

# Genetics in support of fisheries and aquaculture management

17-19 September  
Faro, Portugal

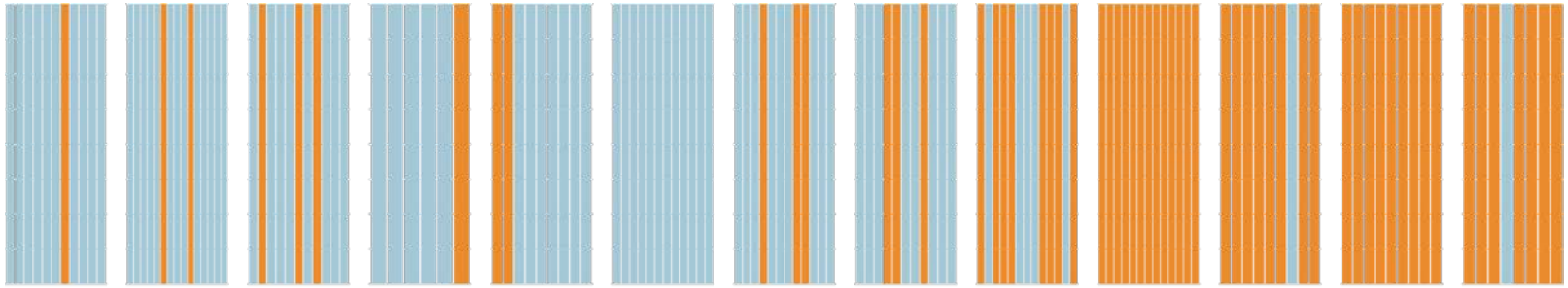


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# Background principles of population genetics

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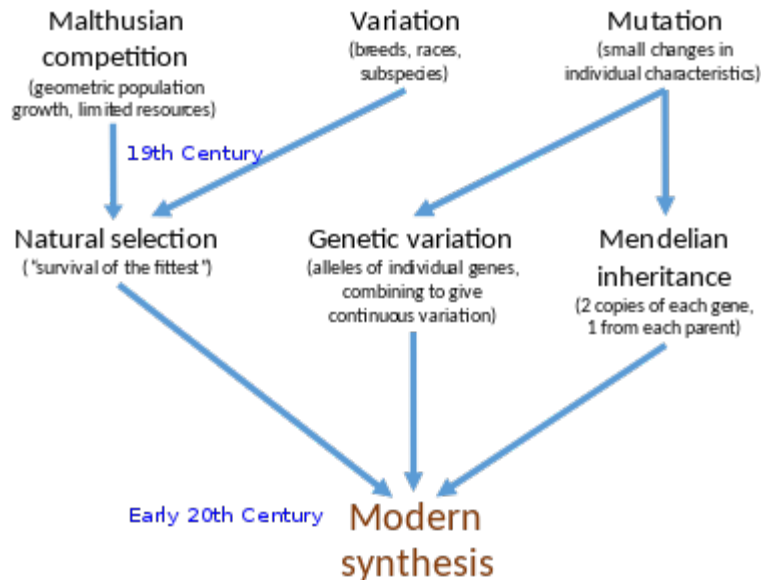


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# The importance of population genetics

- "Nothing in Biology Makes Sense Except in the Light of Evolution"
- Evolution is: "a change in the frequency of an allele within a gene pool"



Theodosius Grygorovych Dobzhansky

**"Nothing in Evolution makes sense except in the light of population genetics" (Michael Lynch)**



# Why is population genetics important?

- What are the risks of getting a child with a genetic disease?
- Has farming or breeding caused a loss in genetic variability in farmed plants and animals?
- Will wild rare animals or plants be able to avoid inbreeding and survive?
- How are populations from different parts of the world related to each other?
- Have genomes adapted to different environmental conditions across the globe?
- How do populations and species evolve over time?

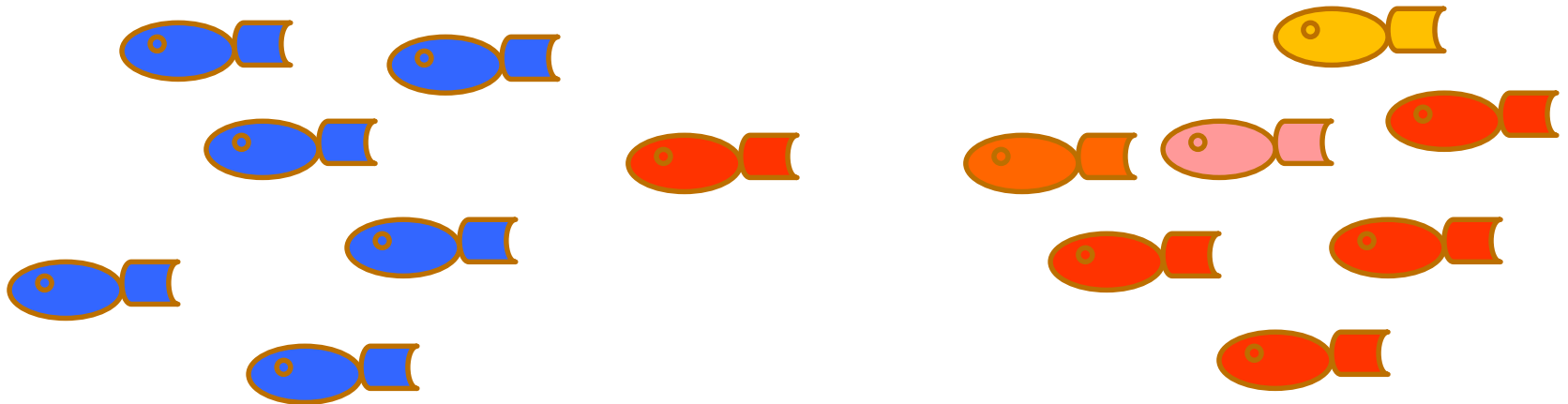
# What is a (genetic) population?

- The individuals of a species living within an country?
- The individuals of a certain species living within a restricted geographical region, e.g. the Scandinavian peninsula?
- Group of individuals within a species who are reproductively isolated (to some degree) from other individuals of the same species?

# Concepts and definitions

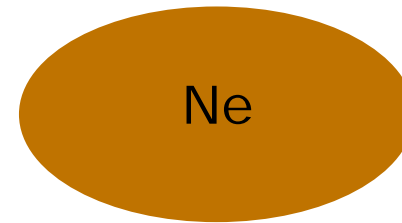
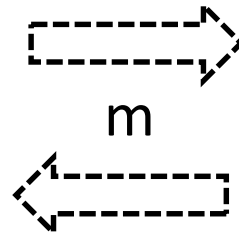
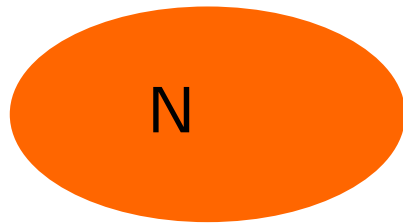
## Populations

A group of individuals of the same species that are **reproductively isolated** (at least to some extent) from other conspecifics



