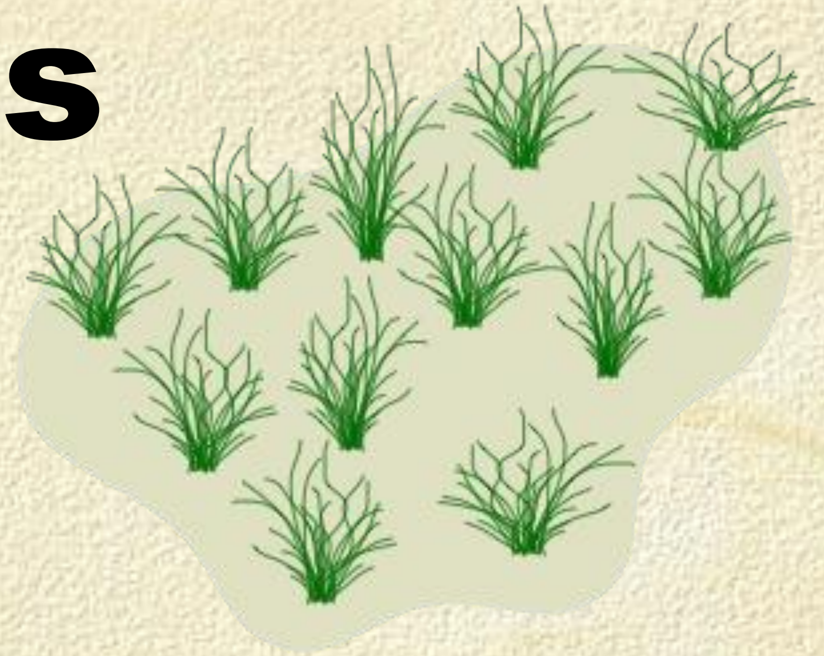
Population Genetics

Species

- A **biological species** is:
  a grouping of organisms that can interbreed and are **reproductively isolated** from other such groups.
- Species are recognized on the basis of their **morphology** (size, shape, and appearance) and, more recently, by genetic analysis.
- For example, there are up to **20 000 species of butterfly**; they are often very different in appearance and do not interbreed.

Populations

- From a **population genetics** viewpoint:
  - A **population** comprises the total **number of one species** in a particular area.
  - All members of a population have the potential to interact with each other. This includes **breeding**.
- Populations can be very large and occupy a large area, with fairly **continuous distribution**.
- Populations may also be limited in their distribution and exist in **isolated pockets** or “islands”, cut off from other populations of the same species.

