CELS191 Cell & Molecular Biology
Te Mātau Koiora o te Pūtau me te Rāpoi Kota

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Research & Interests:

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CELS191 2025

Molecular Biology & Genetics

Lecture 21

Population Genetics & Natural Selection

Professor Jon Waters
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Lecture 21 Objectives

After you have revised this lecture you should be able to:

- Calculate genotype frequencies using Hardy-Weinberg equilibrium.
- Identify that random genetic drift occurs rapidly in small populations.
- Explain how populations evolve over time in response to selection.

POPULATIONS EVOLVE, INDIVIDUALS DON'T

- **Population:** localised group of individuals of the same species

- **Gene Pool:** total aggregate of genes (and their alleles) in the population at one time

Why might we need to estimate frequencies of genotypes in a population?

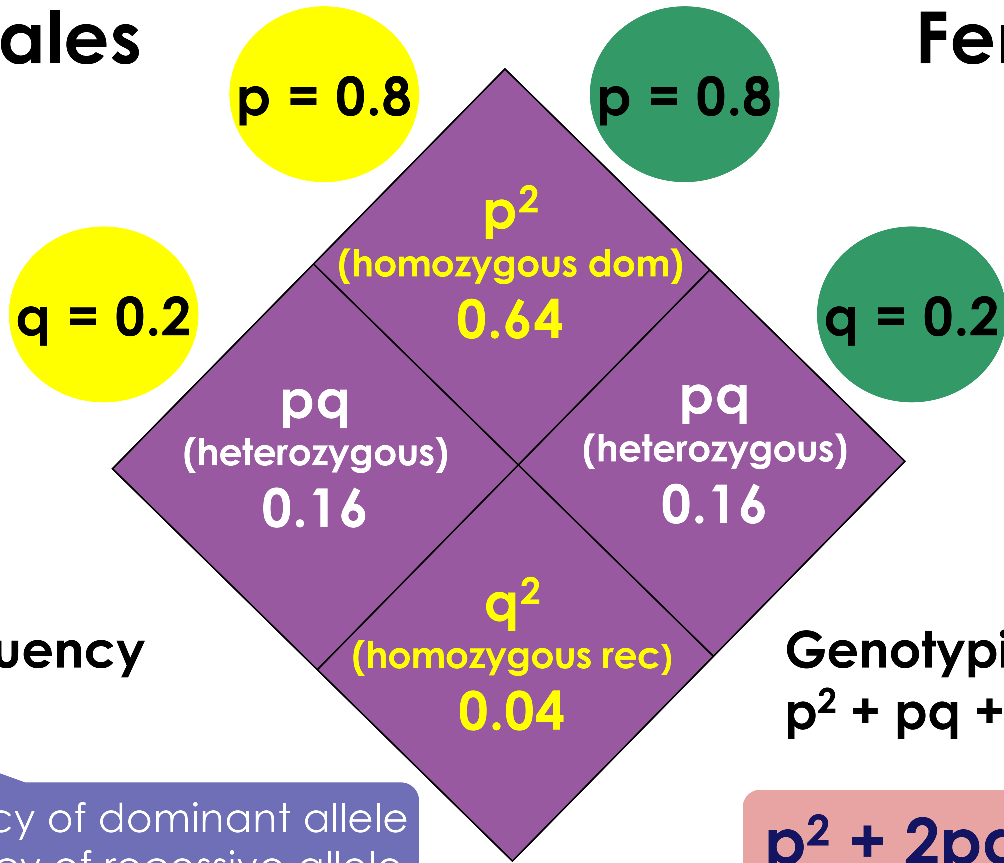
1. To predict how many individuals will inherit a genetic disease

2. To estimate the proportion of individuals who are ‘carriers’ of a genetic disease

HARDY-WEINBERG EQUATION

How do we predict genotypes in a population?

Males **Females**



Allele frequency
$p + q = 1$

p = frequency of dominant allele
q = frequency of recessive allele

Genotypic frequency
$p^2 + pq + pq + q^2 = 1$

$p^2 + 2pq + q^2 = 1$

Example Question 1

1 in 2500 children are born with cystic fibrosis.
What proportion of the population are carriers for the disease?

A: Carriers = <u>heterozygous</u> = 2pq......therefore:

Estimate q:
$q^2 = 1/2500$
$q = 1/50 = 0.02$
(recessive allele)

Estimate p:
$1 - q = p = 0.98$
(dominant allele)

Estimate 2pq:
$2pq = 2(0.98 \times 0.02)$
$= 0.0392$

= about 4% of the population are carriers

Example Question 2

If 1 in 10,000 children is born with Tay-Sachs disease, what percentage of the population are expected to be carriers for this autosomal recessive disease?

(A) 0.10%
(B) 0.20%
(C) 0.99%
(D) 1.98%

Estimate q:
$q^2 = 1/10000$
$q = 1/100 = 0.01$

Estimate p:
$1 - q = p = 0.99$

Estimate 2pq:
$2pq = 2(0.99 \times 0.01)$
$= 0.0198$

= about 2% of the population are carriers

Example Question 3

If the proportion of *X* chromosomes with the allele for haemophilia (a) in a population is 0.4, how many females will be affected by the disease? And how many males?

$X^a = q = 0.4$

Females = $X^aX^a = q^2 = 0{\cdot}4^2 = 0.16$ or **16%**

Males = $X^aY = q = 0.4 =$ **40%**

Remember, for an x-linked trait: females need to be homozygous recessive to get the disease; males need just one recessive allele

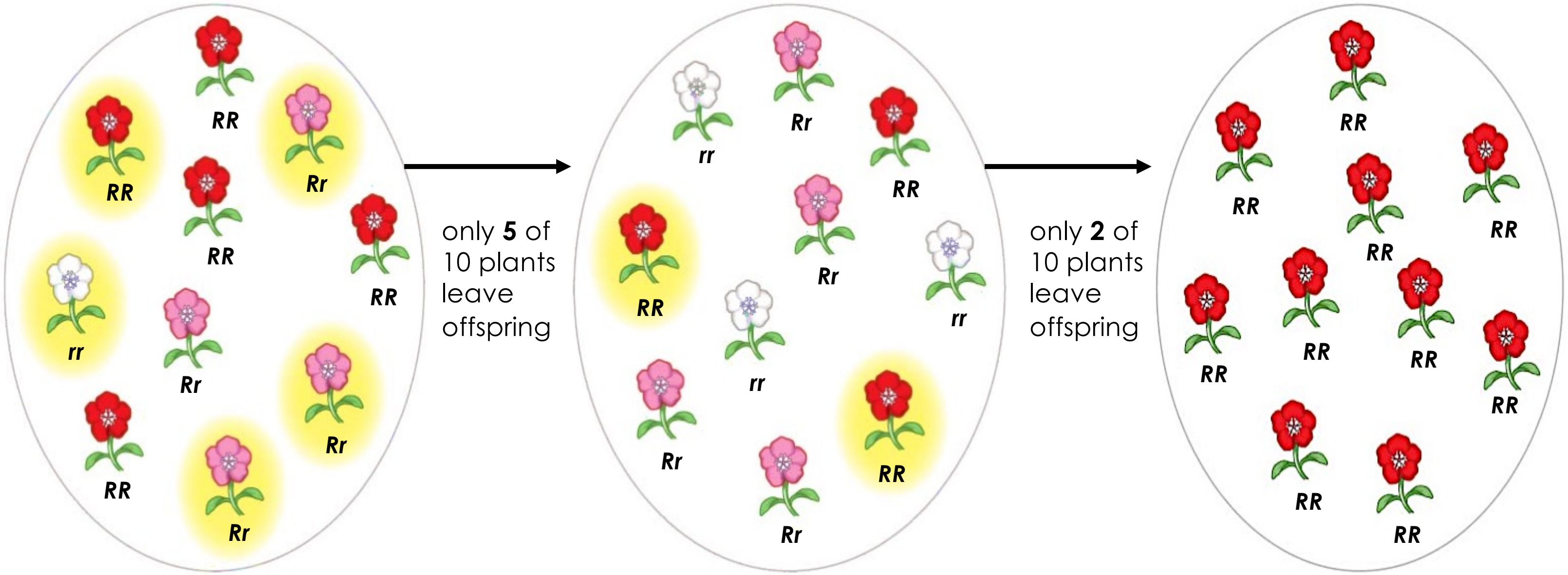
Allele frequencies can change via...