# Concepts and definitions

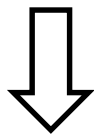
## Populations



### Ecological population

Demographic cohesiveness

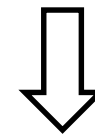
$$m < 0.1$$



### Evolutionary population

Genetic cohesiveness

$$N_e m < 25$$

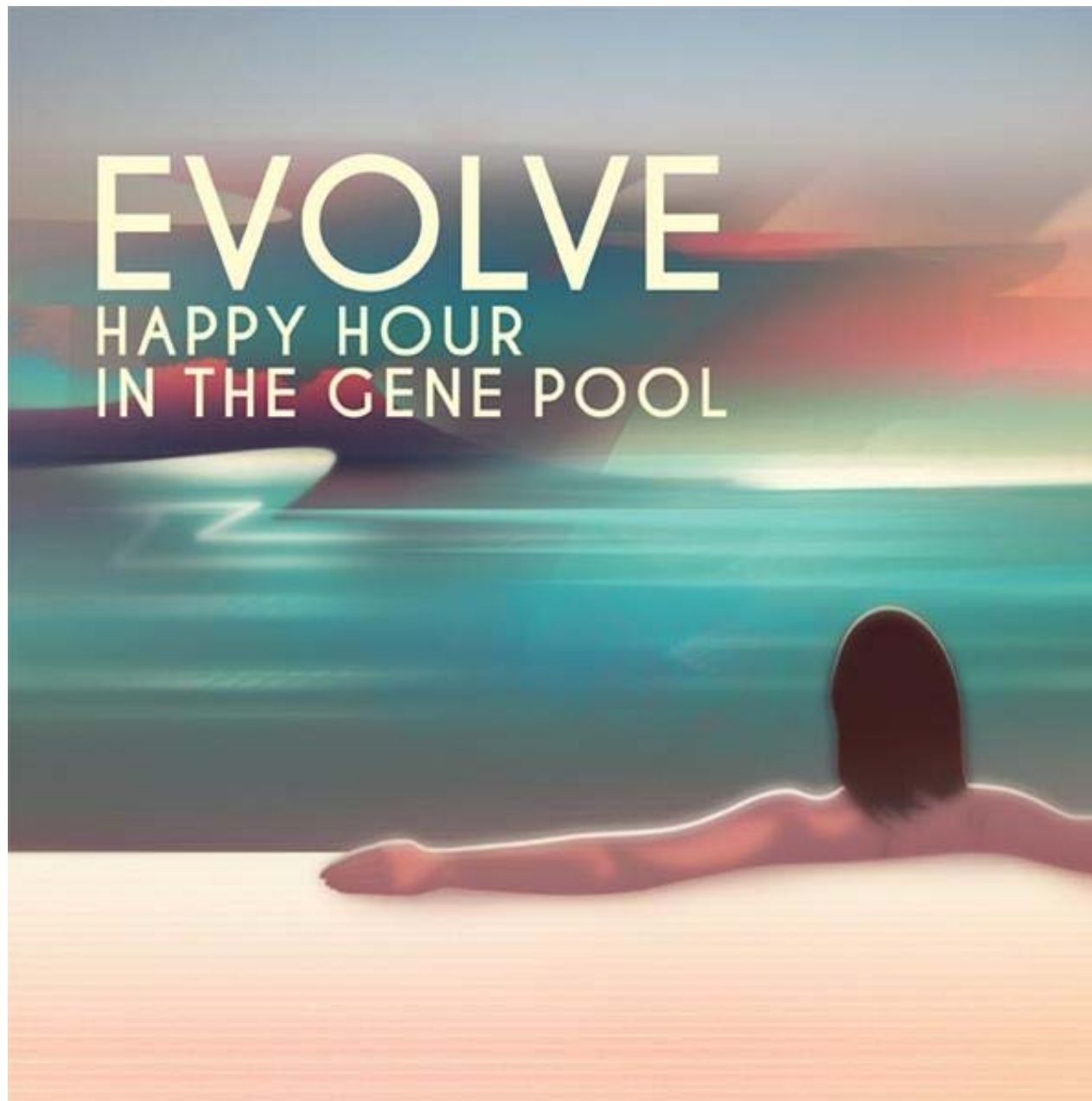


## Population differentiation

**Demographic independence**

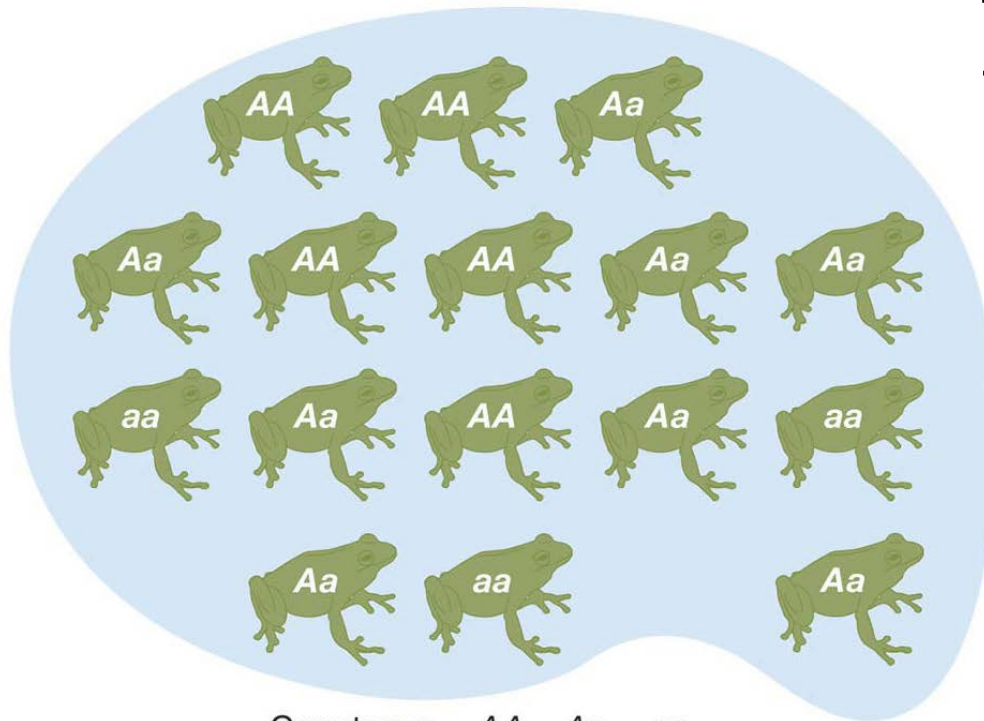
**Genetic independence**





# The Gene-Pool Concept

**The gene pool is the sum total of alleles in a population**

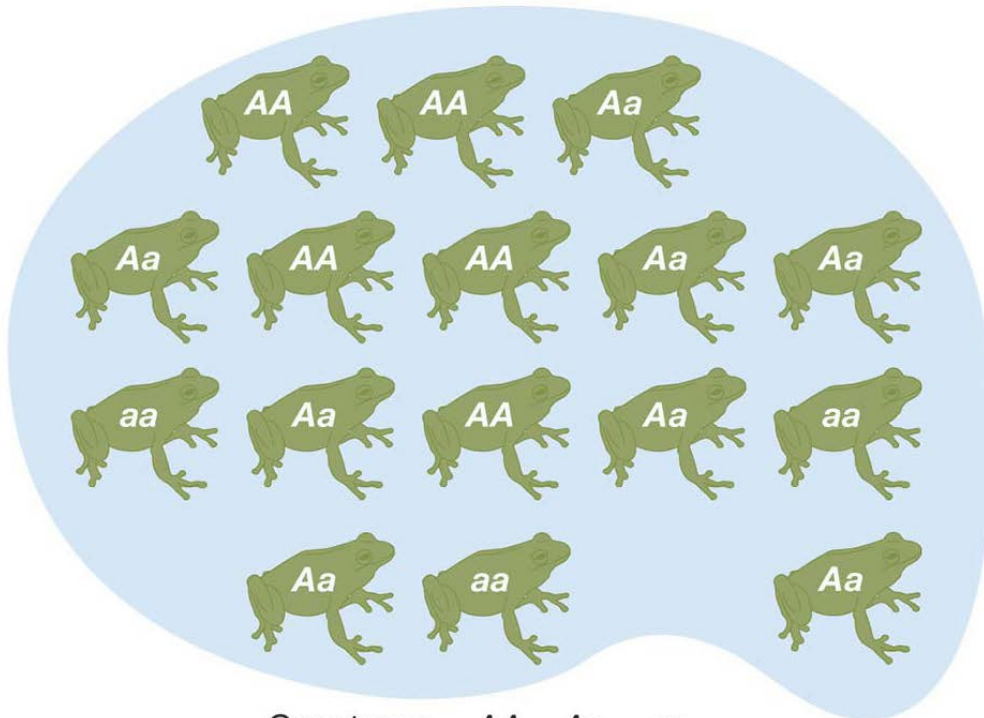


Genotypes	AA	Aa	aa
Number	5	8	3
Alleles	A	a	
Number	18	14	

The gene pool is a fundamental concept for the study of genetic variation in populations: it is the sum total of all alleles in the breeding members of a population at a given time. We can describe the variation in a population in terms of genotype and allele frequencies

