PART 1

What is a Population?

The concept of a **POPULATION** is central to the fields of ecology, evolution and conservation

In general, populations are considered independent units, and are managed as such

HOWEVER

There are many definitions of what a population actually is, depending on the field, organism and (personal) preference
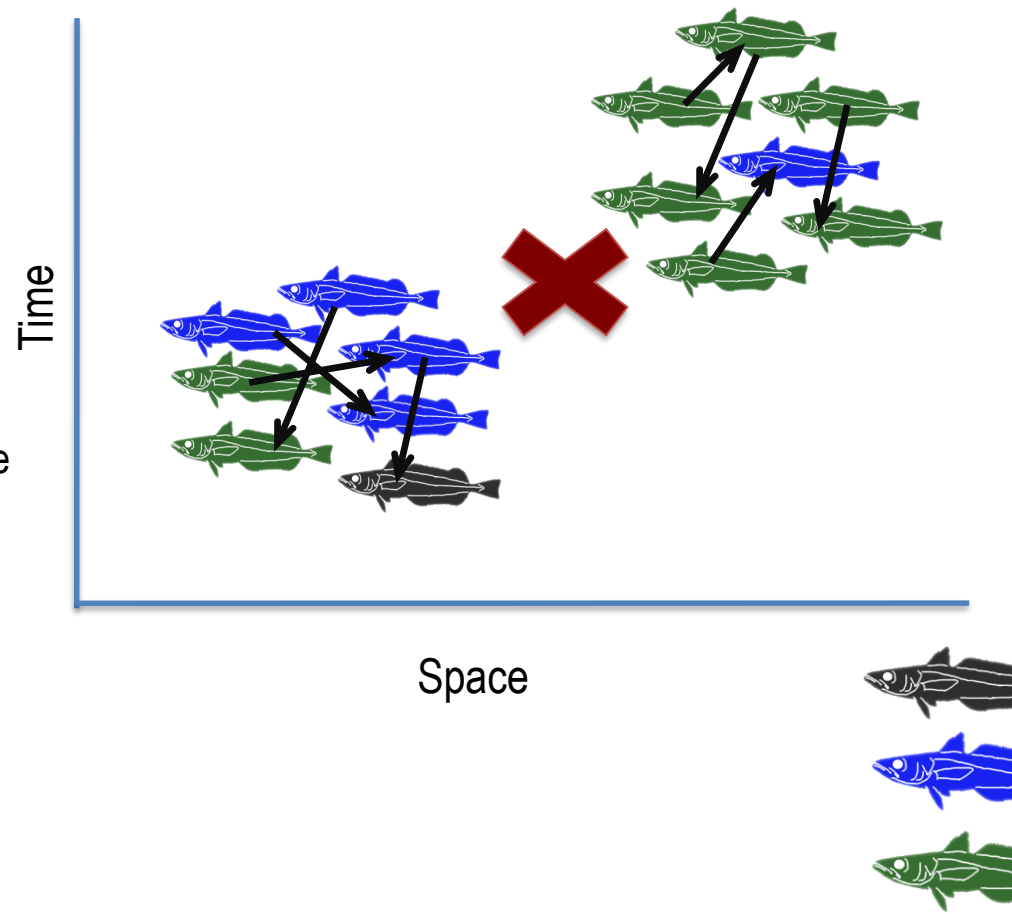
1.   ECOLOGICAL POPULATIONS

There are several definitions of what a population is, but all revolve around the notion of an isolated group of individuals that lives in the same area

E.g.

1.1. A group of individuals of the same species occupying a particular space at a particular time

1.2. A group of individuals of the same species that live together in an area of sufficient size where all requirements for reproduction, survival and migration can be met

1.3. A group of individuals occupying the same biogeographical area or biome
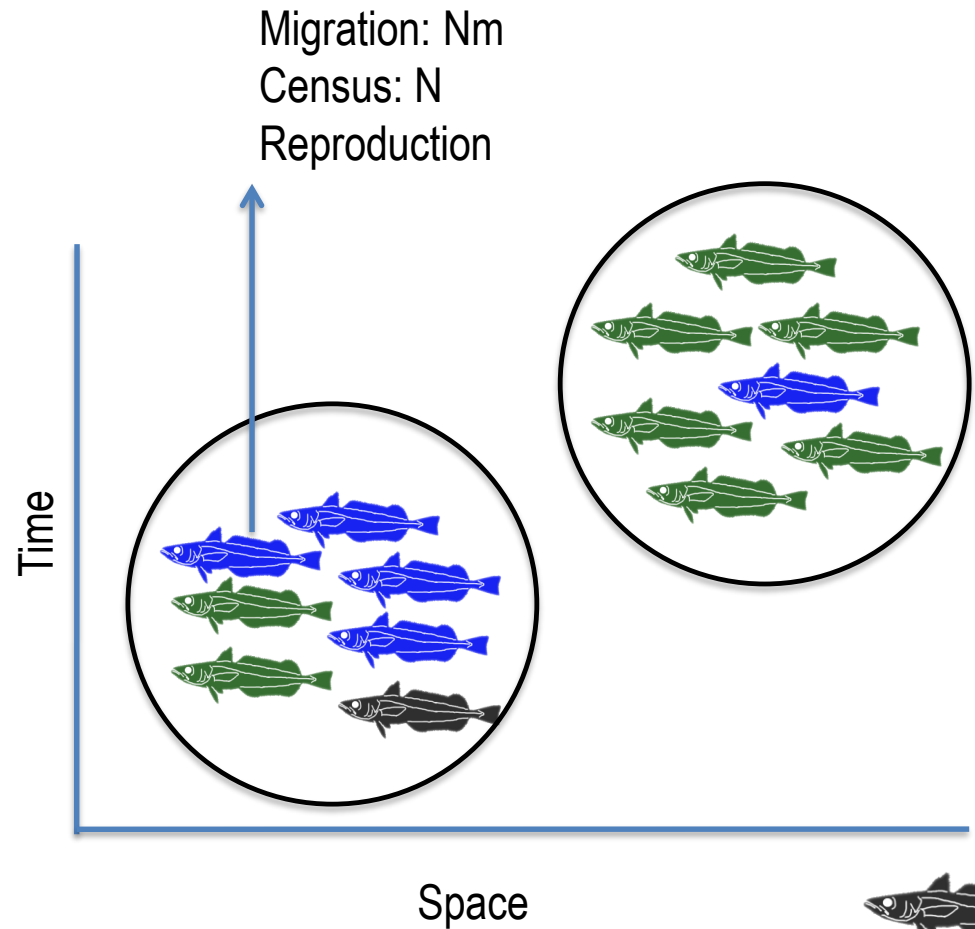
Time

Space

2.   STATISTICAL POPULATIONS

A group that we want to make inferences about

E.g.

2.1. An aggregate about which we want to draw inferences by sampling

2.2. The totality of individual observations about which inferences are to be made, existing within a specified sampling area limited in time and space

Migration: Nm
Census: N
Reproduction

Time

Space

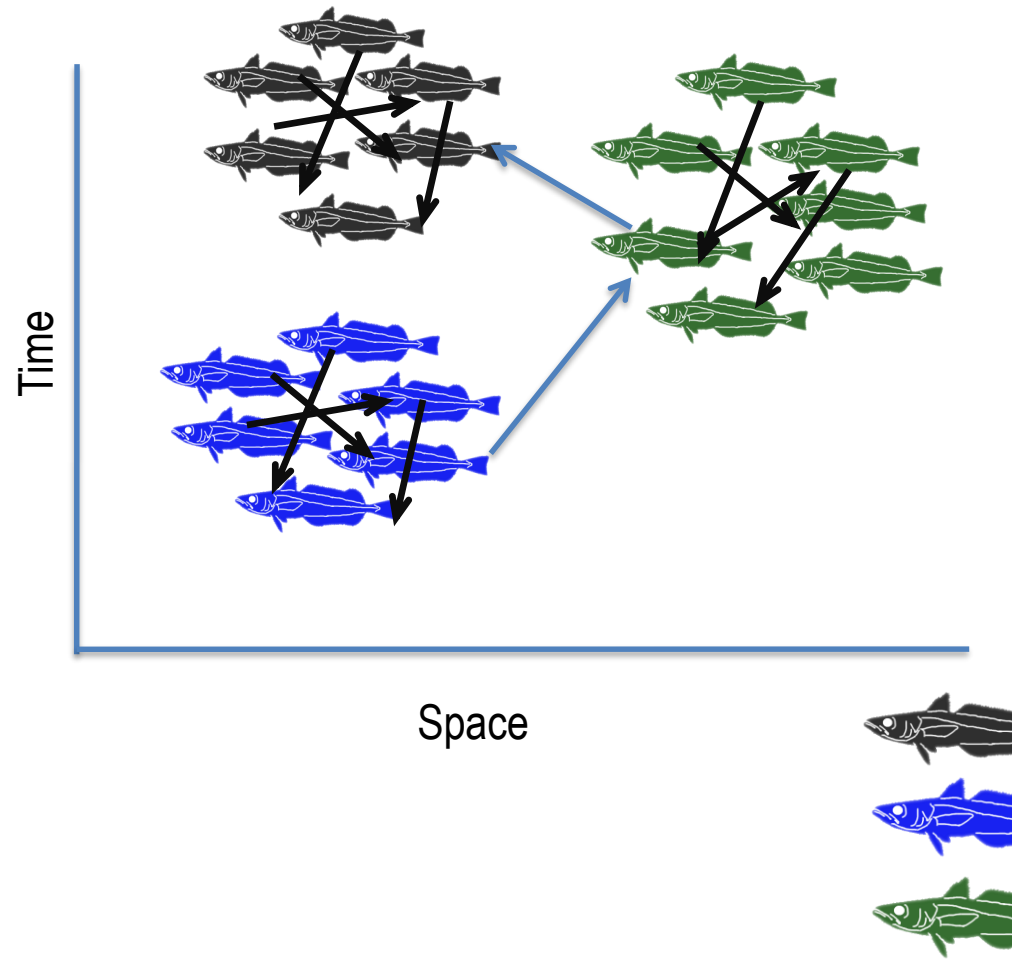## 3.   EVOLUTIONARY POPULATIONS

There are many definitions but all centered around the idea of reproductive cohesiveness and co-existence in time and space