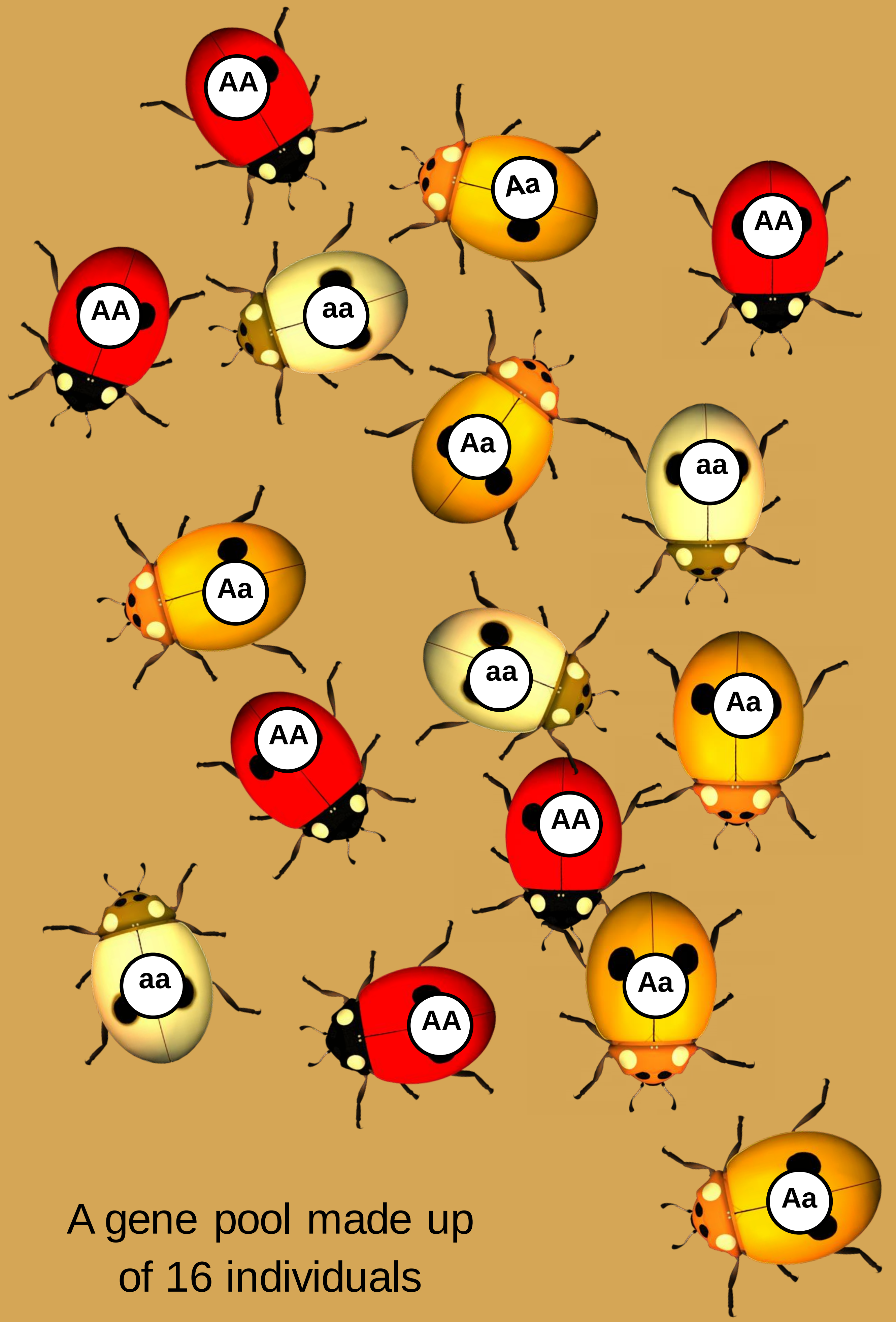


Continuous distribution
Example: human population, Arctic tundra plant species

Fragmented distribution
Example: Some frog species

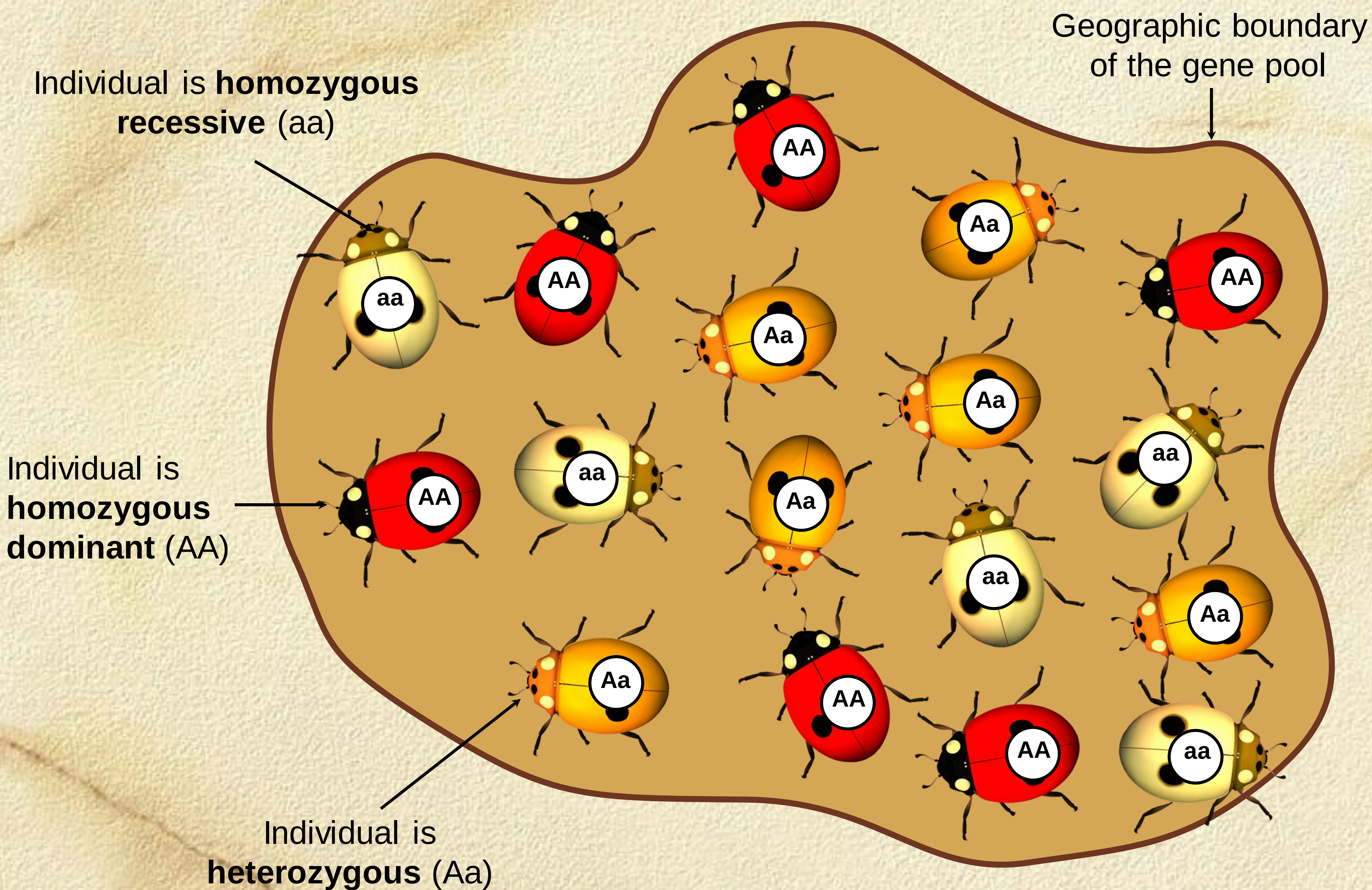
Gene Pool

- A **gene pool** is defined as the sum total of all the genes present in a population at any one time.
  - Not all the individuals will be breeding at a given time.
  - The population may have a distinct geographical boundary.
  - Each individual is a carrier of part of the total genetic complement of the population.