# The Hardy Weinberg law

$p=?$        $q=?$



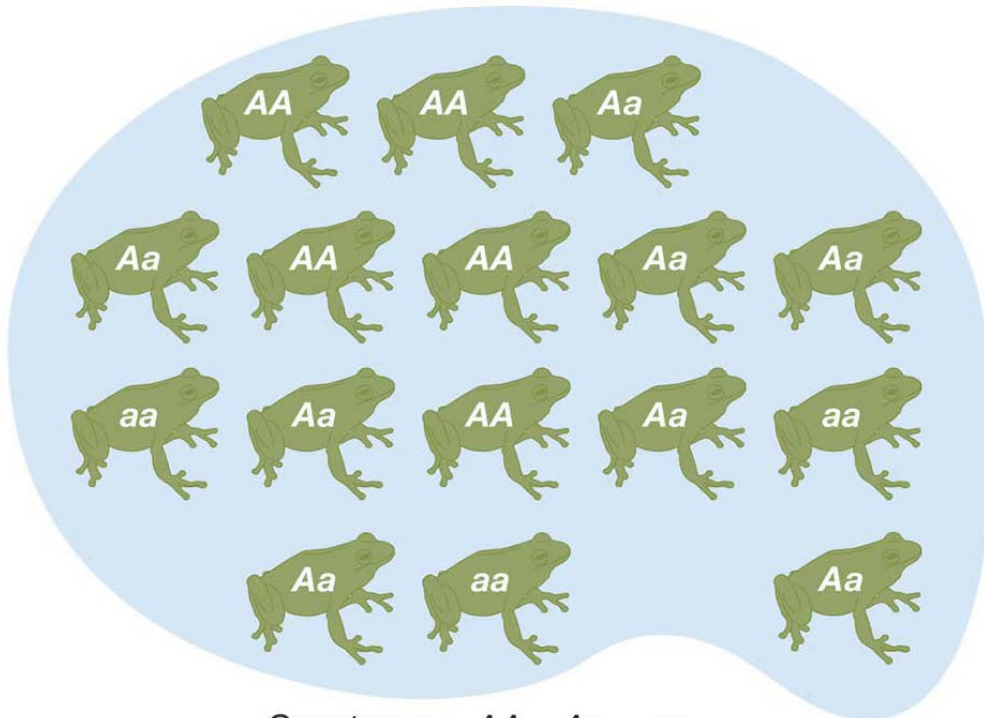
Genotypes	AA	Aa	aa
Number	5	8	3
Alleles	A	a	
Number	18	14	

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# The Hardy Weinberg law

$$p = 0,56$$

$$q = 0.44$$



What is the probability of getting an AA homozygote in the next generation?

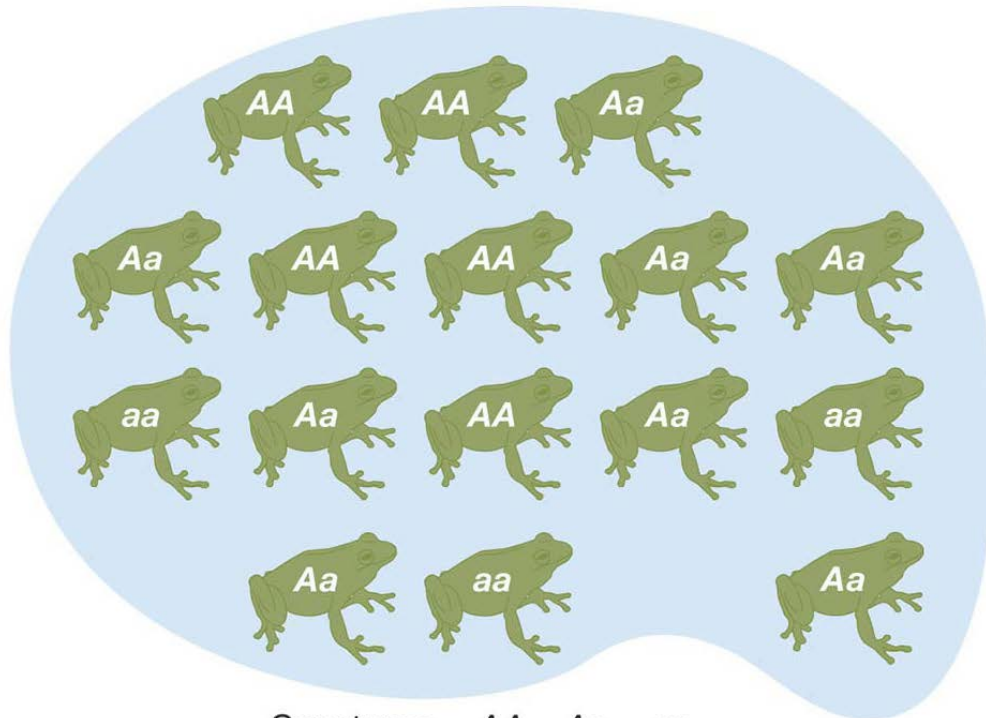
Genotypes	AA	Aa	aa
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# The Hardy Weinberg law

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Genotypes	AA	Aa	aa
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What is the probability of getting an AA homozygote in the next generation?