E.g.
3.1. A community of individuals of a sexually reproducing species within which mating occurs

3.2. A major part of the environment in which selection takes place

3.3. A group of interbreeding individuals that exist together in time and space

4. Many other variants

Stock
A species, group or population of fish that maintains and sustains itself over time in a definable area
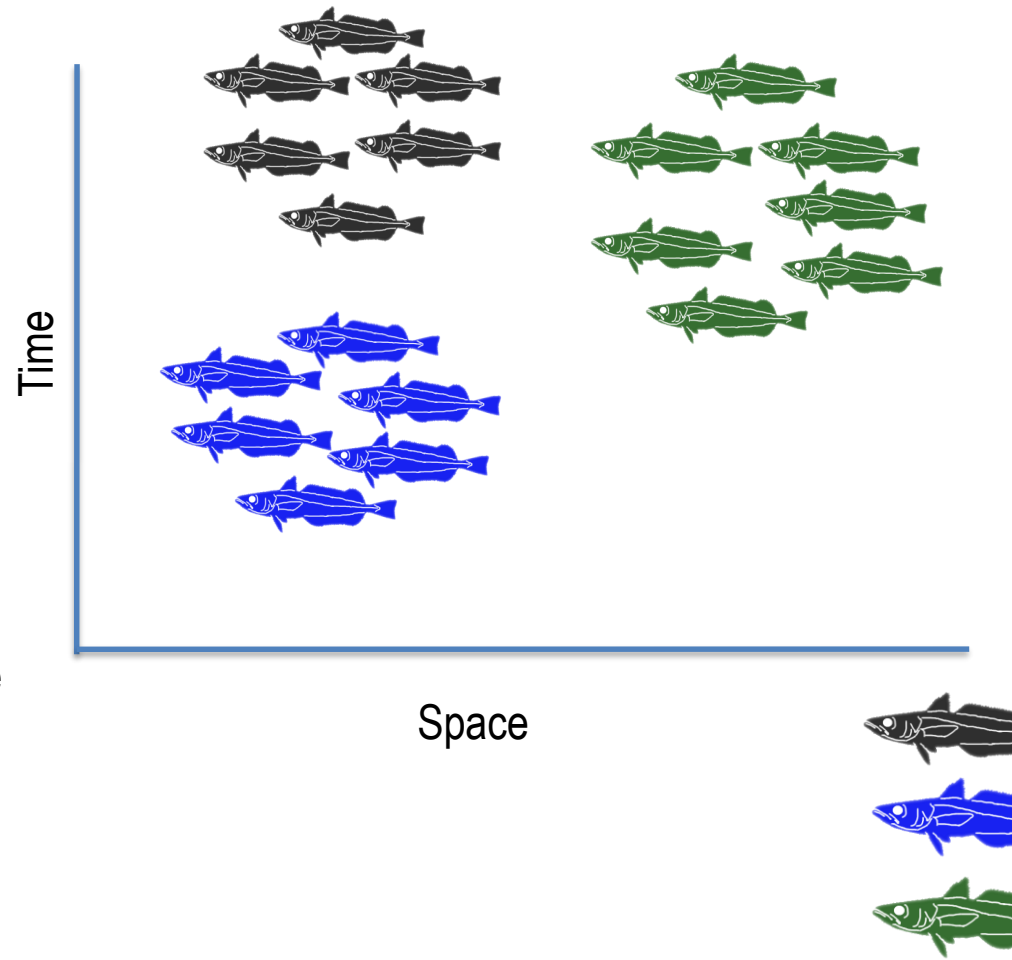
Demes
Separate evolutionary units

Natural population
Can only be defined by natural ecological or genetic barriers

Metapopulation
A group of spatially separated populations of the same species that interact at some level

Why do we actually need to define a population?

Since it is the central point for many studies/paradigms/theories it would be expected an objective and quantitative definition.

Independent researchers in independent fields in independent studies could apply it to the **same problem** and reach the **same conclusion**
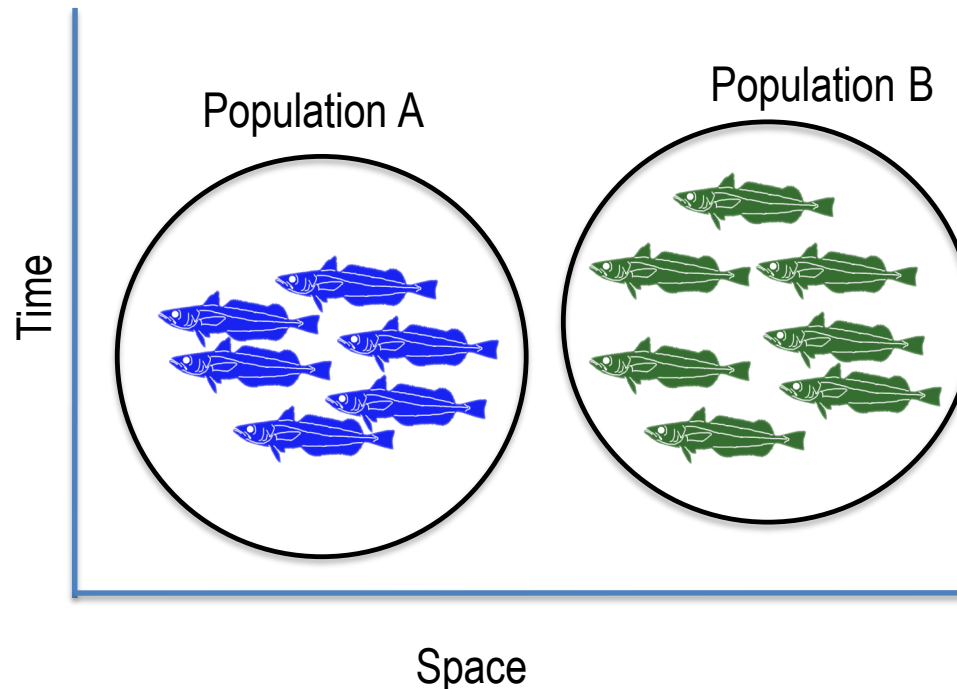
By relying on different definitions we over-complicate the problem

Also, because most methods used to identify populations are qualitative, different researchers, with the same information, can reach different conclusions

Evolutionary population

Ecological population

Stock

Deme
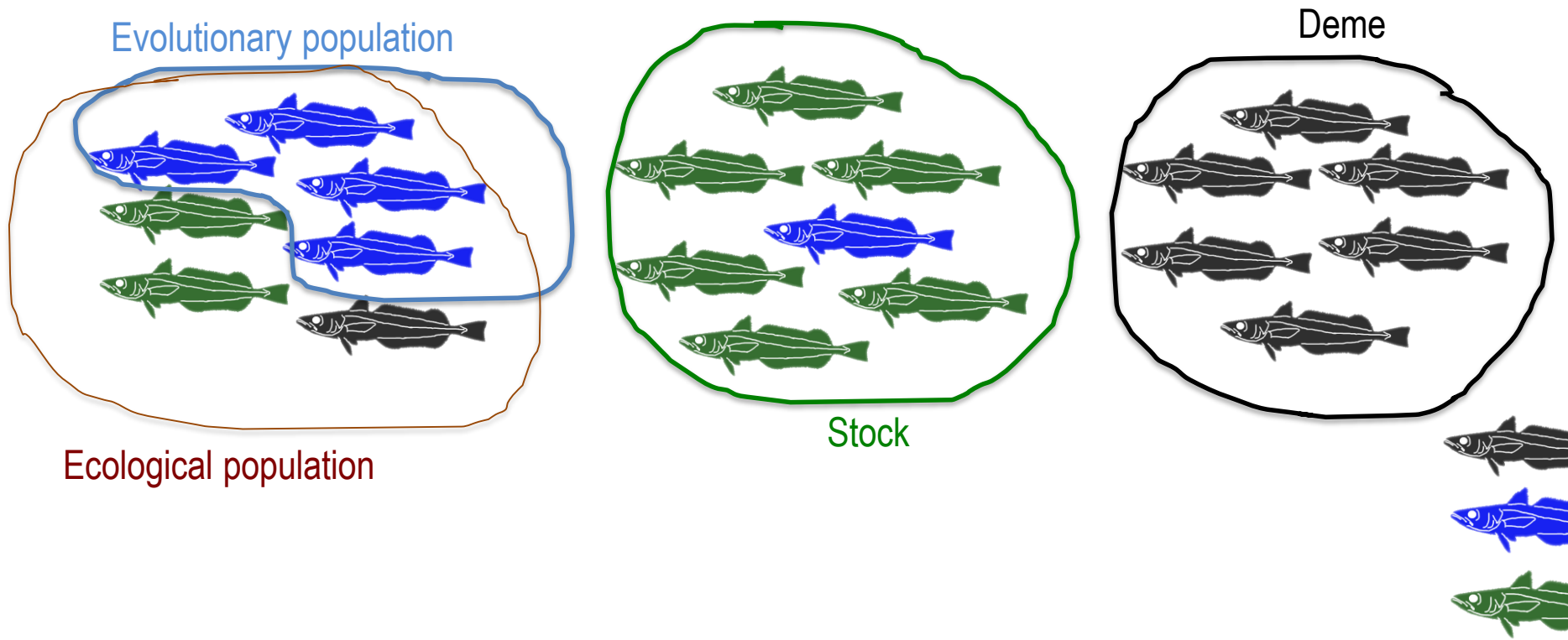
The advance of molecular studies, in particular the development of the field of POPULATION GENETICS, requires a uniform vision of what a population is