A gene pool made up of 16 individuals

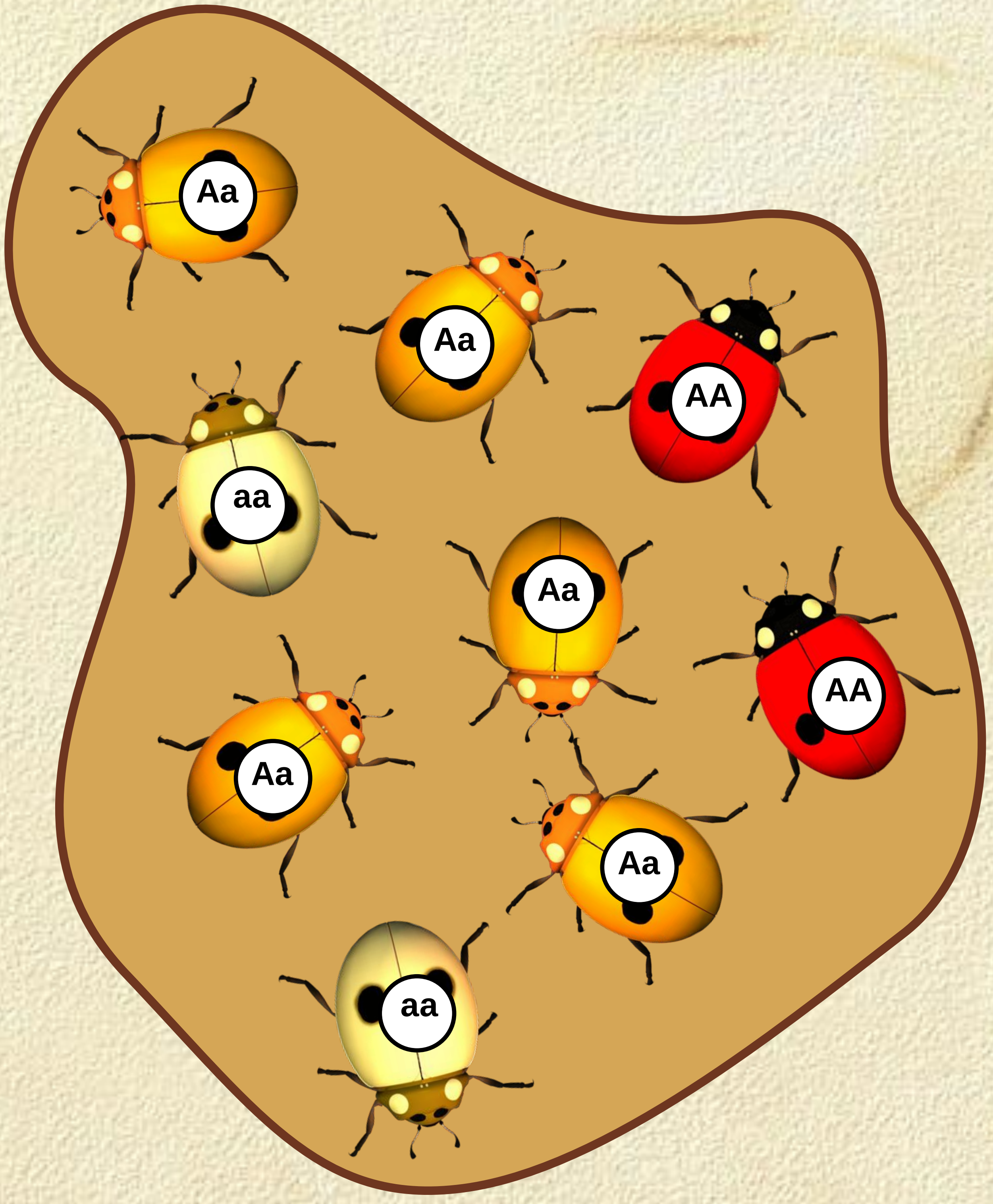
Gene Pool



A gene pool made up of 16 individual organisms with gene A, and where gene A has two alleles