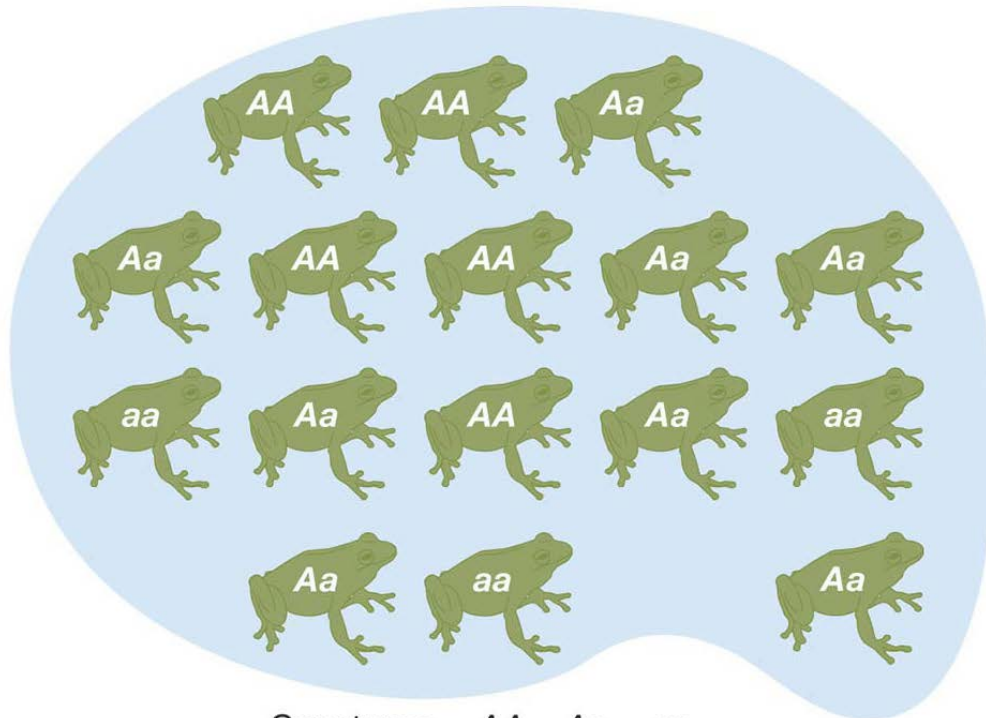
$$p \times p = p^2 = 0.3136$$

aa homozygote:  
 $q \times q = q^2 = 0.1936$

# The Hardy Weinberg law

$$p = 0,56$$

$$q = 0.44$$



Genotypes	AA	Aa	aa
Number	5	8	3
Alleles	A	a	
Number	18	14	

What is the probability of getting an Aa heterozygote in the next generation?

$$(p \times q) \times 2 = 2pq = 0.4928$$

# Factors affecting genetic variation in populations

- Four evolutionary forces:
- Mutation
- Migration
- Random genetic drift
- Selection

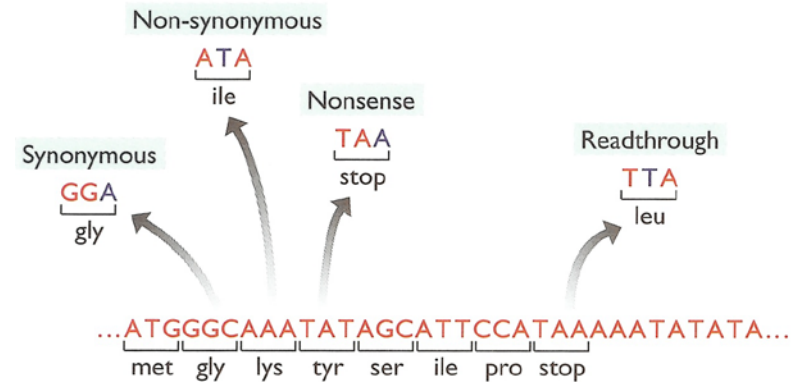


*"Why Dug! I haven't seen you in years! You haven't evolved a bit!"*

# Evolutionary forces

## Mutation

Random changes in DNA code



**Figure 14.11** Effects of point mutations on the coding region of a gene.

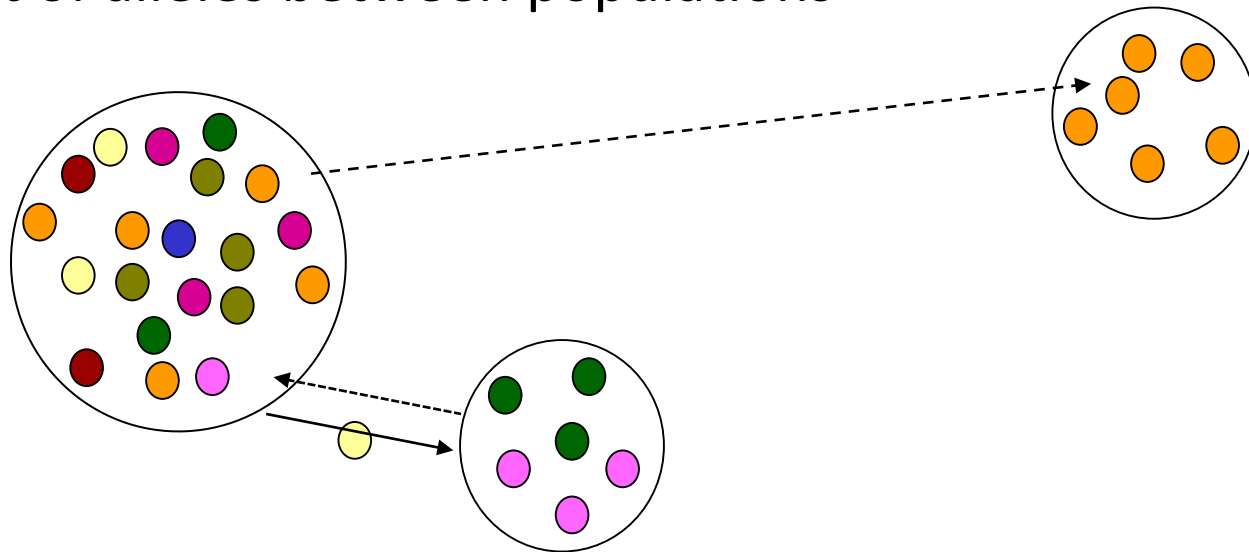
- Most mutations are harmful -> removed through natural selection
- A few mutations are beneficial -> favoured and increase in frequency in the population
- Mutation rates are generally very low, but varies among species and genomic regions



# Evolutionary forces

## Gene flow

Movement of alleles between populations



Is gene flow = migration??

No!