- **Non - random mating**
  - *assortative mating*
  - *inbreeding*
- **Random Genetic Drift**
- **Bottleneck Effect**
- **Founder Effect**
- **Natural Selection**
- **Gene Flow or Migration**
- **Mutation**

Modified Textbook Figure 23.9

Random Genetic Drift

A random change in allele frequencies due to sampling error over generations



Generation 1
p (frequency *R*) = 0.7
q (frequency *r*) = 0.3

Generation 2
p = 0.5
q = 0.5

Generation 3
p = 1.0
q = 0.0

The Bottleneck Effect

Original population has equal frequencies of blue and white with a few yellow

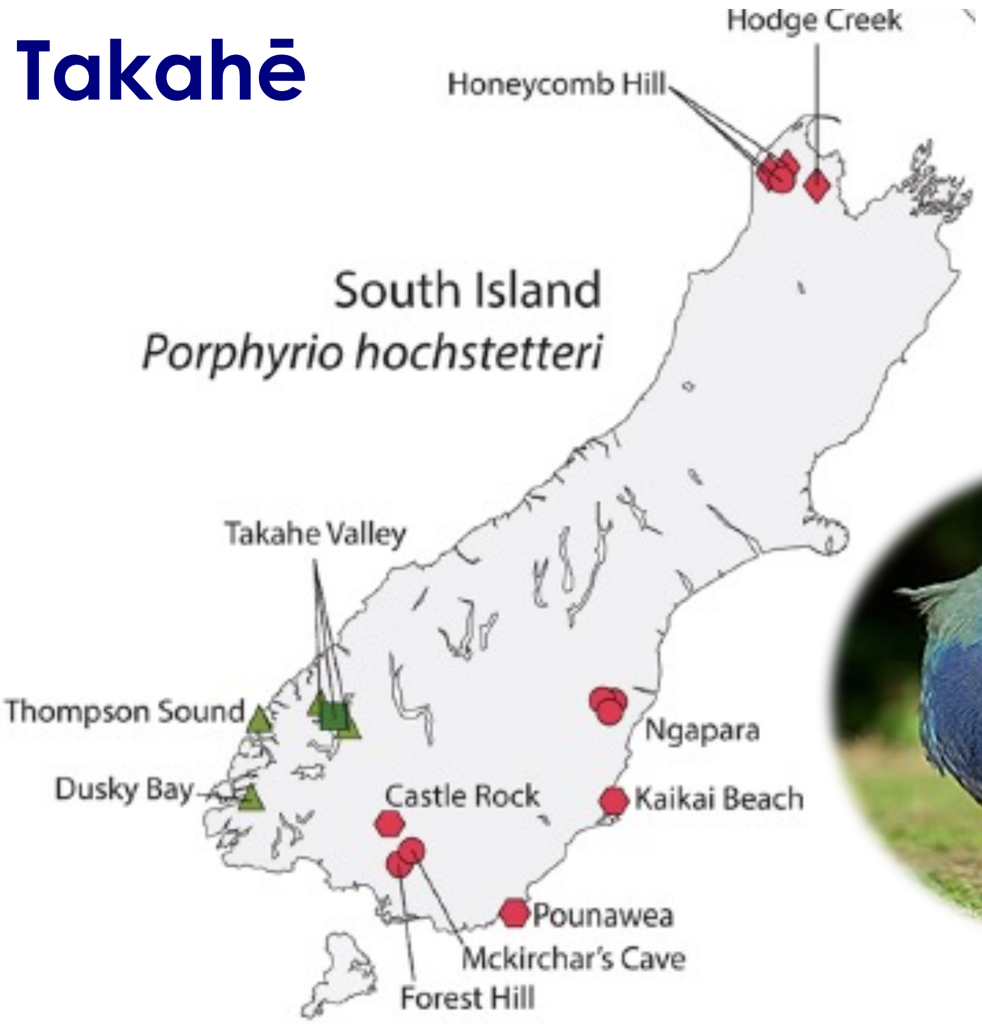
Population reduction or Bottleneck event