Molecular markers and new statistical assignment tools lead to an explosion of identifiable genetic units that can be connected to a certain degree, but still be used for management purposes
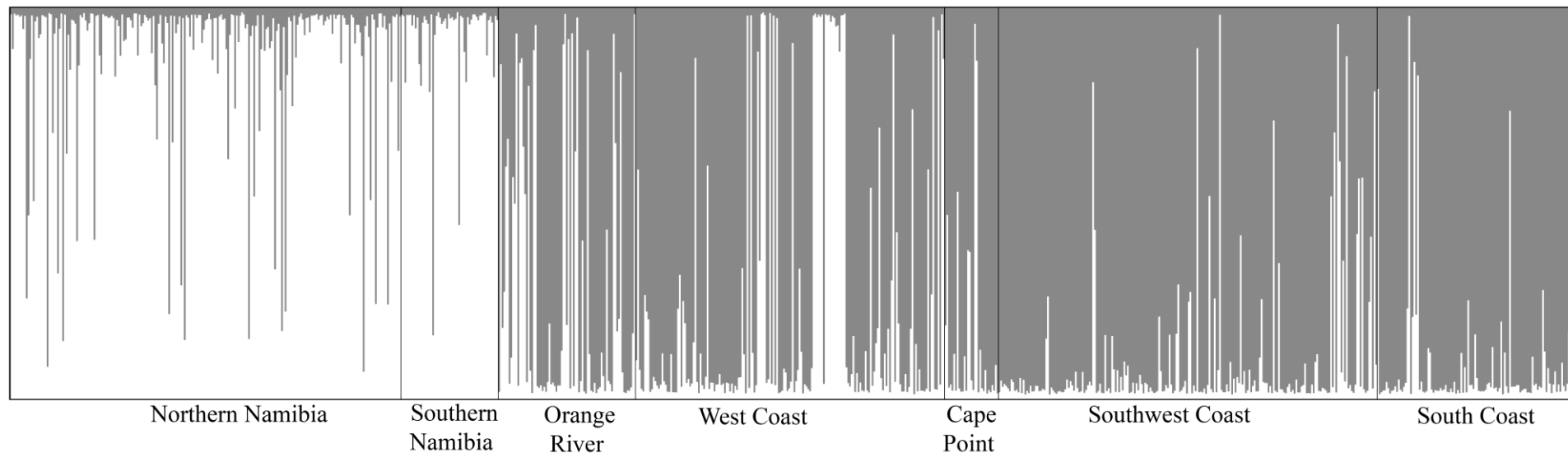
Structure assignment plot

But what is a population or what turns a group of individuals into a population remains debatable

So, we need metrics – quantifiable (as opposed to qualitative) tools that allow us to clearly define isolated groups of individuals
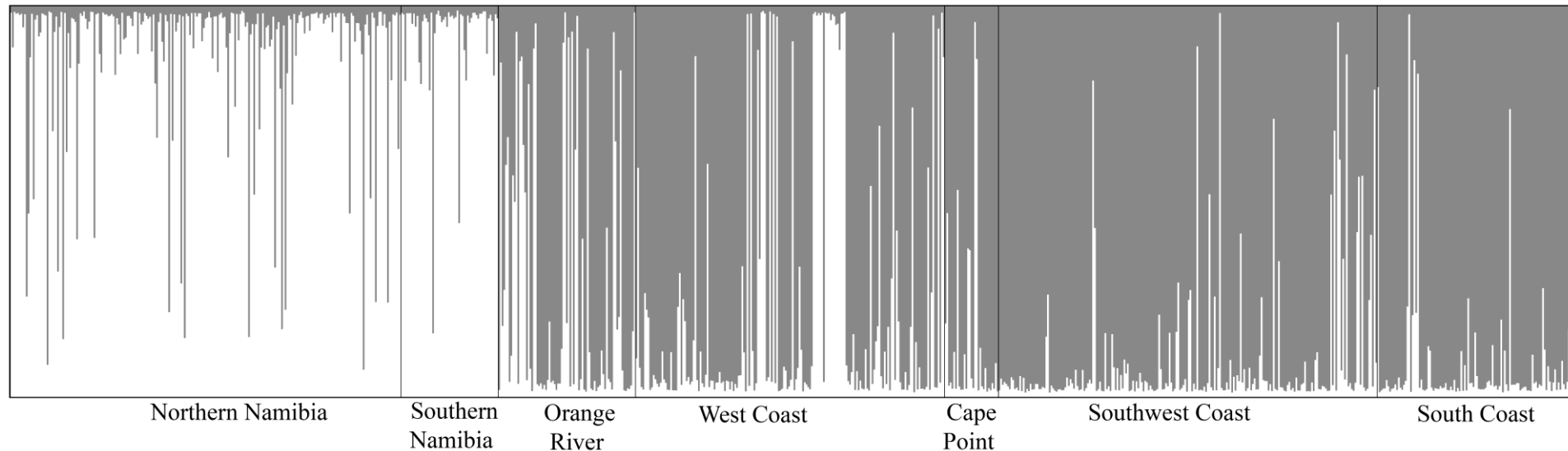
Structure assignment plot

Biological definition

Ecological perspective

A group of individuals of the same species that co-occur in time and space and have an opportunity to **INTERACT** with each other

Evolutionary perspective

A group of individuals of the same species living in close enough proximity that any member of the group can **MATE** with other members

Demographic forces
(competition, social, etc.)

Genetic forces
(reproduction, gene flow)

Biological definition

Random mating

Gene Pool

The one we use in Population Genetics

Evolutionary perspective

A group of individuals of the same species living in close enough proximity that any member of the group can **MATE** with other members
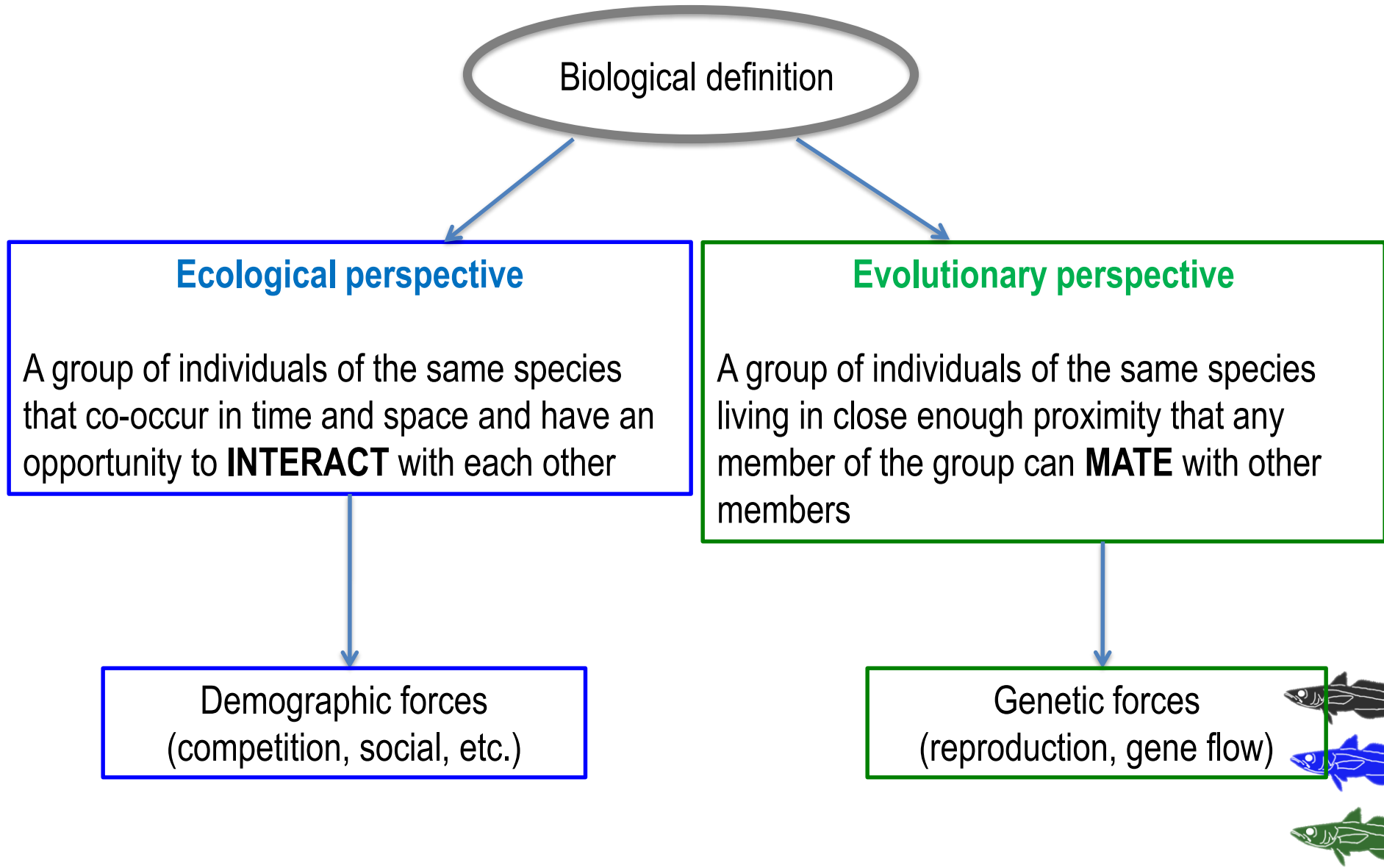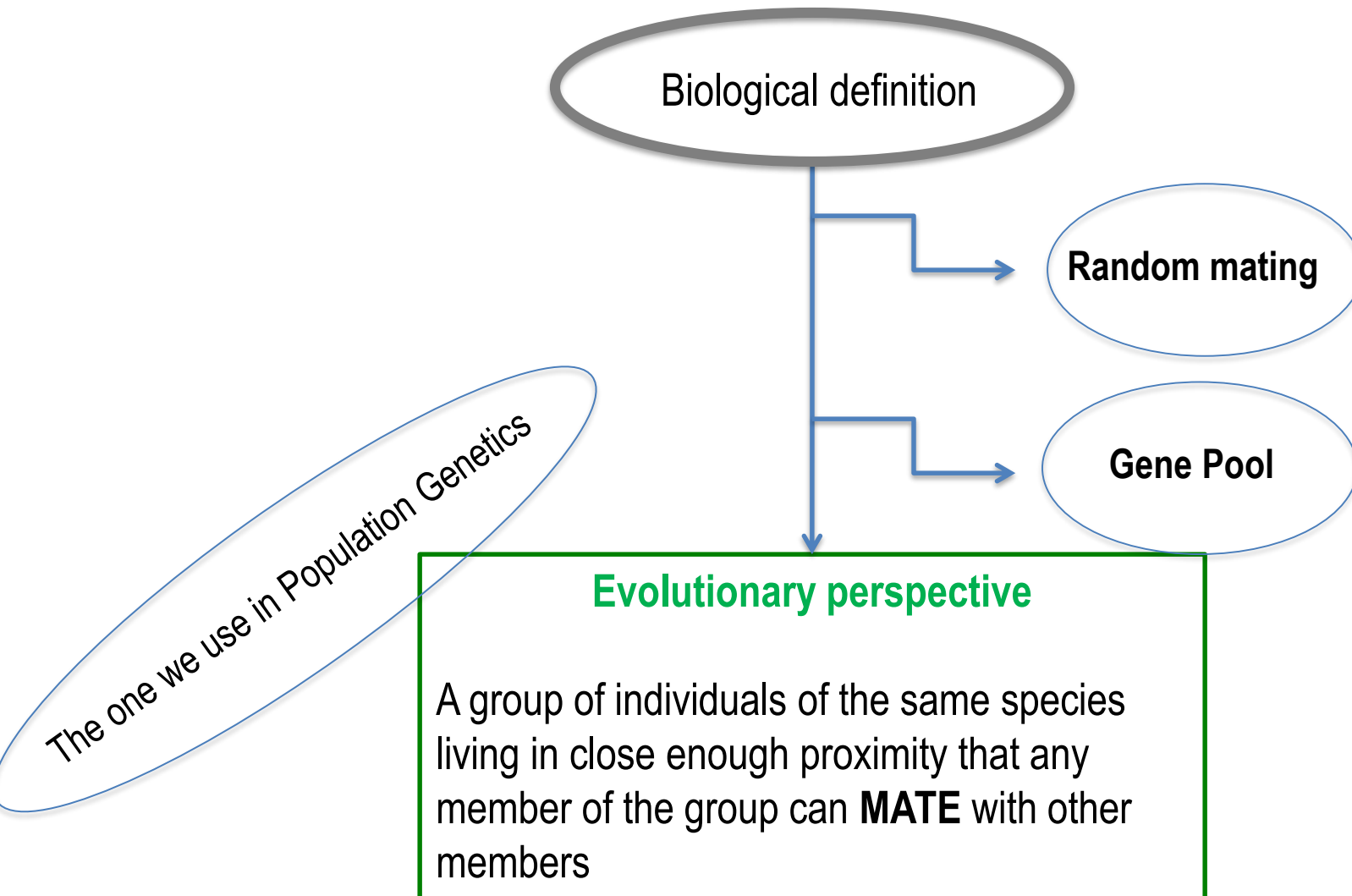
Two very important concepts in Population Genetics