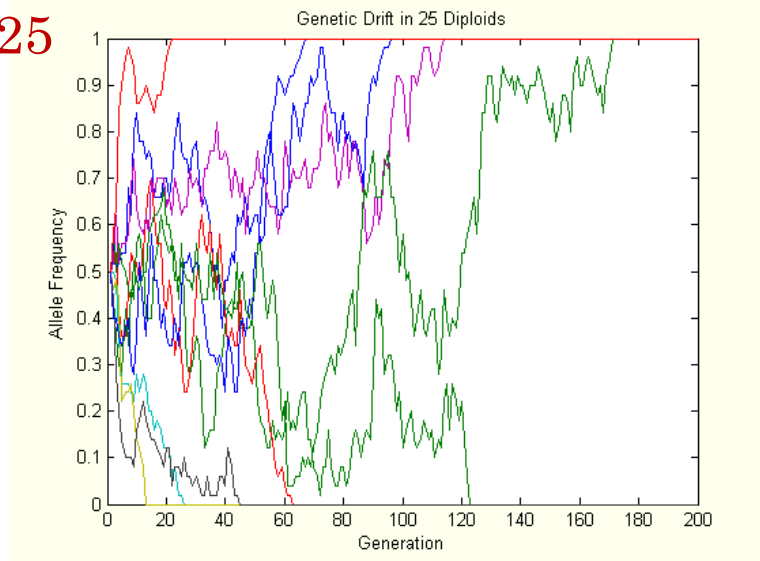
Gene flow requires successful reproduction in new population

# Evolutionary forces

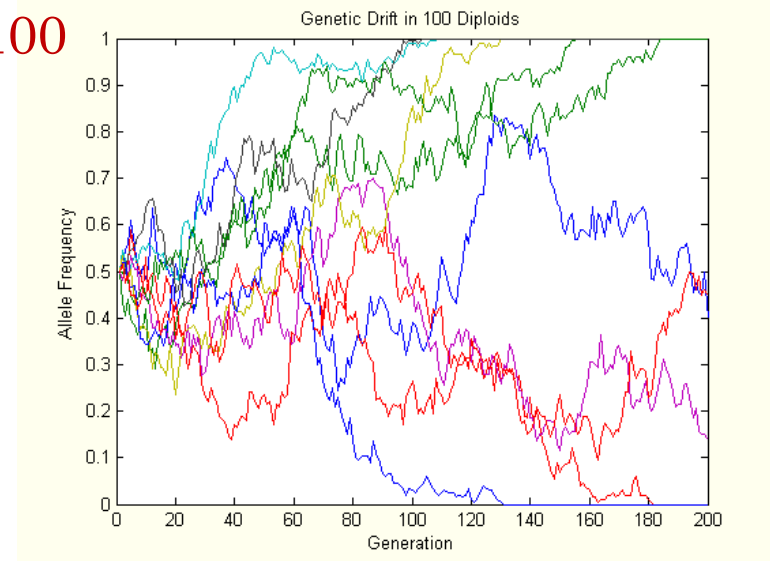
## Genetic drift



$N_e=25$



$N_e=100$



# Effective population size

The **effective population size ( $N_e$ )** is the size of an ideal population, which loses genetic variation at the same rate as the actual population

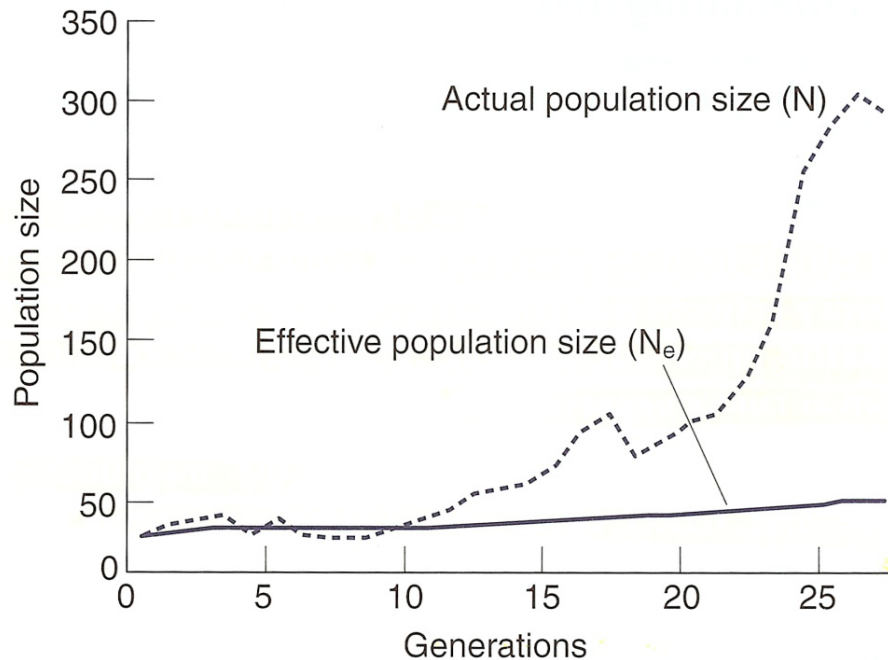
= "how does the population behave genetically?"

*So, what is an ideal population?*

- 1) Random mating
- 2) 1:1 sex ratio
- 3) Non-overlapping generations
- 4) Random number of offspring (Poisson distribution)

# Effective population size

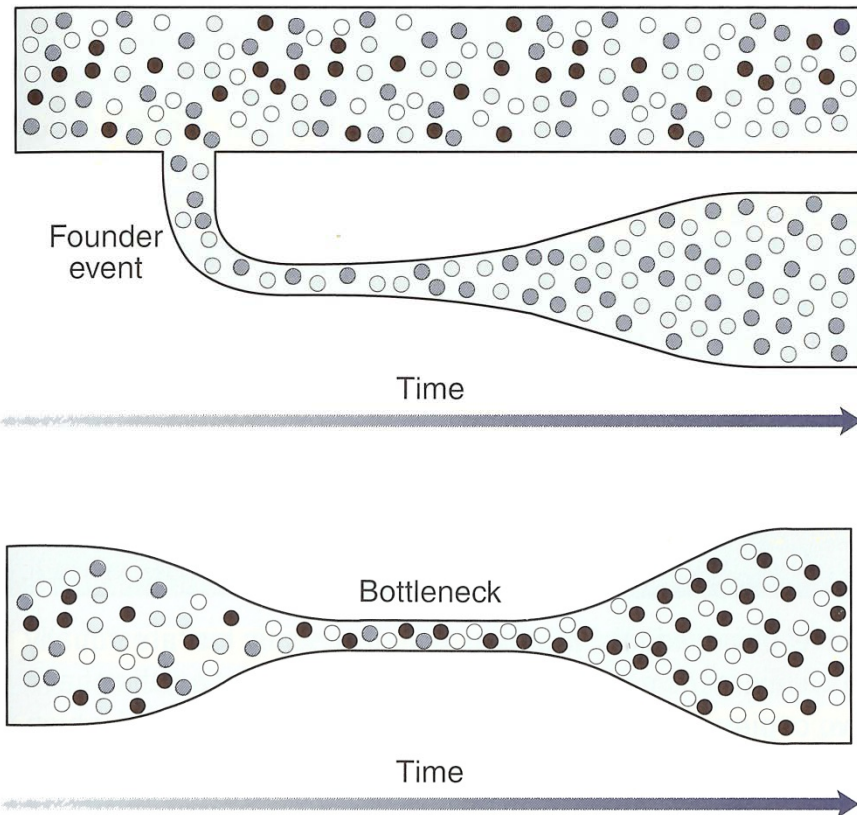
## Effective population size ( $N_e$ ) and census size ( $N$ )



- $N_e$  is often smaller than  $N$
- $N_e$  is disproportionately affected by the smaller population sizes
- In a recently expanded population,  $N_e$  (and the amount of genetic variation) is still largely determined by the smaller ancestral population size.