Yellow allele completely lost, white allele now rare

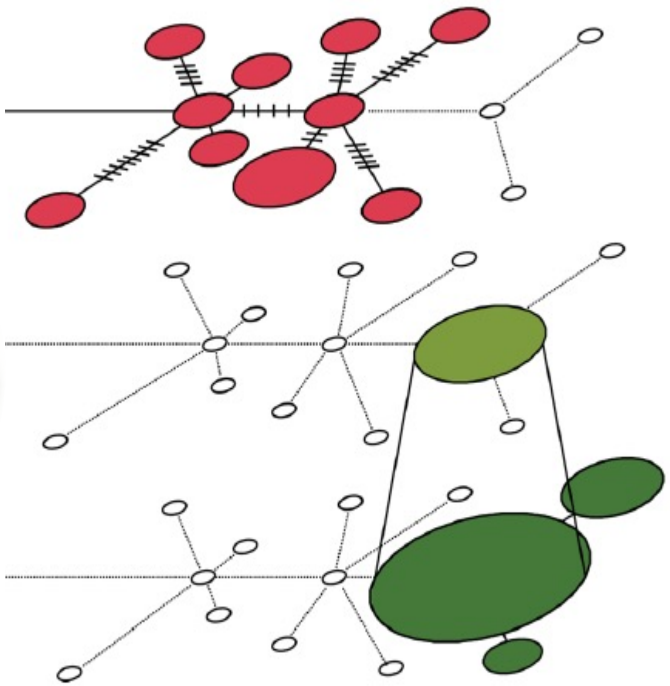
Context Slide

Image: Verry et al (2023) Molecular Ecology

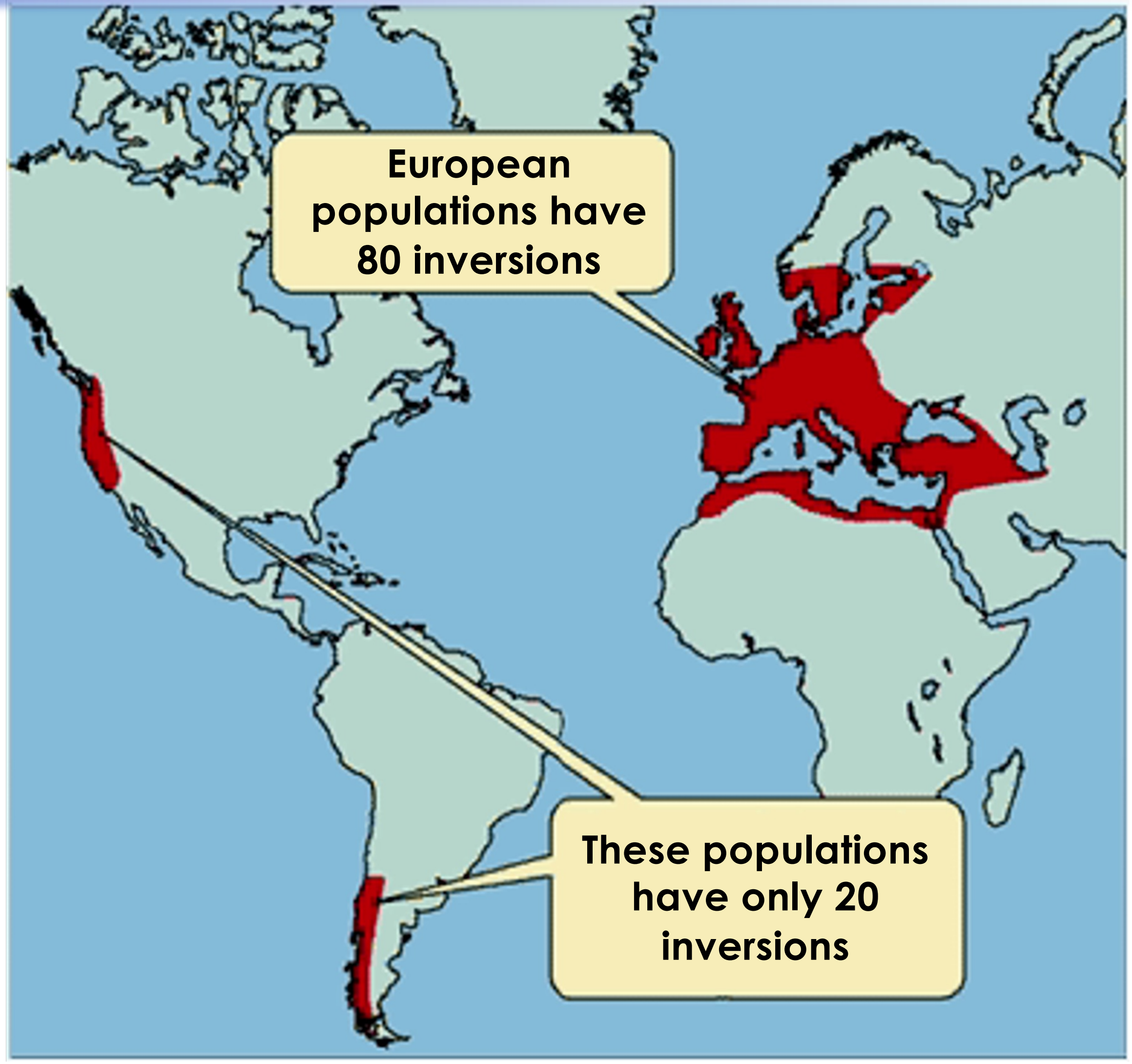
Takahē



Bottlenecks in NZ's endangered birds



The Founder Effect

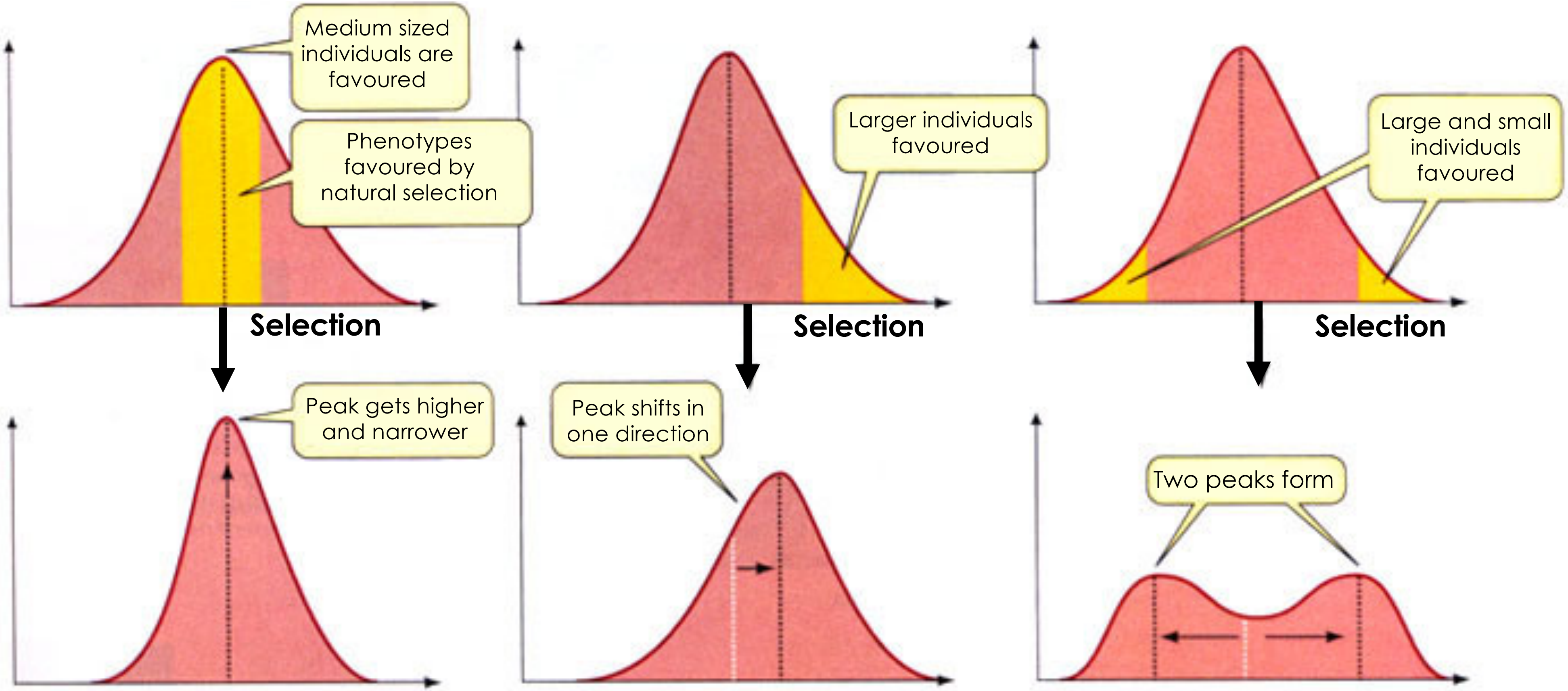


Within 20 years of arriving in the New World, populations of *D. subobscura* have increased dramatically, but have reduced genetic variation