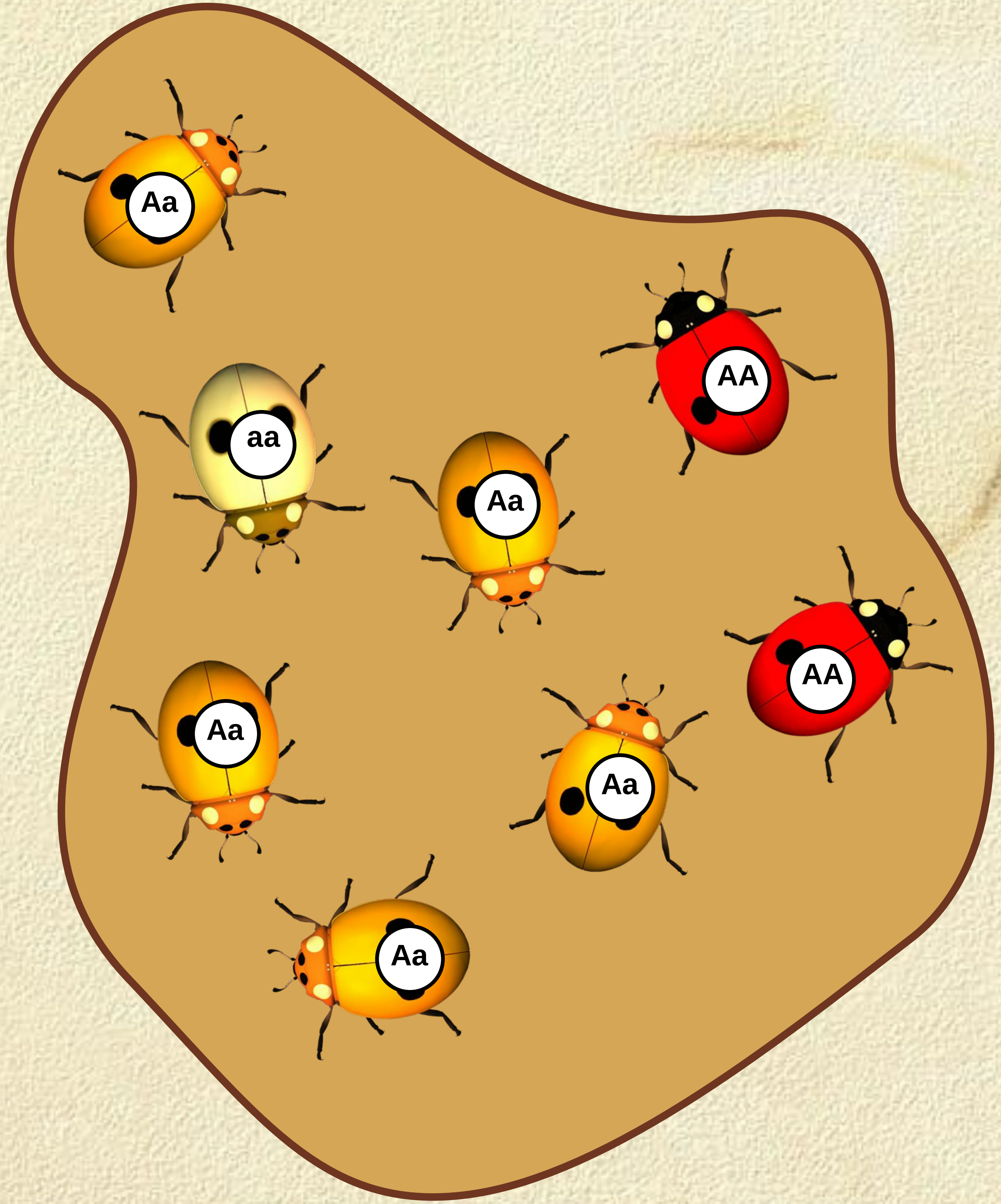
Analyzing a Gene Pool

- By determining the frequency of **allele types** (e.g. A and a) and **genotypes** (e.g. AA, Aa, and aa) it is possible to determine the **state** of the gene pool.
- The state of the gene pool will indicate if it is **stable** or **undergoing change**. Genetic change is an important indicator of evolutionary events.
- There are twice the number of alleles for each gene as there are individuals, since each individual has two alleles.