# Effective population size

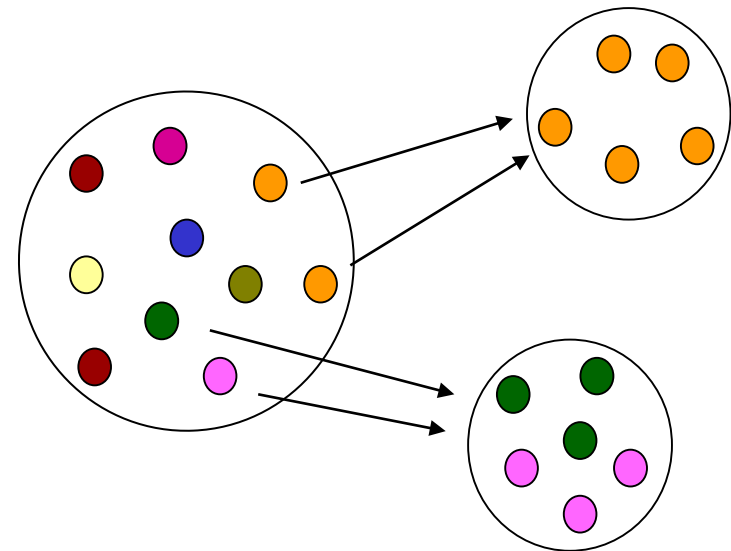
## Founder events and bottlenecks



### Dramatic reduction in $N_e$

-> Increased genetic drift

-> Loss of genetic variation



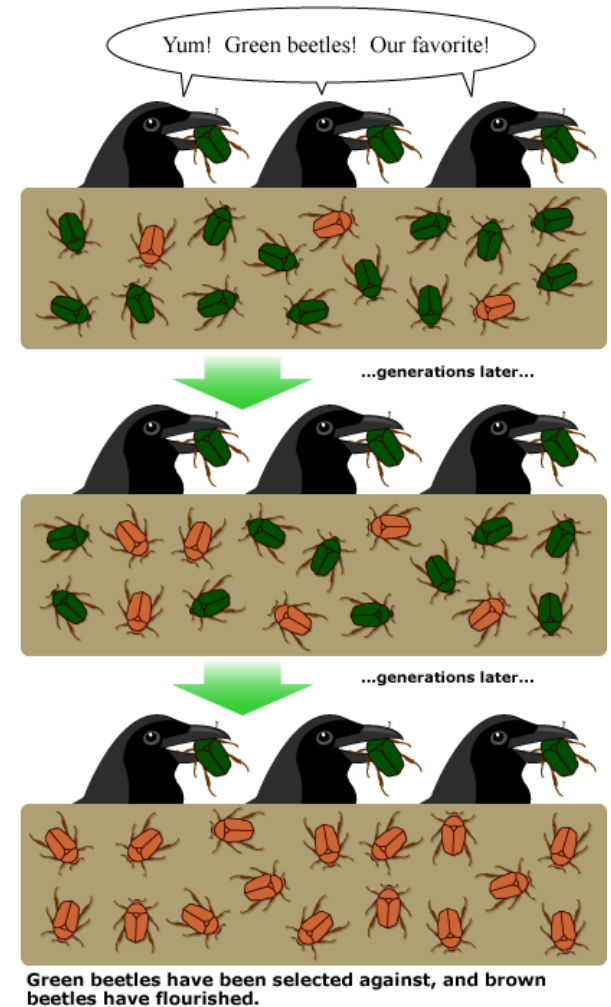
# Evolutionary forces

## Selection (natural and artificial)

Selection requires

- 1) Genetic variation between individuals
- 2) Inheritance from parents to offspring
- 3) Fitness differences between genetic variants

Natural selection, in a nutshell:



[http://evolution.berkeley.edu/evolibrary/article/bergstrom\\_02](http://evolution.berkeley.edu/evolibrary/article/bergstrom_02)

# Evolutionary forces

## Selection (natural and artificial)



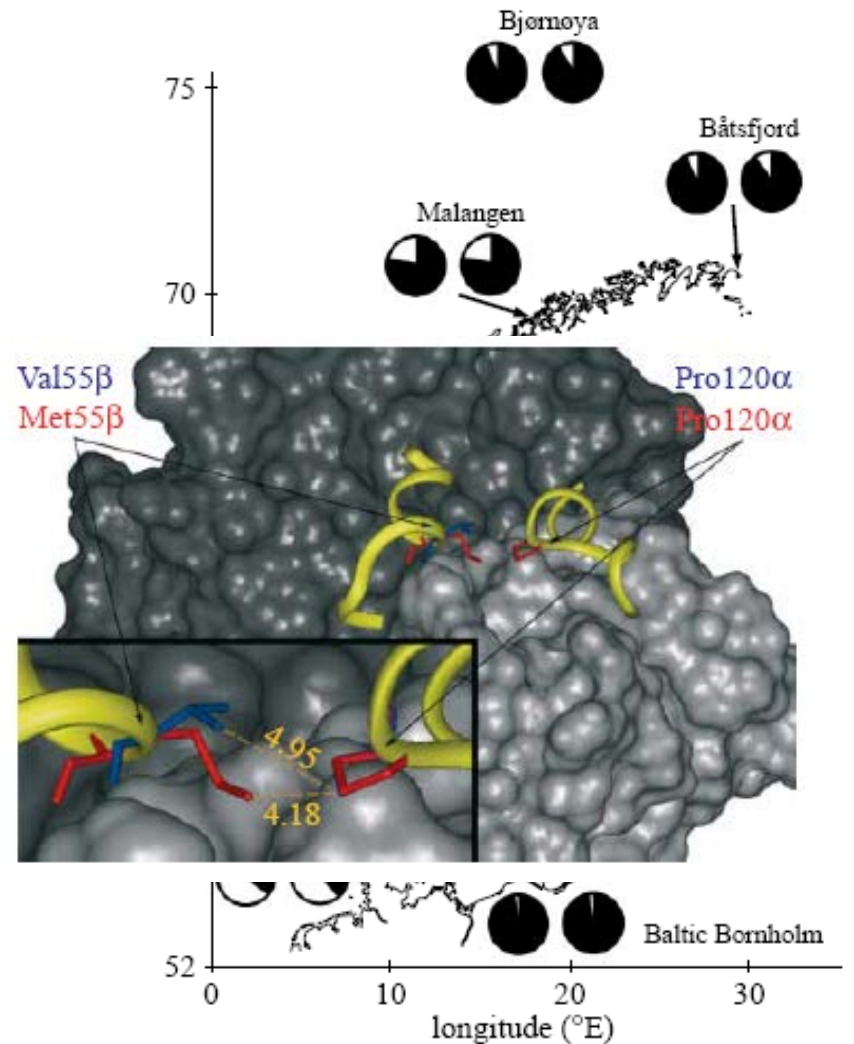
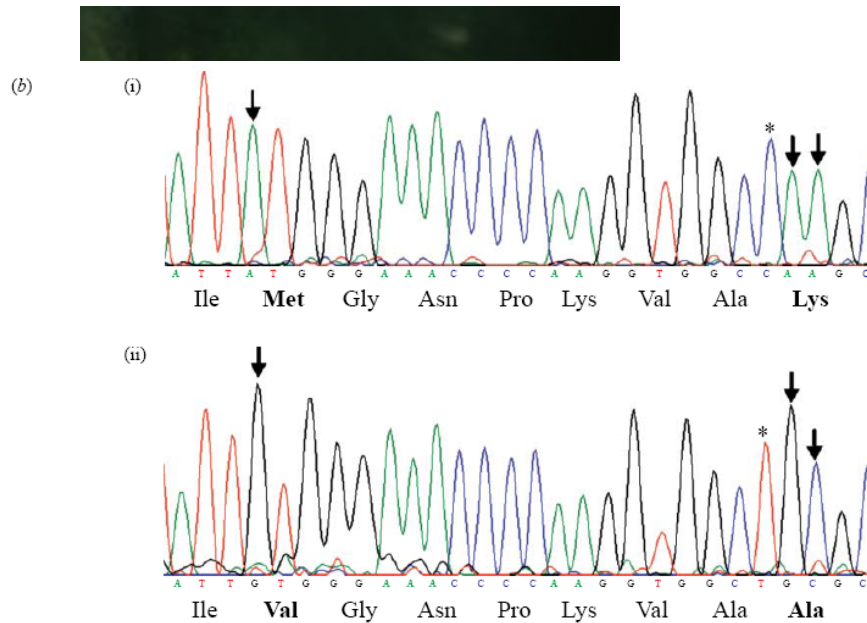
## Natural Selection