Stabilising Selection

Directional Selection

Disruptive Selection



Reduces variation but does NOT change the mean

Changes the mean value towards one extreme

Favours the two extremes producing two peaks

Context Slide

Directed selection on common wild mustard

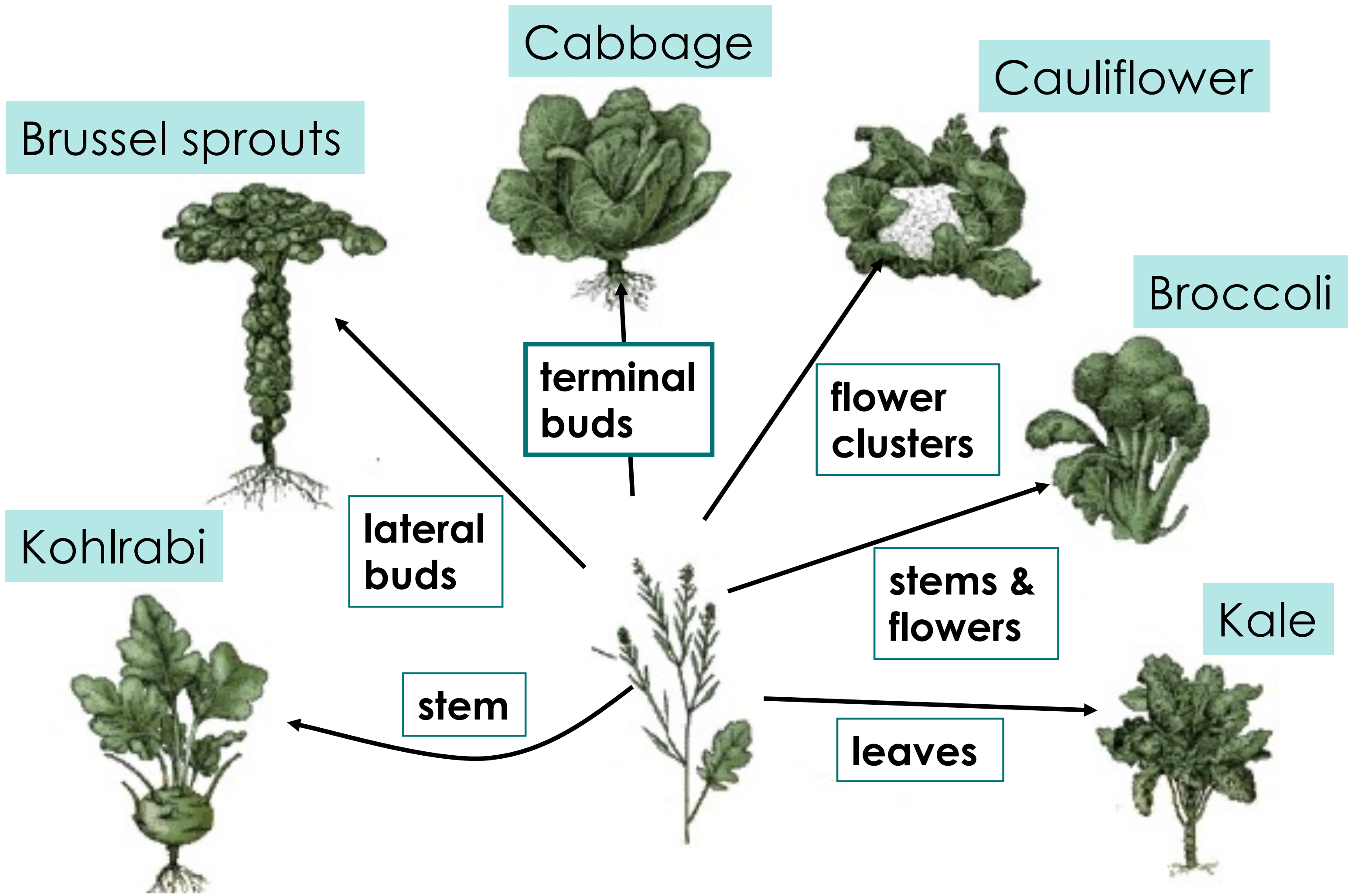
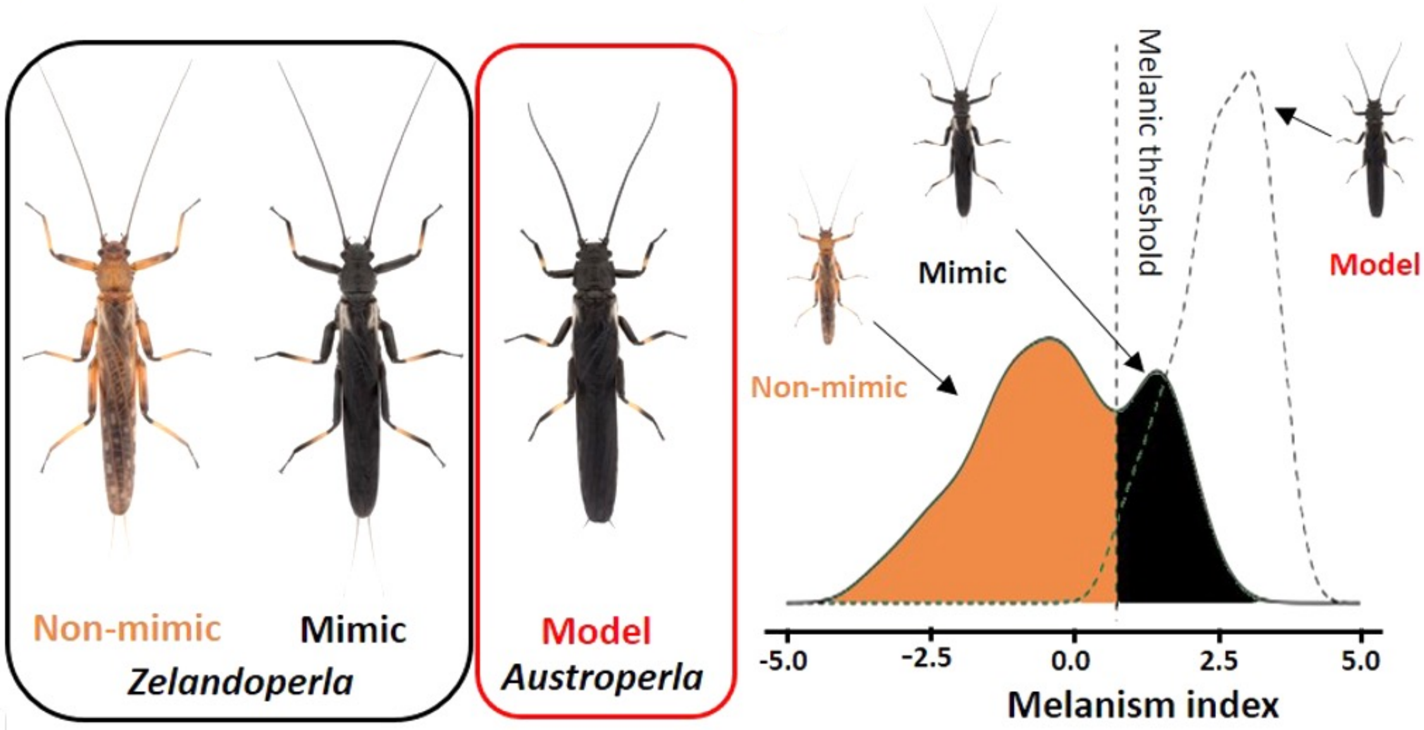