Random mating

Gene Pool

Any individual has the same chance to mate with any other individual in a population
(as opposed to assortative mating)

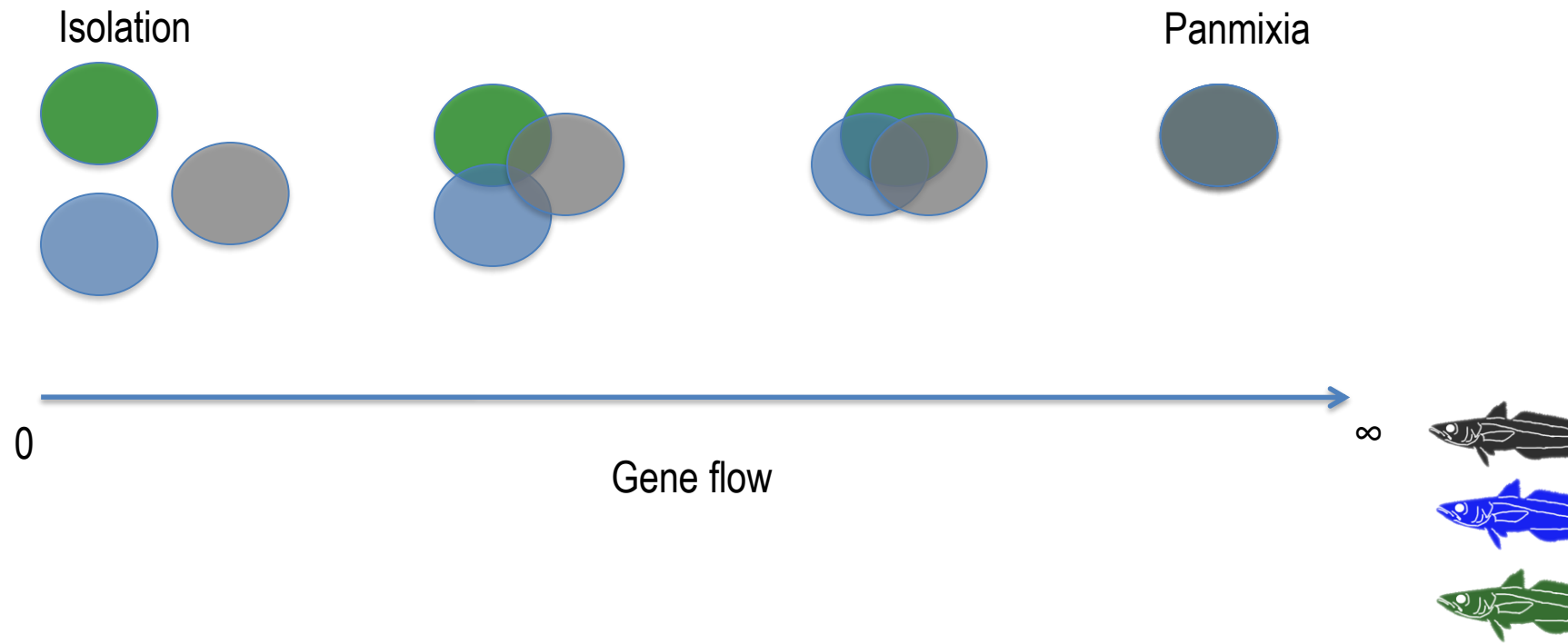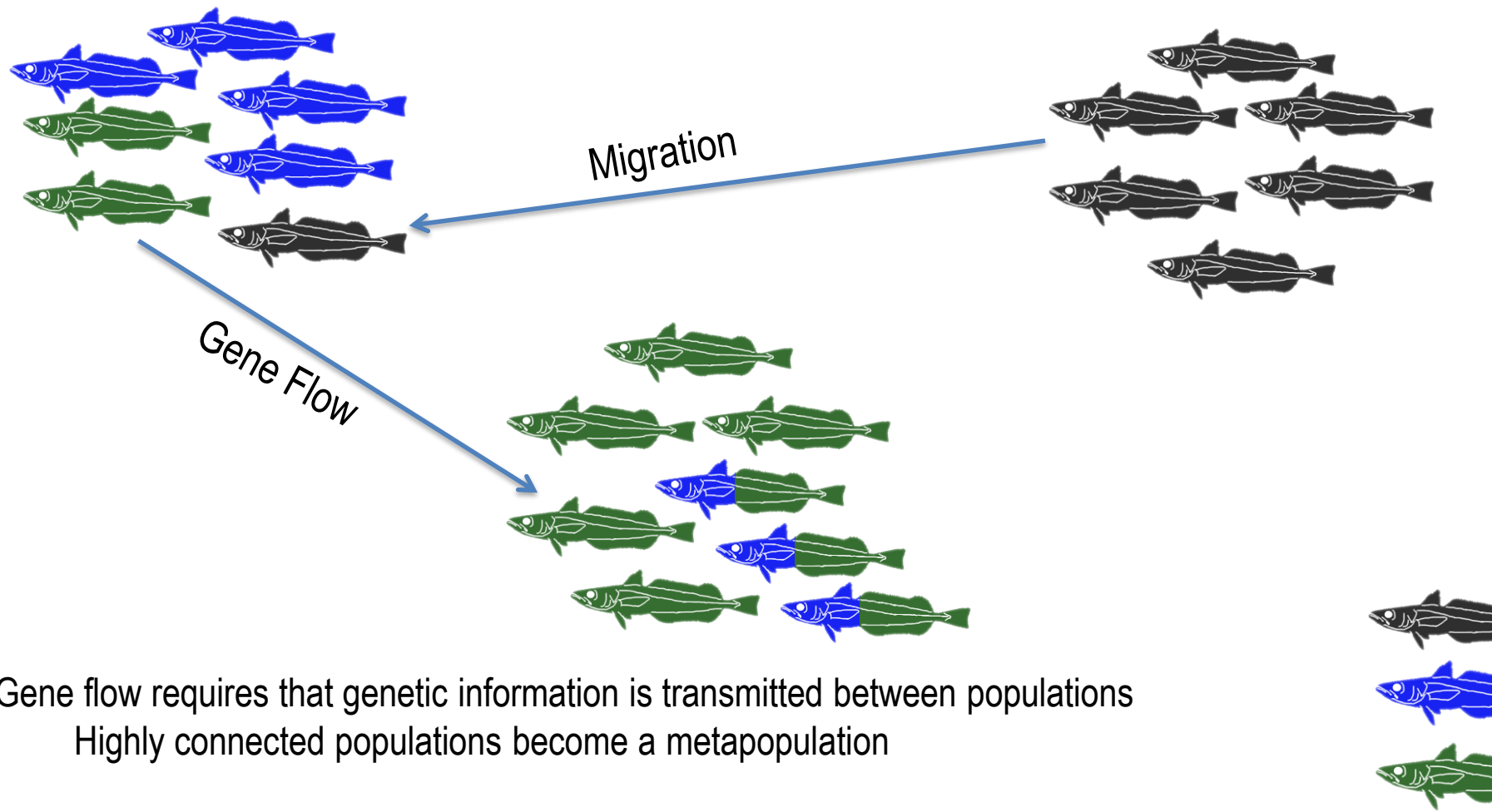Individuals that share the same gene pool form one single population

GENE FLOW

Gene Flow is a vital part in the identification of a population

How much is too much that an independent population looses its integrity and becomes fused with its neighboring populations?

