_{ij} is the number of nucleotide differences between the i^{th} and j^{th} chromosome.

Advantages of d_{XY}



d_{XY}



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Identifying Population Structure

Q: Given a set of samples how can we assess if there is population structure?

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A: Principle Component Analysis (PCA)

PCA Steps

1. Zero-center the allele count matrix
2. Calculate the covariance matrix
3. Perform eigendecomposition

The Allele Count Matrix (C)

$$\mathbf{C}\left(i,j\right)=\begin{bmatrix} c_{11} & c_{12} & \cdots & c_{1j} \\ c_{21} & c_{22} & \cdots & c_{2j} \\ \vdots & \vdots & \ddots & \vdots \\ c_{i1} & c_{i2} & \cdots & c_{ij} \end{bmatrix}$$

Where $\mathbf{C}\left(i,j\right)$ consists of m (individuals) $\times$ n (sites), and $\mathbf{C}\left(i,j\right) \in \{\ 0,1,2\}$ alternative/derived allele count.

The Standardized Allele Count Matrix (M)

$$\mu(j) = \frac{\sum_{i=1}^{m} \mathbf{C}(i,j)}{m}$$

$$\mathbf{M}\,(i,j) = \frac{\mathbf{C}(i,j)-\mu(j)}{\sqrt{(m\times 2)p(j)(1-p(j))}}$$

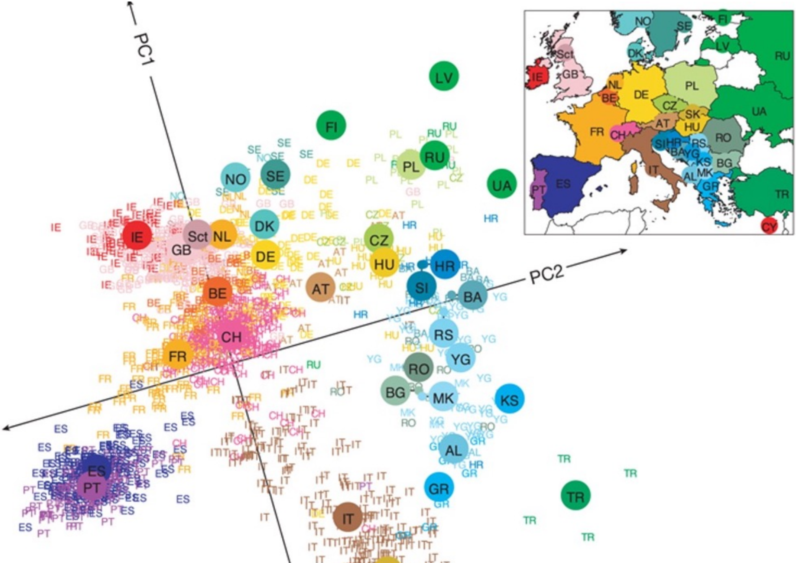
Where $\mu(j)$ is the column mean, $p(j) = \mu(j) \div 2$ is the allele frequency for site j, and $\sqrt{(m \times 2)\, p(j)\, (1 - p(j))}$ is the standard deviation of the binomial distribution and is proportional to the rate change in allele frequency per generation due to genetic drift.

Covariance Matrix (X)

$$\mathbf{X} = \frac{1}{m-1}\mathbf{M}\mathbf{M}^{\mathbf{T}}$$

Where X is a square matrix consisting of the the covariance between all individuals.

PCA In Action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