It Still Works

# Evolutionary forces

## Selection (natural and artificial)

Haemoglobin variation in Atlantic cod



1 2 3

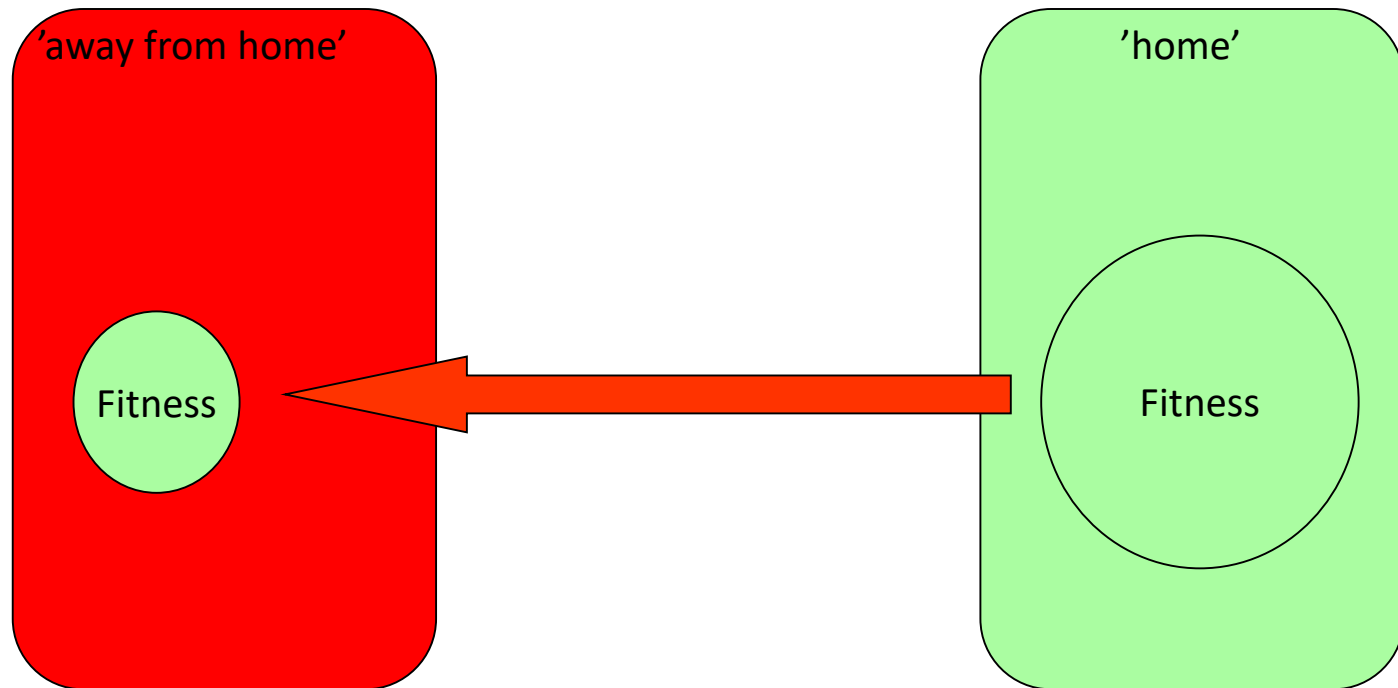


# Evolutionary forces

## Selection (natural and artificial)

### Local adaptation - 'Home' versus 'away'

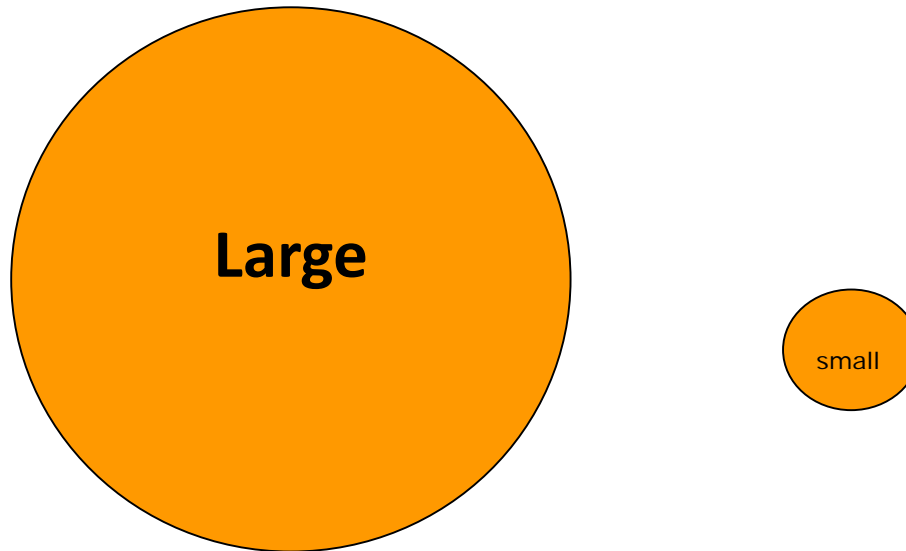
- Individual has higher fitness 'at home' compared to in a different environment



# Evolutionary forces

## Balance between genetic drift and selection

Where is genetic drift and selection most important?