Isolation

Panmixia

0   ∞

Gene flow

Gene Flow and Migration are not the same thing

Migration

Gene Flow

Gene flow requires that genetic information is transmitted between populations
Highly connected populations become a metapopulation

Isolation

Panmixia

0

∞
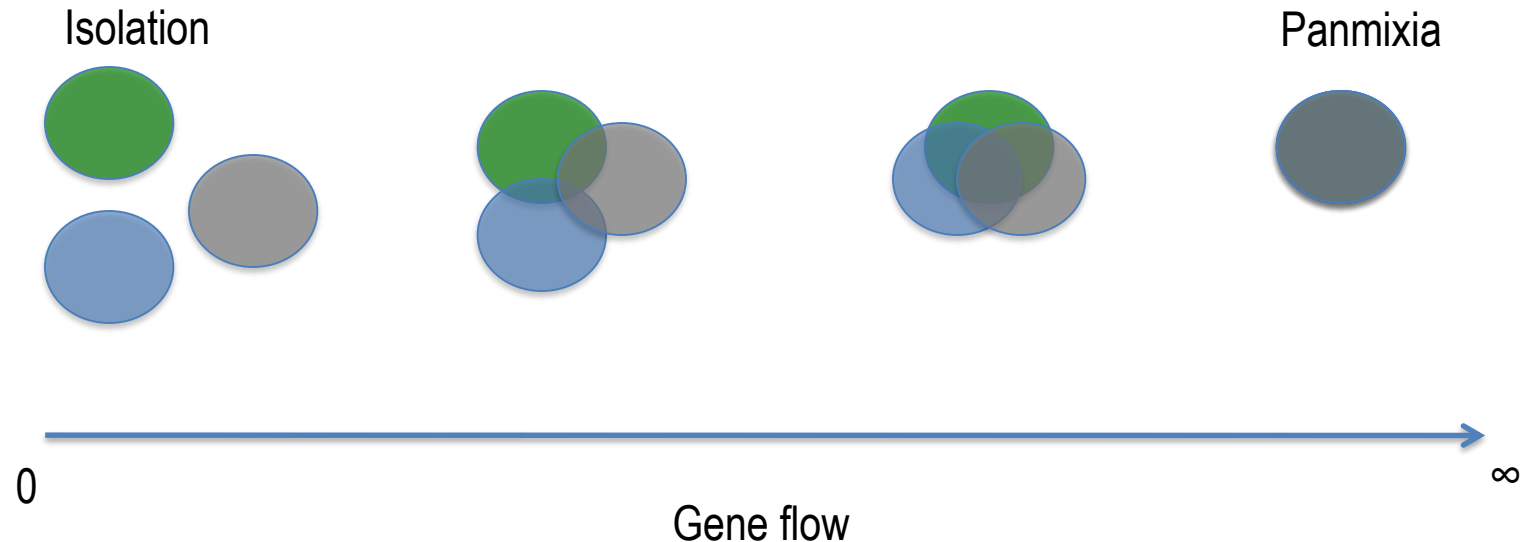
Gene flow

Reproductive cohesiveness is determined by the level of gene flow between populations, and the number of effective migrants (the ones that reproduce)

But we can have sub-populations anywhere in the middle of the gene flow continuum, and any departure from panmixia may imply the presence of genetically isolated populations

Isolation

Panmixia

0

∞

Gene flow
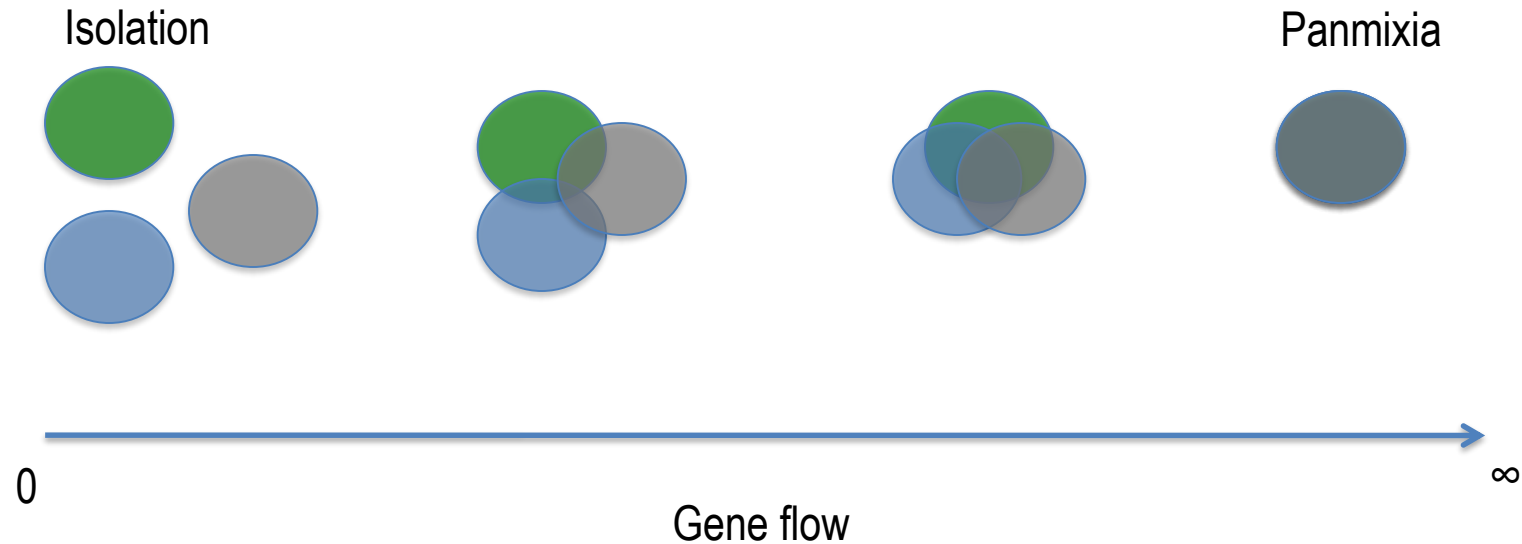
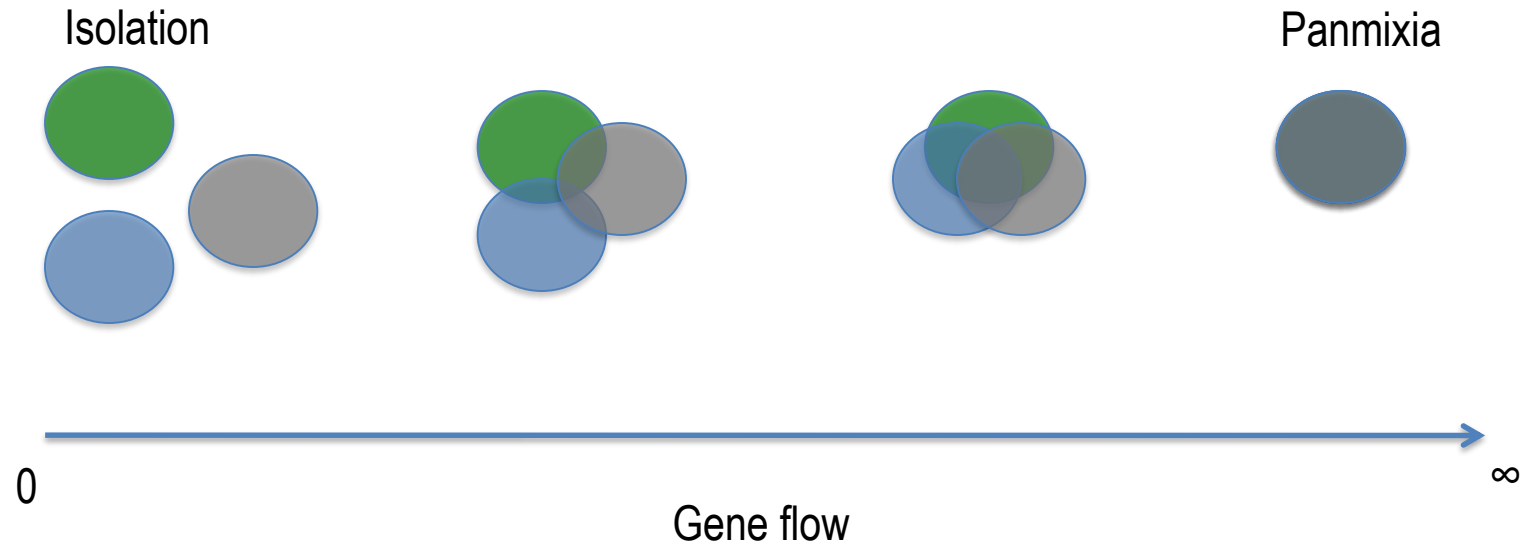How much is too much?

Several attempts have been made to quantify the minimum number of effective migrants between populations, in order to clarify their boundaries.