Image: Ni et al (2024) Science

Directional selection on insect colour

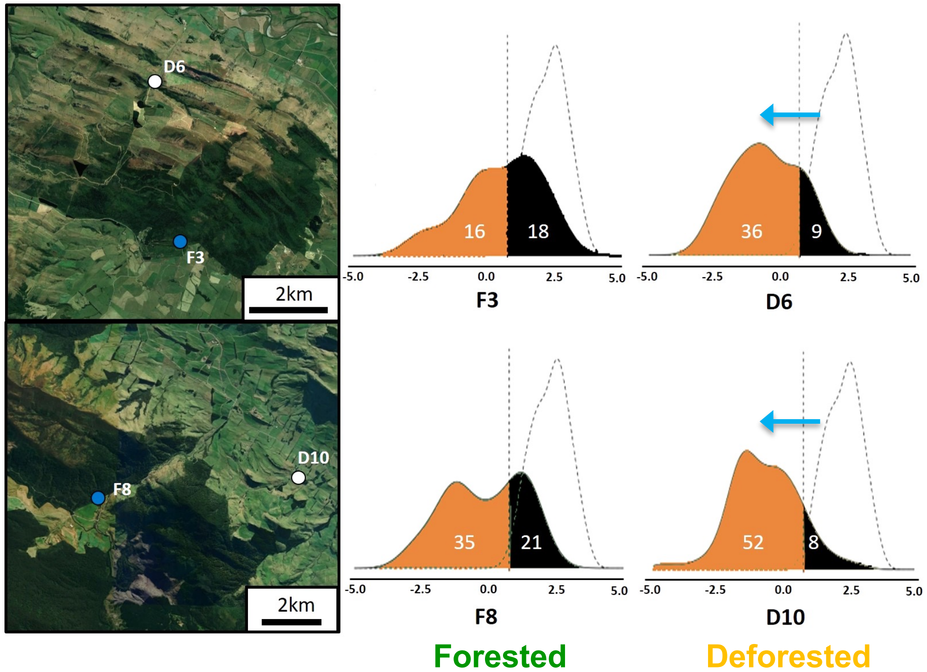


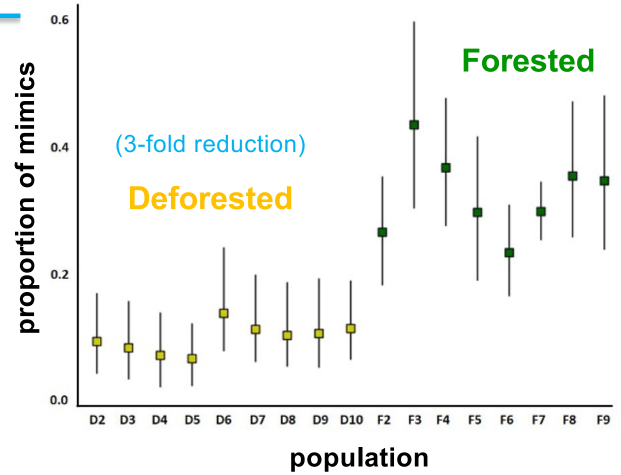
Context Slide

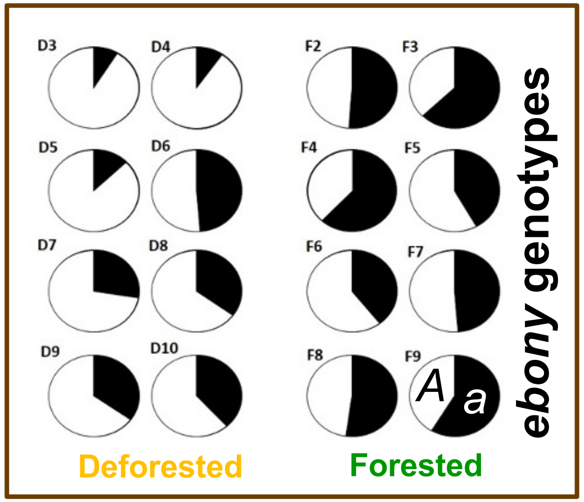
Image: Ni et al (2024) Science

Directional selection on insect colour

Phenotypic shifts:
fewer dark individuals in deforested habitats