- **Selection** operates most efficiently in large populations
- In small populations, even disadvantageous alleles may become fixed due to **genetic drift**

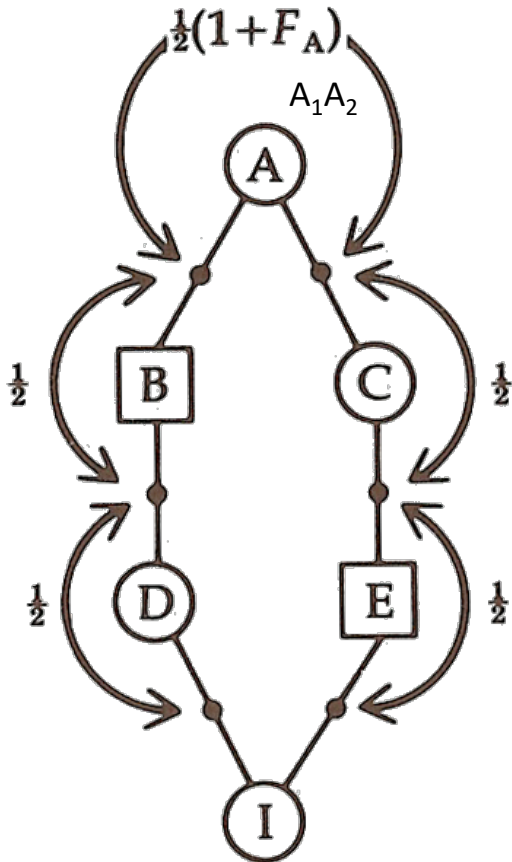
# Inbreeding

- **Definition:** Reproduction between (genetic) relatives
- **Result:** Increased homozygosity, reduced heterozygosity
  - difference between expected ( $H_e$ ) and observed ( $H_o$ ) heterozygosity
- **Effect:** May lead to **inbreeding depression** through exposure of deleterious recessive alleles to selection

## Relevance to wild populations?

- > Poor growth and survival, deformities, reduced fertility
- > Largest effect in small populations

# Inbreeding



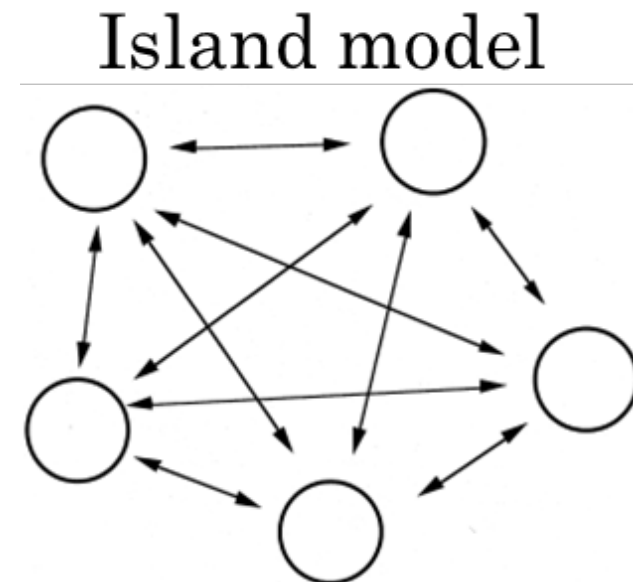
Departures from Hardy-Weinberg Equilibrium proportions

Genotype	Random mating	Inbreeding (=non-random mating)
AA	$p^2$	$p^2 + Fpq$
Aa	$2pq$	$2pq - F2pq$
aa	$q^2$	$q^2 + Fpq$

**Inbreeding coefficient**  $F_{is} = (H_s - H_i) / H_s$  - the mean reduction in H of a locus within an individual due to non-random mating within a subpopulation

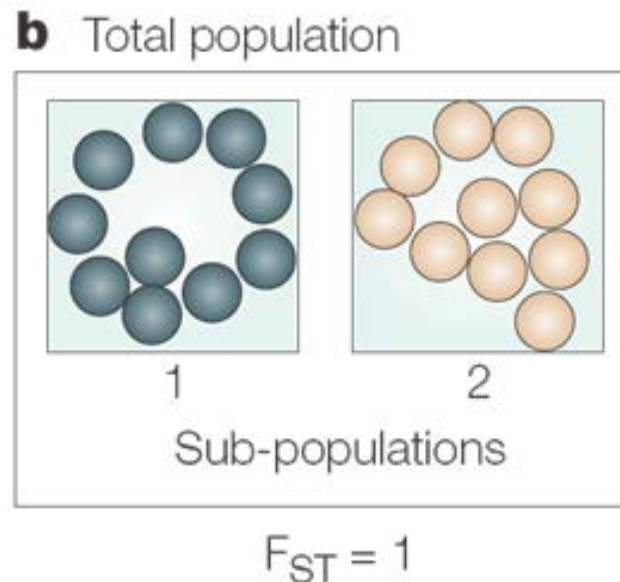
## Wright's F statistics

- Inbreeding/co-ancestry coefficients
- Measures how the genetic variation is distributed between
  - Individuals (I)
  - Subpopulations (S)
  - Total population (T)



## Wright's F statistics

- Inbreeding/co-ancestry coefficients
- Measures the reduction in heterozygosity (H) due to random genetic drift



# Genetic variation and population structure

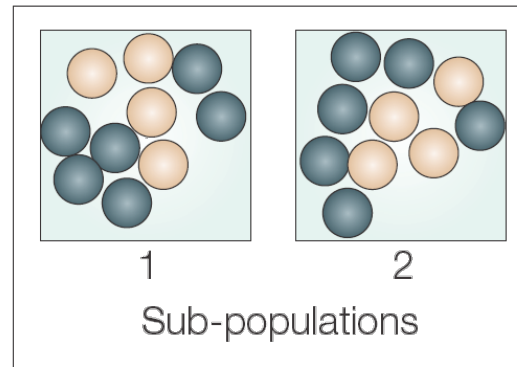
## Wright's F statistics

Proportion of total genetic variance caused by differences among populations

$$F_{ST} = \frac{\sigma_{GB}^2}{\sigma_{GB}^2 + 2\sigma_{GW}^2}$$

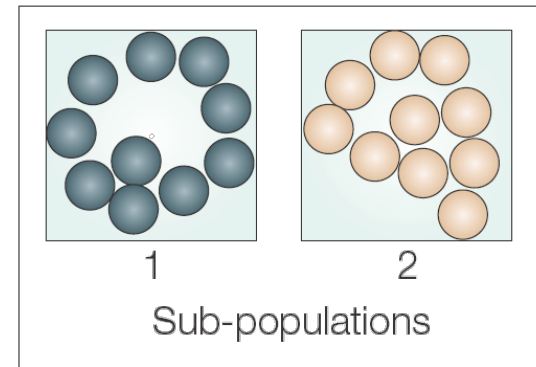
$$F_{ST} = \frac{H_T - H_S}{H_T}$$

**a** Total population



$F_{ST} = 0$

**b** Total population



$F_{ST} = 1$

**$F_{ST} = 1$** : Populations fixed for different alleles

**$F_{ST} = 0$** : Allele frequencies are the same across populations

# Genetic variation and population structure

$$F_{ST} = \frac{H_T - H_S}{H_T}$$

← Sub-population  
← Total population

Two alleles, two populations:

	A <sub>1</sub>	A <sub>2</sub>	
Freq. in pop 1	0.3	0.7	H <sub>T</sub> = 2*0.5*0.5 = 0.5
Freq. in pop 2	0.7	0.3	H <sub>S</sub> = ((2*0.3*0.7)+(2*0.7*0.3))/2
Overall freq	0.5	0.5	= 0.42

$$F_{ST} = (0.5 - 0.42) / 0.5 = \underline{0.16}$$



WHEN? WHY?  
WHERE? WHO?  
WHAT? HOW?  
HOW? WHEN?  
WHERE? WHO?  
HOW? HOW?  
WHERE? WHICH? WHOSE? WHEN? WHY?  
WHICH?  
WHOSE?

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HOW?  
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