Isolation

Panmixia

0

∞

Gene flow

While some studies suggest that even **5** effective migrants are enough to homogenize populations and lose independency, there is no consensus regarding the actual number of migrants required

Isolation

Panmixia

0

∞
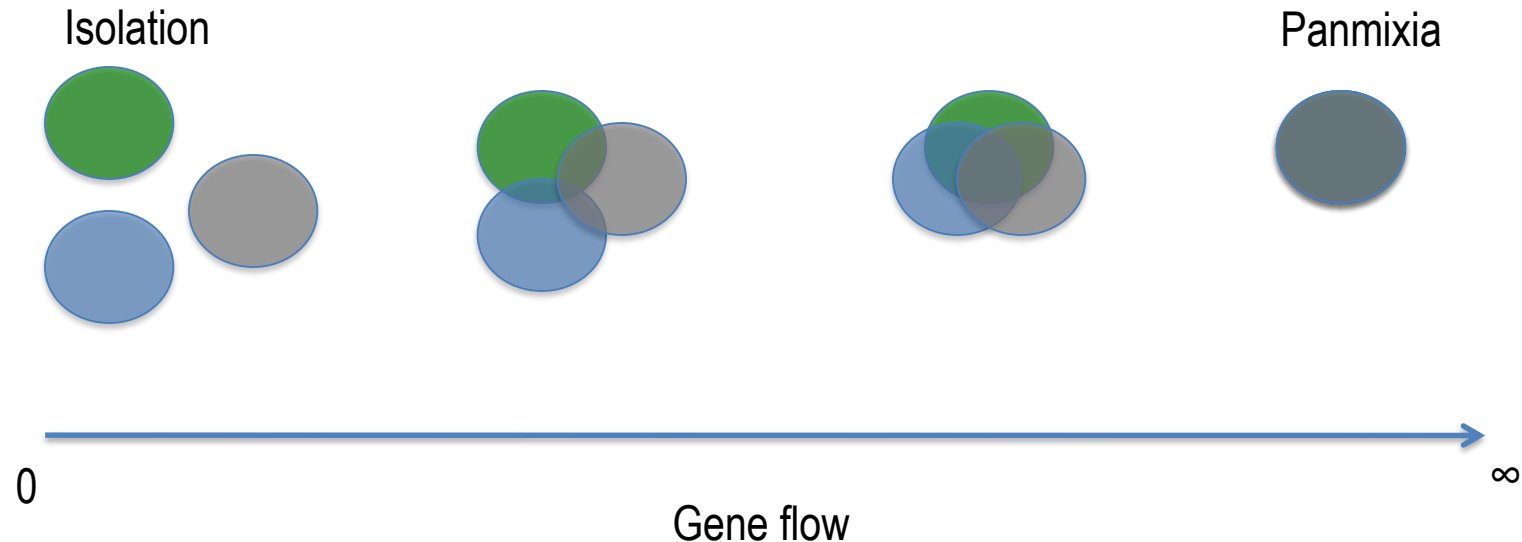
Gene flow

The correct answer to how much gene flow is too much gene flow is: Depends!

It depends on the species (abundance, genetic diversity)
It depends on the aims of the study (evolutionary vs. conservation)
It depends on the markers one uses (mutation rate)

PART 2

The four pillars of Population Genetics
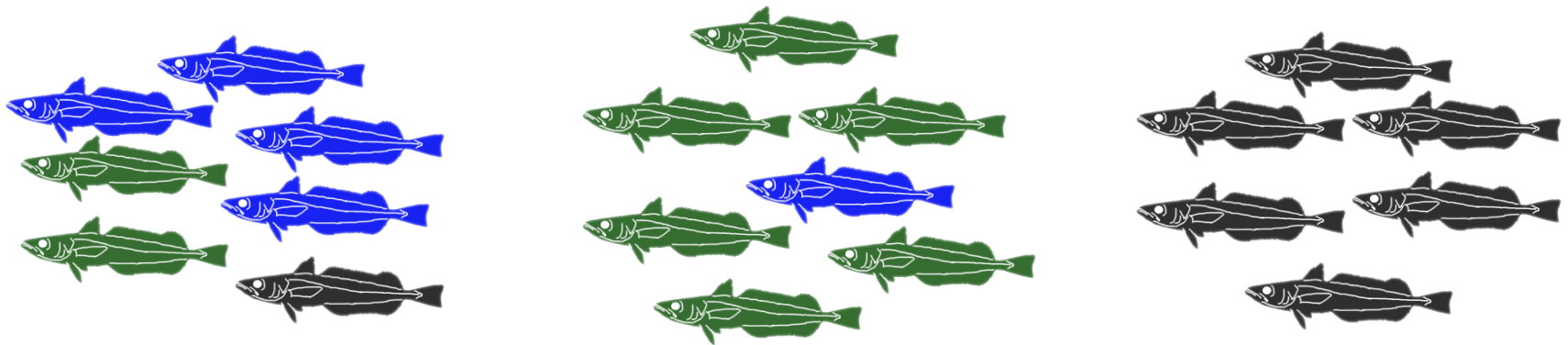
Population Genetics

Study of genetic variation within (and between) populations, by tracing changes in the frequencies of genetic variants in a population over time and space
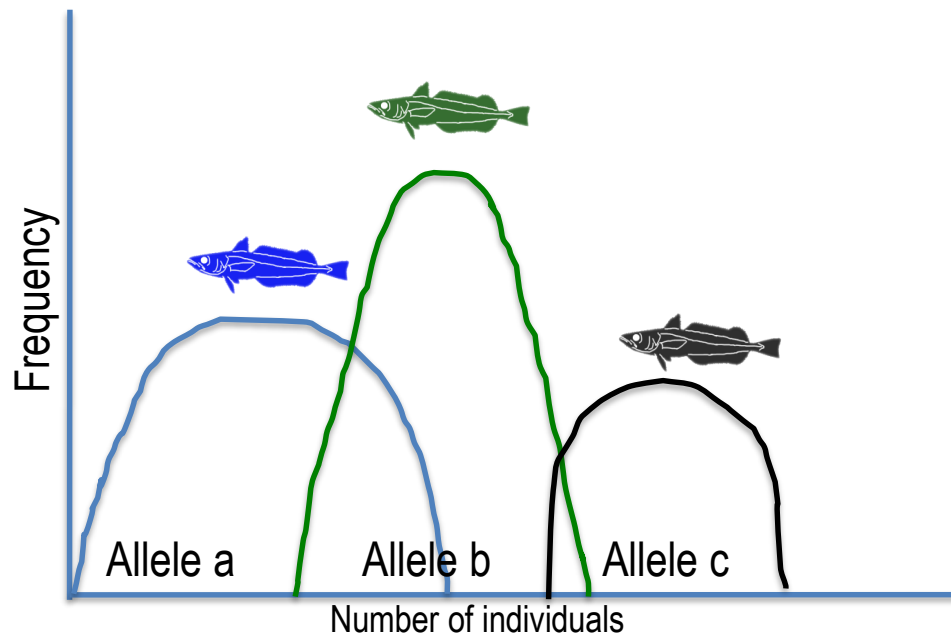
Because these changes are at the core of evolution and speciation, population genetic is often studied along with evolutionary genetics

How can there be variation in a Population?

Source of variation

↓

Mutation

Maintenance of variation

↓   ↓   ↓

Genetic Drift		Natural Selection

Gene Flow
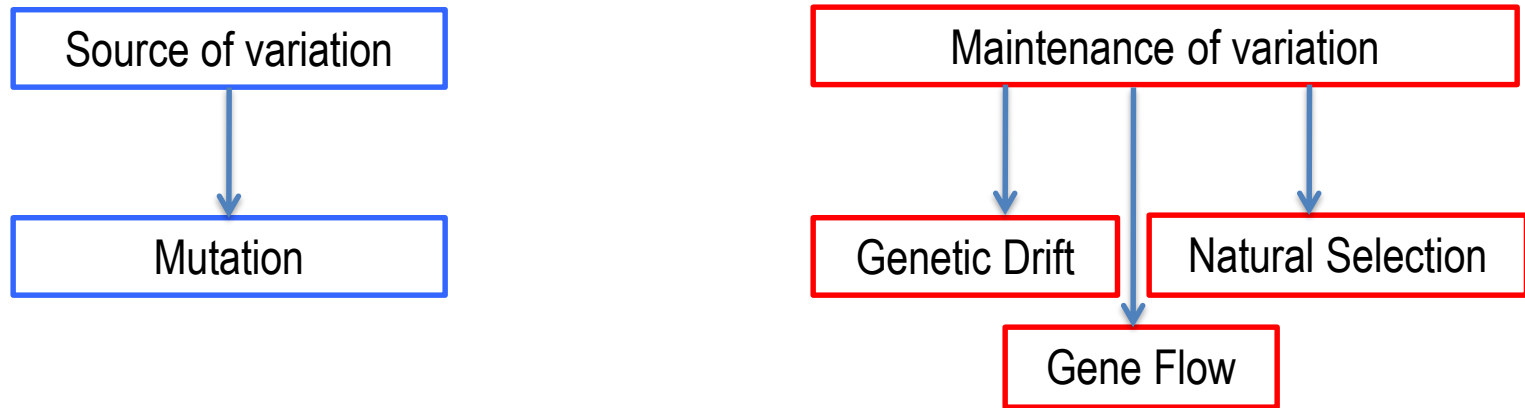
The origin of a change in a region or gene sequence is mutation, which generates new variants (alleles)

Mutations can remain in a population if they are not deleterious to the organism, and be spread within and between populations through three main mechanisms
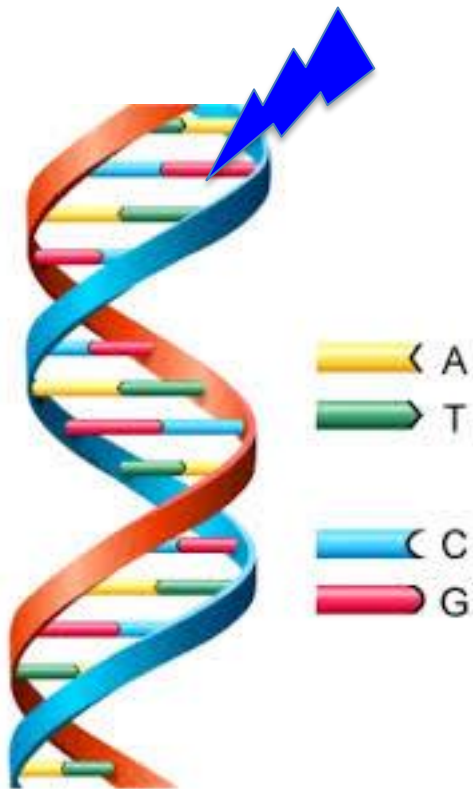
Mutation can change the nucleotides at any given site in the DNA, but it is not random