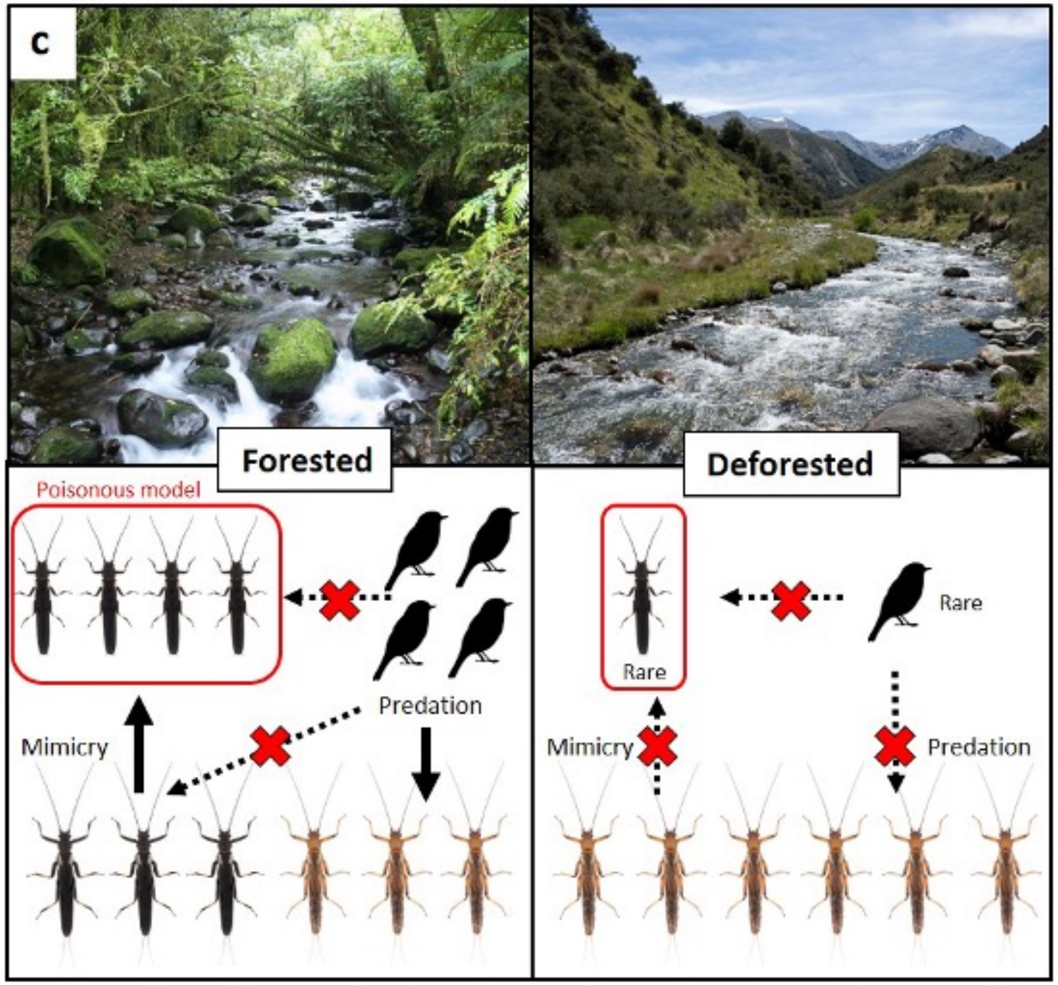




Context Slide

Image: Ni et al (2024) Science

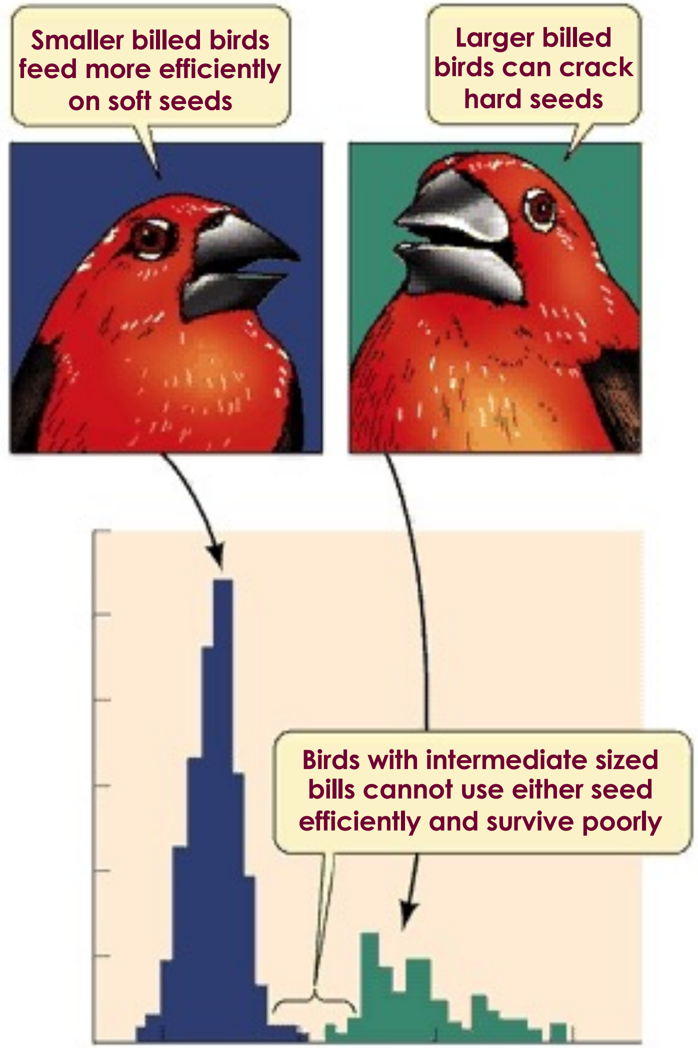
Directional selection on insect colour



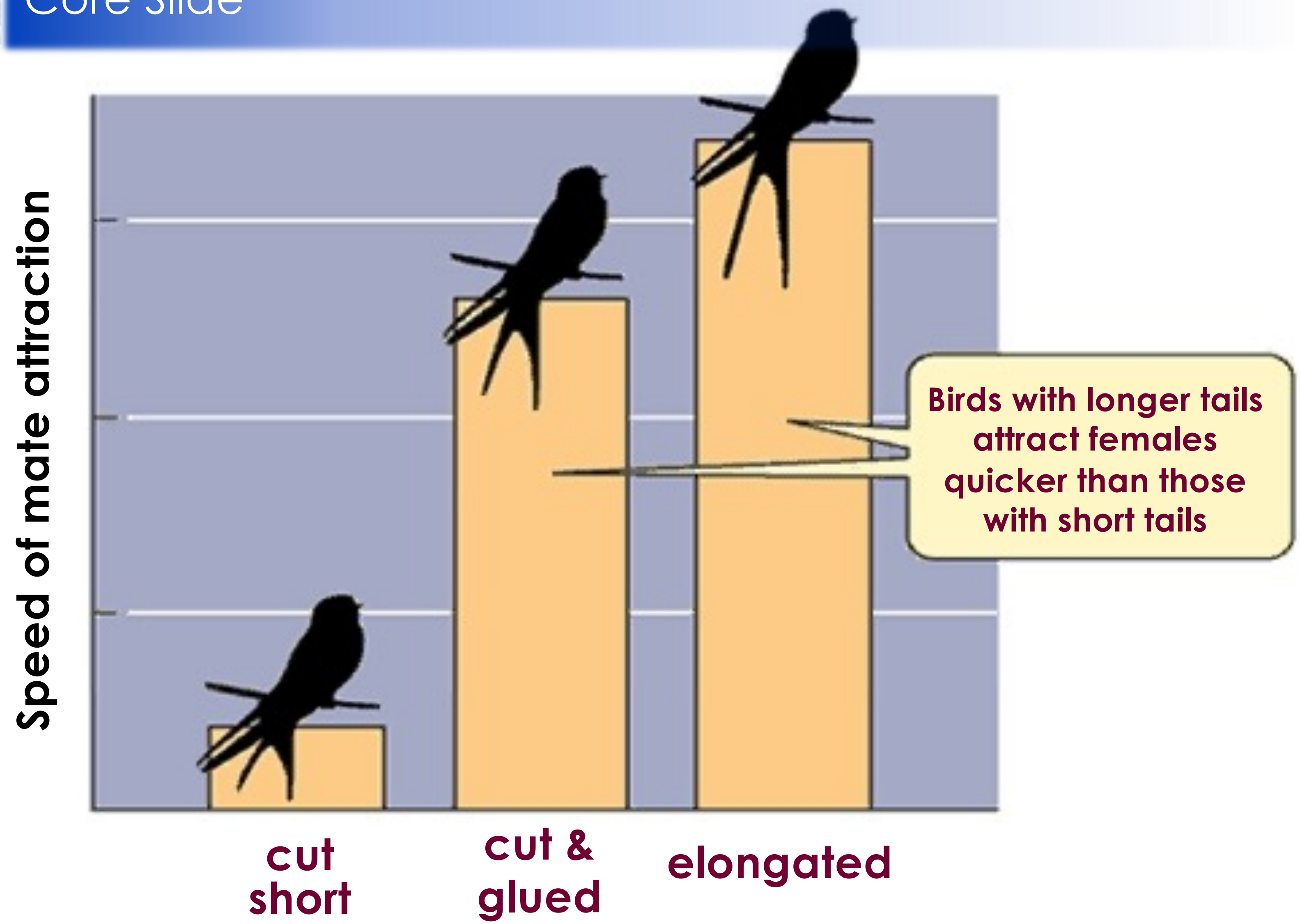
West African Black-bellied Seedcrackers display two bill morphologies

What type of selection is this?