Due to chemical bonds, A is more likely to be replaced by a G, and a T by a C

Purines → transitions
Pyrimidines → transversions

ADENINE   THYMINE   GUANINE   CYTOSINE

A

T

C

G
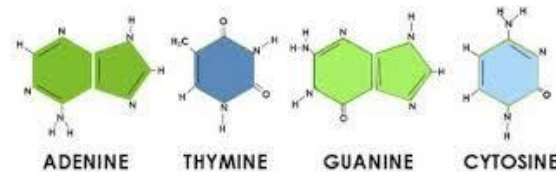
Genetic Drift

Alleles can be lost or retained due to random events (chance)

Has the same expected effect throughout the genome

Its ability to alter a genetic composition of a population depends on the effective population size:
in large populations, drift has a reduced effect
in a small population it can significantly affect the genetic make-up in a matter of generations
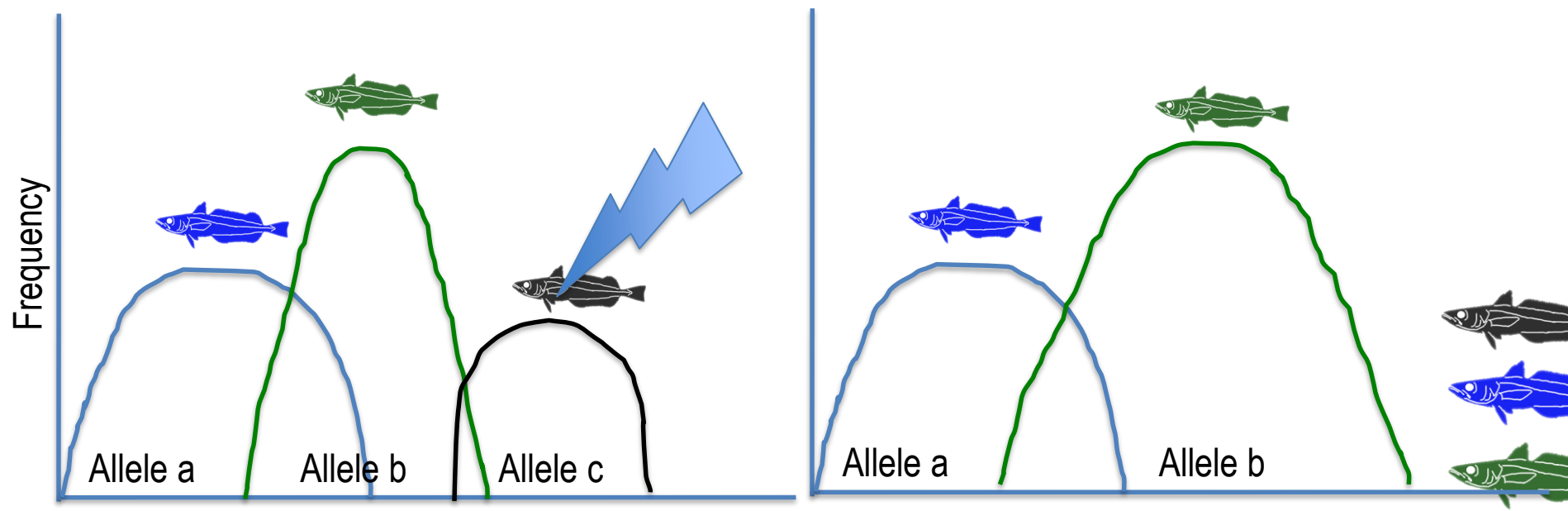
Genetic Drift

Stable populations can reach a mutation-drift equilibrium, where the gain of genetic variants is compensated by the loss due to genetic drift

When populations are in mutation-drift equilibrium it is easy to make predictions regarding gene flow, population differentiation and evolutionary history

Genetic drift is non-directional
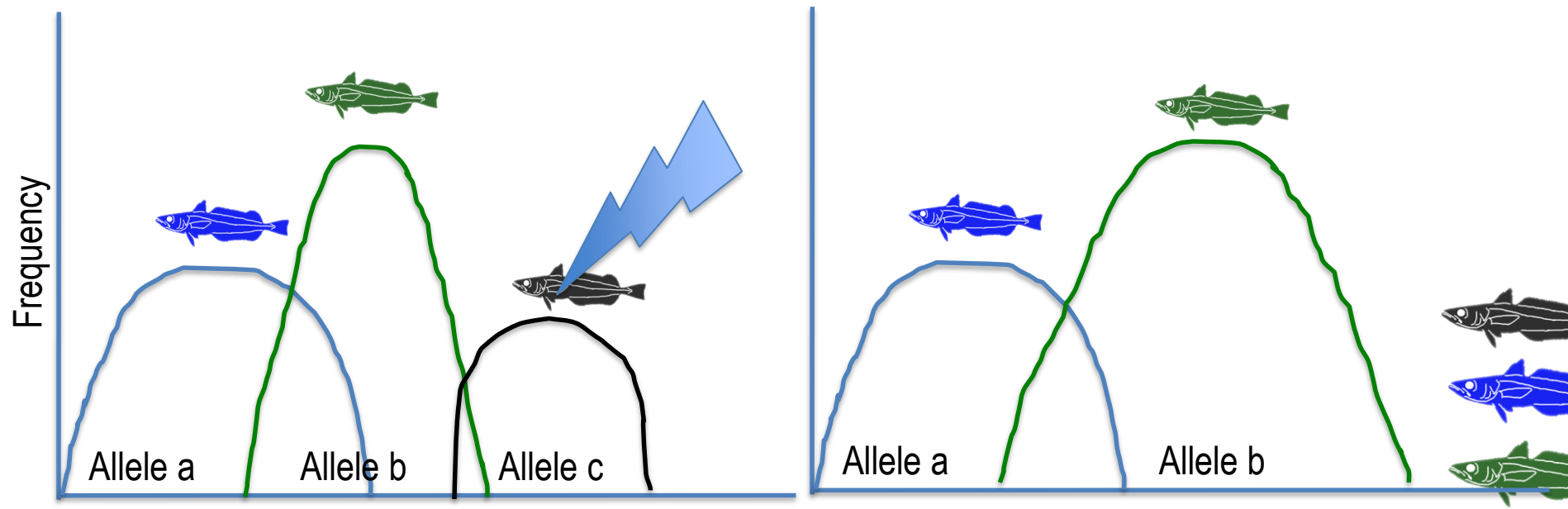
Genetic Drift

Very common mechanisms: Founder effect and bottlenecks

Founder effects – in this case, only a few individuals from the original population successfully colonize a new environment. Not all the original genetic variability is represented in the new population

e.g. The colonization of St. Helena Island

After the retreat of the British soldiers with the death of Napoleon (XIX century), only 15 people were left.
In 1961 there was a volcanic eruption and the whole island was moved back to England (N = 260 pax).

A genetic study revealed a higher incidence of *retinitis pigmentosa*, a recessive eye disease, in the islanders than expected

Genetic Drift

Very common mechanisms: Founder effect and bottlenecks

Bottleneck – reduction of a population by a random event for at least one generation

Time ▶

Catastrophic event
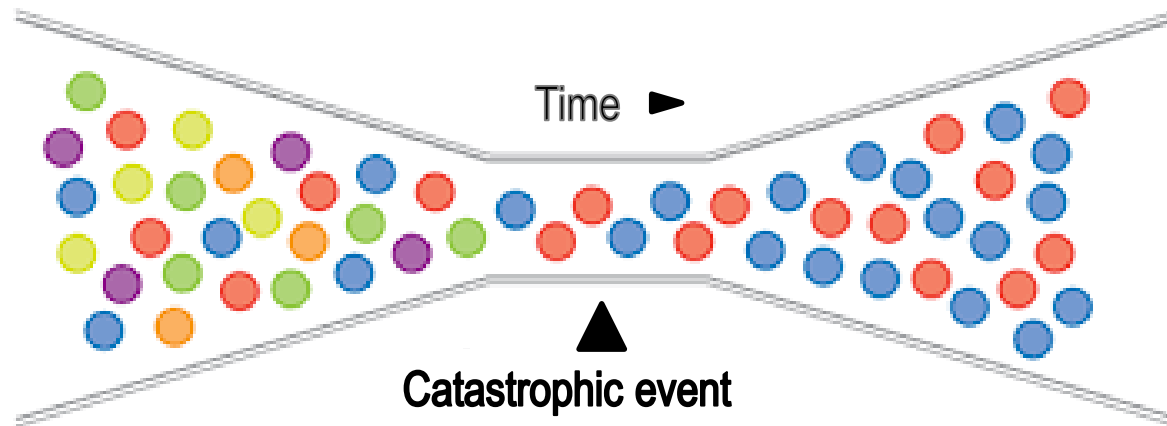
Like with a founder effect, the genetic composition of the resulting population does not reflect the genetic variability of the original population

Gene Flow

The spread of new variants can be greatly facilitated by gene flow among populations

Has the same effect throughout the genome

The homogenization can occur even at low levels of gene flow

And gene flow can be **asymmetrical**

Natural selection

Although it is believed that natural selection is the dominant force shaping the course of evolution is actually quite difficult to quantify
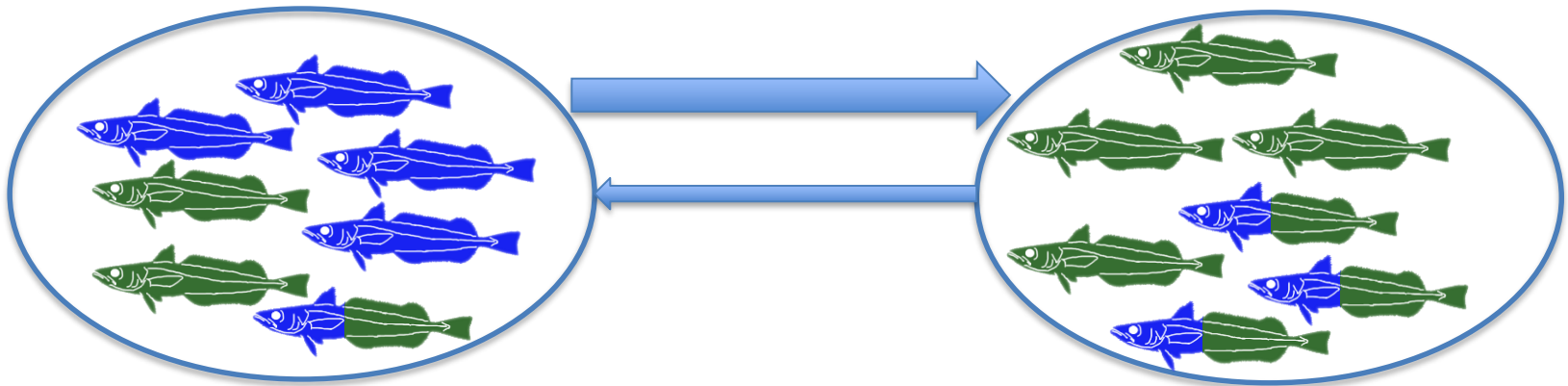
Alleles can be fixed or lost at a higher frequency than expected by random chance

Its effects are very localized in the genome (not widespread) – affecting a few genes at a time

It reflects external pressures to obtain the best fit phenotype/behaviour/morphology

Can also occur in a few generations
e.g. evolution of a freshwater form in marine sticklebacks in 50 years
(Lescak *et al.* 2015)

Natural selection

Sticklebacks have three forms: marine, estuarine and freshwater

In 1964, an earthquake caused a geological uplift of islands in the Prince William Sound and the Gulf of Alaska

This trapped marine fish in a brackish (and later freshwater) environment

Majority of species reverted back to a freswater form after 10 to 50 years

The speed of change will depend on the size of the population but also on the strength of the driver

Commonly used genetic diversity measures

Heterozygosity or Gene diversity
- Proportion of genotypes composed by different alleles
- Can be extrapolated to quantify the amount of DNA variation for several DNA sequences
- Not sensitive to variations in the number of loci
- Varies between expected (HE) and observed (HO)
- Uses the allelic frequencies in a population

$$HE = 1 - \Sigma pi^2 \quad HO = 1 - \Sigma pij^2$$

pi – frequency of allele p in population
pij – observed frequency

Why expected and observed heterozygosity?

As mentioned before, the field of population genetics operates under a mutation-drift equilibrium model, which is represented in the Hardy-Weinberg equation

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

Assumptions: infinite size (constant), random mating, diploid organisms, non-overlapping generations, sexual reproduction, negligible effects of mutation, gene flow and selection

This is an over-simplification, but it allows to describe a population's genetic composition by reducing the number of parameters that need to be considered

Only the frequencies of the alleles at a certain locus need to be used, instead of the frequencies of all diploid genotypes

If populations are not in H-W equilibrium (which is very common) then one of the assumptions has been violated (generally the outcrossing)

Commonly used genetic diversity measures

Number of Alleles
- Number of alleles present per locus in a population
- Varies with sampling size and the type of loci used
- Allelic richness is a rarefied measure of the number of alleles
- Does not have an upper limit – it varies between loci due to mutation rates and models

Inbreeding coefficient
- Measures deviations to the expectation of outcrossing, by estimating the inbreeding load of an individual in relation to its sub-population
- Varies between -1 and 1
- Inbreeding will impact heterozygosity – high inbreeding decreases genetic diversity

Why does Genetic Diversity matter?

The evolutionary potential of a population is proportional to the amount of genetic diversity

High genetic diversity is generally linked to higher resilience to change

For example, high heterozygosity measures have been linked to short-term response to selection, while allelic richness is a better predictor for long-term and total response

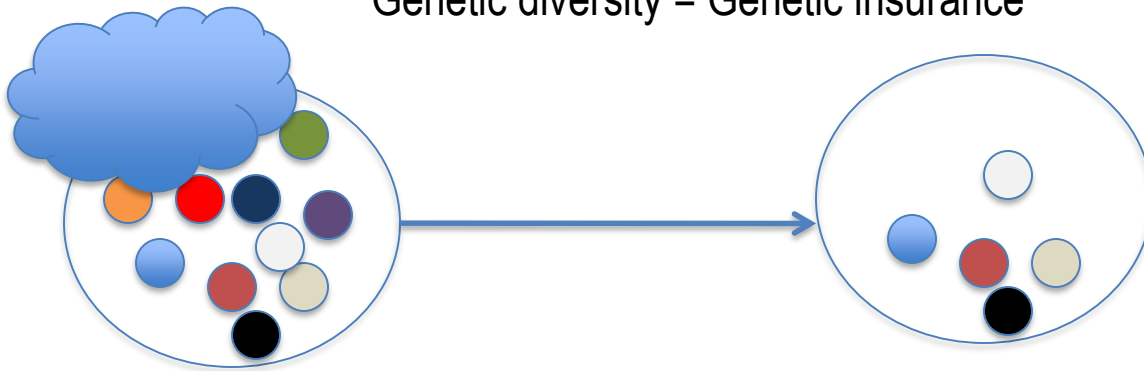Genetic diversity = Genetic insurance
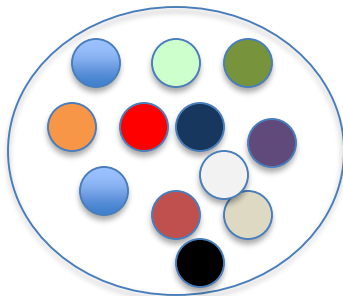
If the conditions change, or if there is a random loss of diversity, populations that have higher genetic diversity are more likely to persist in time

Why does Genetic Diversity matter?

Genetic diversity = Genetic insurance

However, genetic diversity is also linked to effective population size, a.k.a. the total number of breeders/spawners

Species with high effective population sizes have generally higher genetic diversity, and thus should be better equipped to resist random changes (drift)

Species with lower effective population size have lower genetic diversity and are generally more vulnerable

Why does Genetic Diversity matter?

The cheetahs example

12 000 years ago there was a extinction event, that erased 75% of the mammals populations

A few cheetahs survived this bottleneck and the population recovered, but from a small effective population size

Cheetahs are, today, severely genetically depleted and very susceptible to the same infectious diseases

But they haven't gone extinct!

Why?
Environment hasn't changed that much
Remote habitat
Cheating cheetahs! Females have multiple partners

Why does Genetic Diversity matter?

The commercially exploited fishes example

60% of all cod and herring fisheries are either fully exploited or over-exploited

Fishing decreases abundance and species richness, but conclusive proof that it decreases genetic diversity is quite difficult to obtain

Because of traditionally high population numbers, it is difficult to ascertain reductions in heterozygosity, since even when a fishery is collapsed there might still be millions of fish in the sea
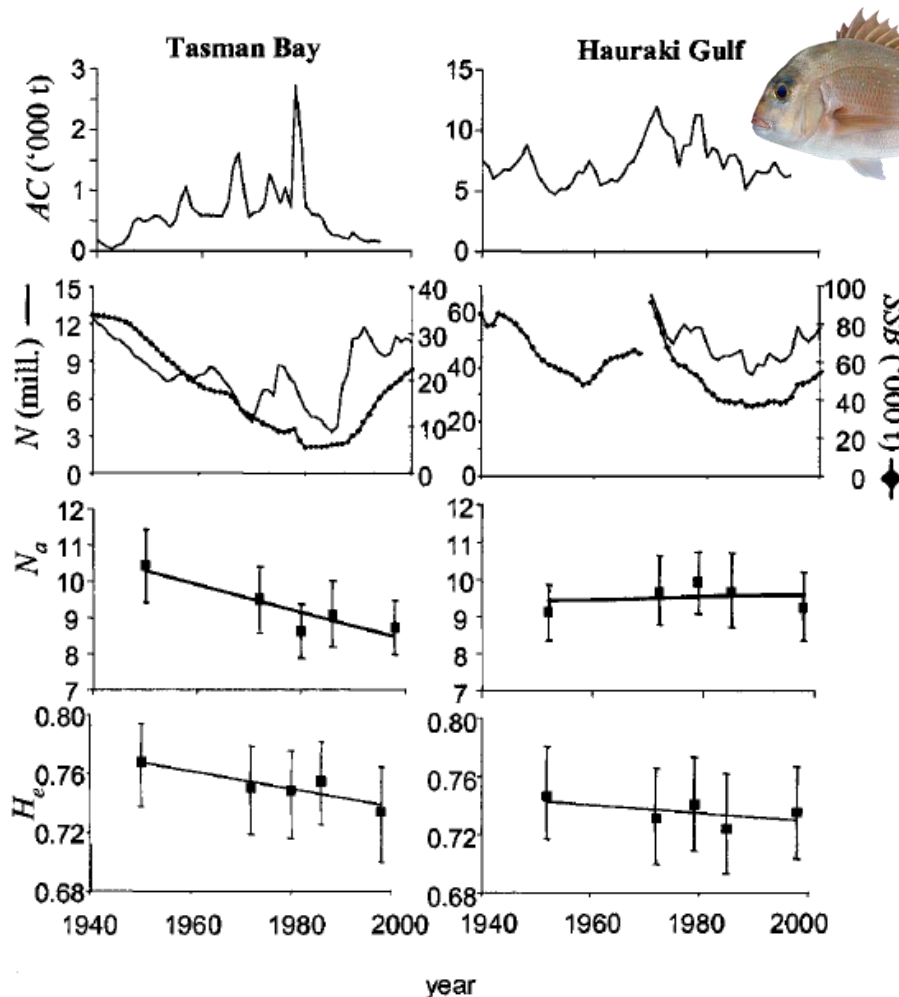

But census size is not effective population size!

Reduction of genetic diversity can actually be detected in marine fishes

Reduction of genetic diversity in *Pagrus aurata* (Hauser et al. 2002)

Microsatellite analysis of a time series of archived samples

Significant decline in genetic diversity of *Pagrus aurata* off New Zealand

Decline linked with historical exploitation levels

QUESTIONS?