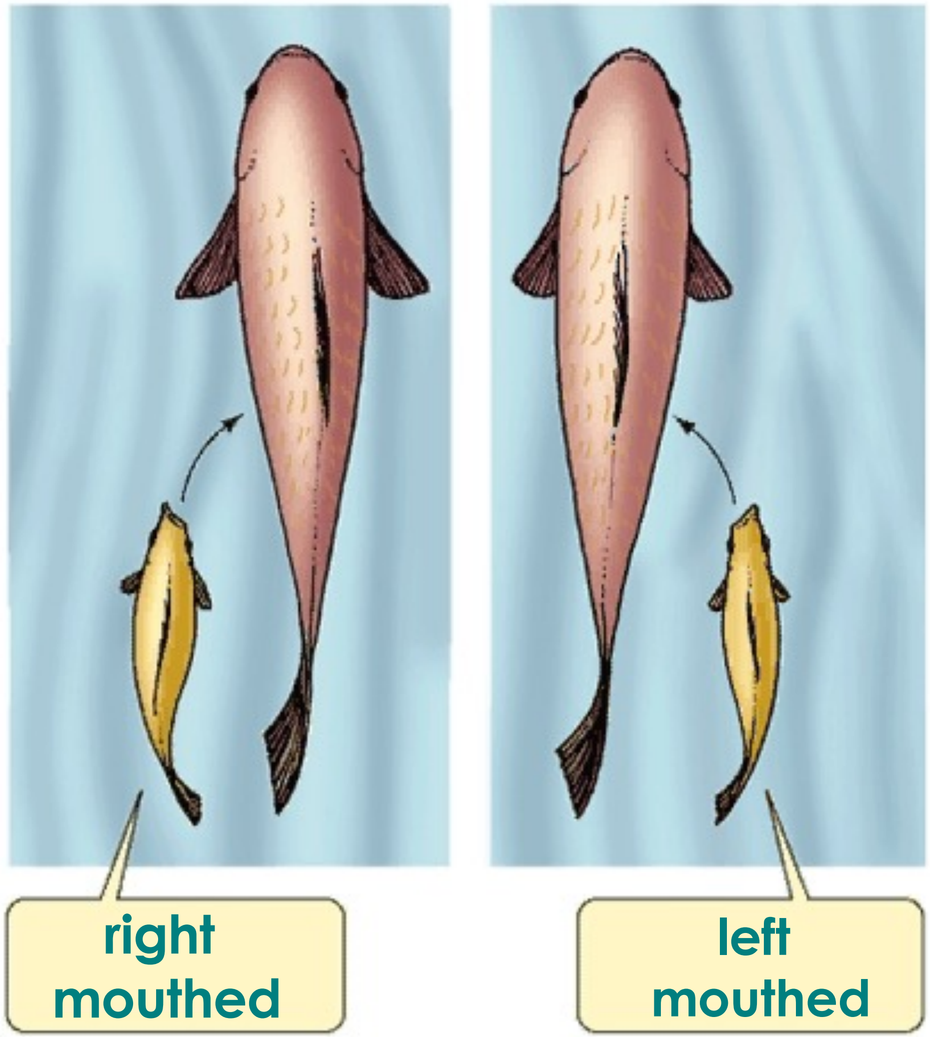
DISRUPTIVE SELECTION



Sexual Selection

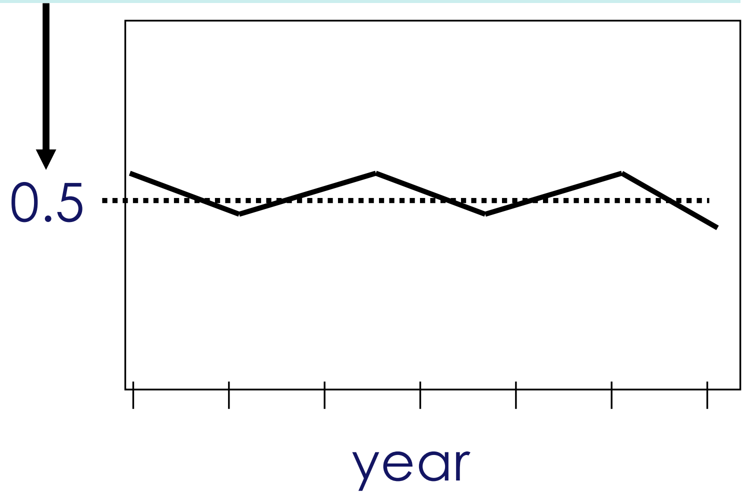


Frequency dependent selection in *Perissodus microlepis*



Natural selection maintains an equal proportion of left and right mouthed individuals in the population

Mean frequency of left mouthed *Perissodus*



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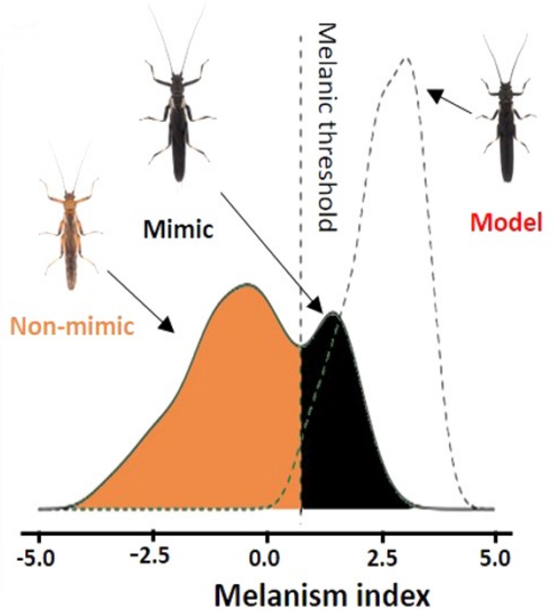
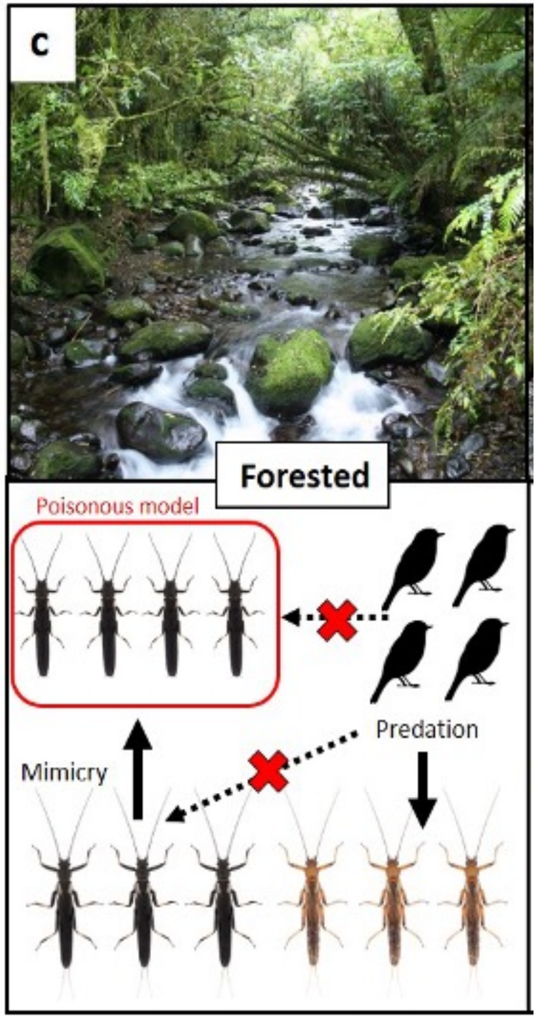




Frequency dependent selection?

Why are there any non-mimics in the population?

What happens if mimics start to outnumber the poisonous 'model'?

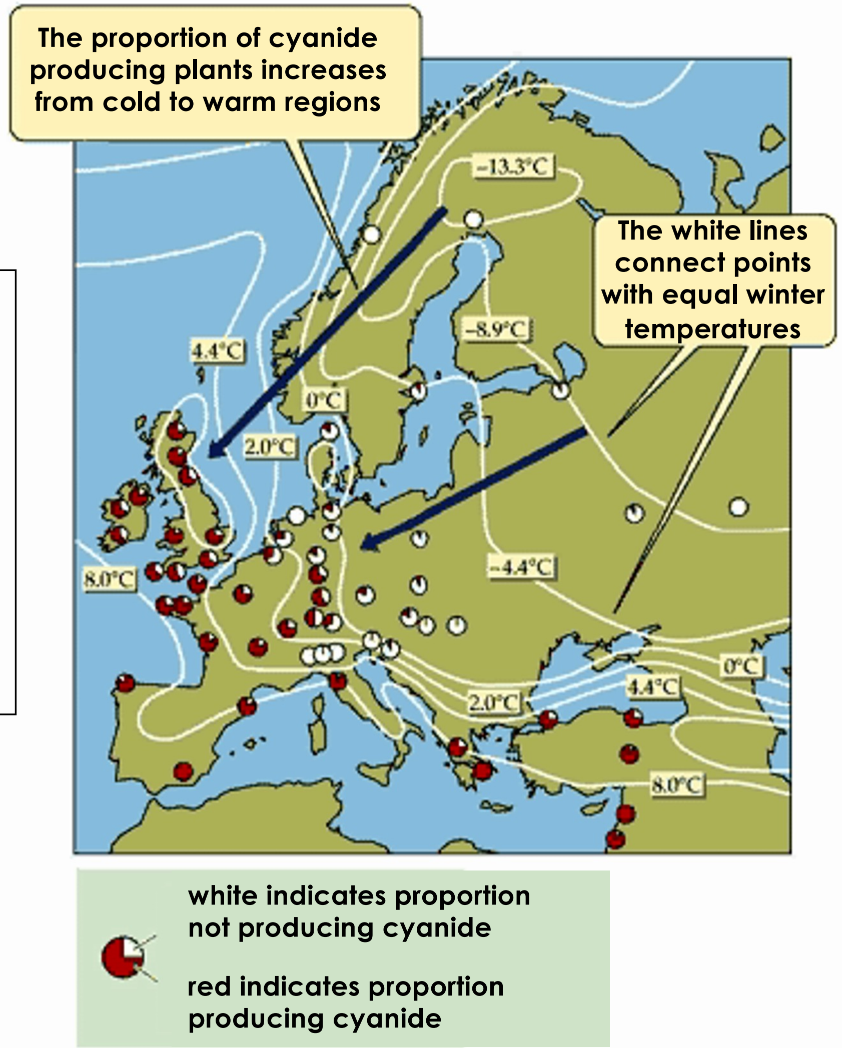
Spatial Distribution of Genetic Variation

Cyanide production in clover

Trifolium repens

The gradual geographic change in genetic/phenotypic composition is termed at:

CLINE



Mutation

Very slow to act and usually disadvantageous, its role is usually of macroevolutionary proportions

Migration

An individual from another population successfully mates (i.e. contributes gametes) to the gene pool.

- brings new alleles
- changes proportions of existing alleles
- changes population size
- makes two populations more similar

Lecture 21 Summary

- The Hardy-Weinberg equation can be used to predict genotype frequencies in a population
- Small populations rapidly lose allelic diversity as a result of genetic drift.
- Different types of natural selection lead to contrasting shifts in the phenotypic and genotypic composition of populations
- In cases of frequency-dependent selection, the fitness of a genotype is influenced by how rare or common it is.

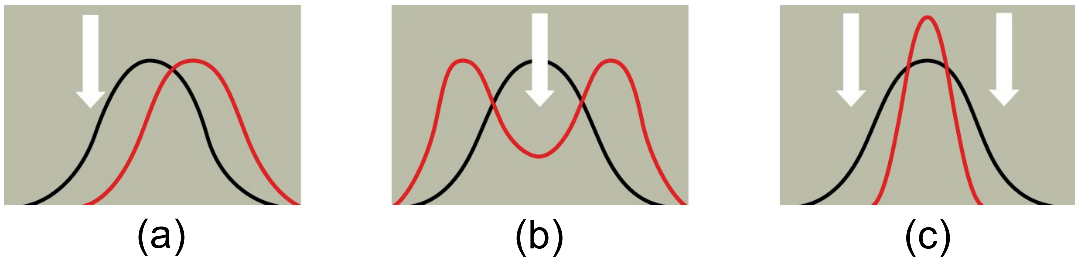
Objective-Based Questions

- Why would we want to estimate genotype frequencies in a population?
- State the Hardy-Weinberg equation for calculating genotypic frequency and define the terms p, q and 2pq.

If p=0.8 in a certain population, what

a) is the value of q?
b) % of the alleles will be the dominant allele?
c) is the frequency of individuals that are homozygous dominant?
d) is the frequency of individuals that are homozygous recessive?
e) is the frequency of heterozygous individuals?

- If 1 in 10000 children is born with Tay-Sachs disease, what percentage of the population are expected to be carriers for this autosomal recessive disease?
- Using an example, explain what is meant by the Founder Effect.
- Identify the modes of selection in figures (a)-(c) on the right. Note: the large white arrows symbolize selective pressures against certain phenotypes.



If you have any questions relating to this part of the course then......

Review the lecture resources on Blackboard, watch the lecture recordings, use the Learning Modules, read your textbook, ask during the lab, come along to a drop-in session, contact me, ask ask ask........

As teachers we appreciate the difficulty of learning this stuff so please do not be afraid to ask for help. However, you will only get better by practising - so go through questions and understand your answers right or wrong. Get a feeling for the concepts - what are the main points? Once you understand this, the rest becomes easier.........**honest!**

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