Analyzing a Gene Pool

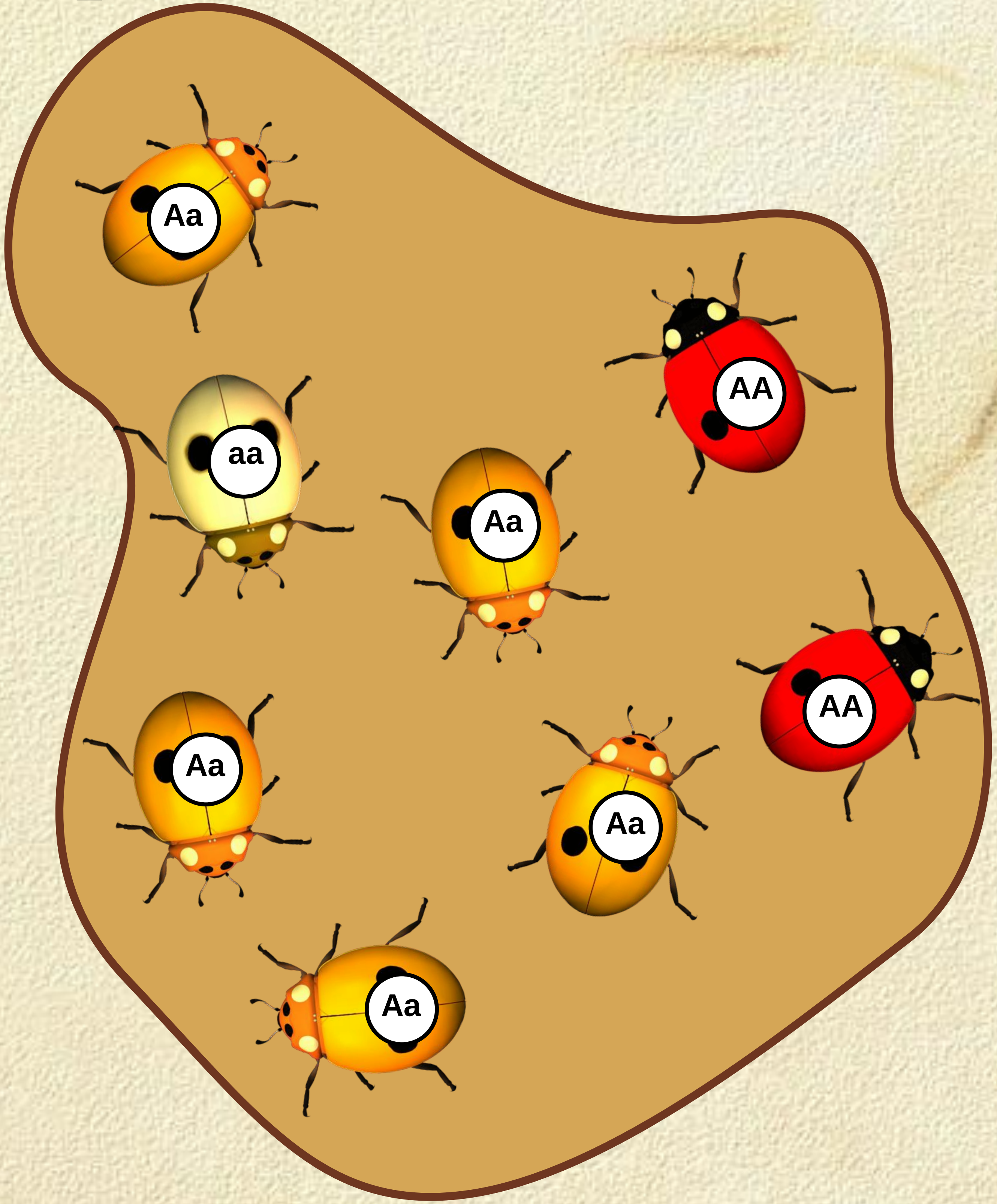
- **EXAMPLE**
  - The small gene pool above comprises 8 individuals.
  - Each individual has 2 alleles for a single gene A, so there are a total of 16 alleles in the population.
  - There are individuals with the following genotypes:
    - homozygous dominant (**AA**)
    - heterozygous (**Aa**)
    - homozygous recessive (**aa**)



Determining Allele Frequencies

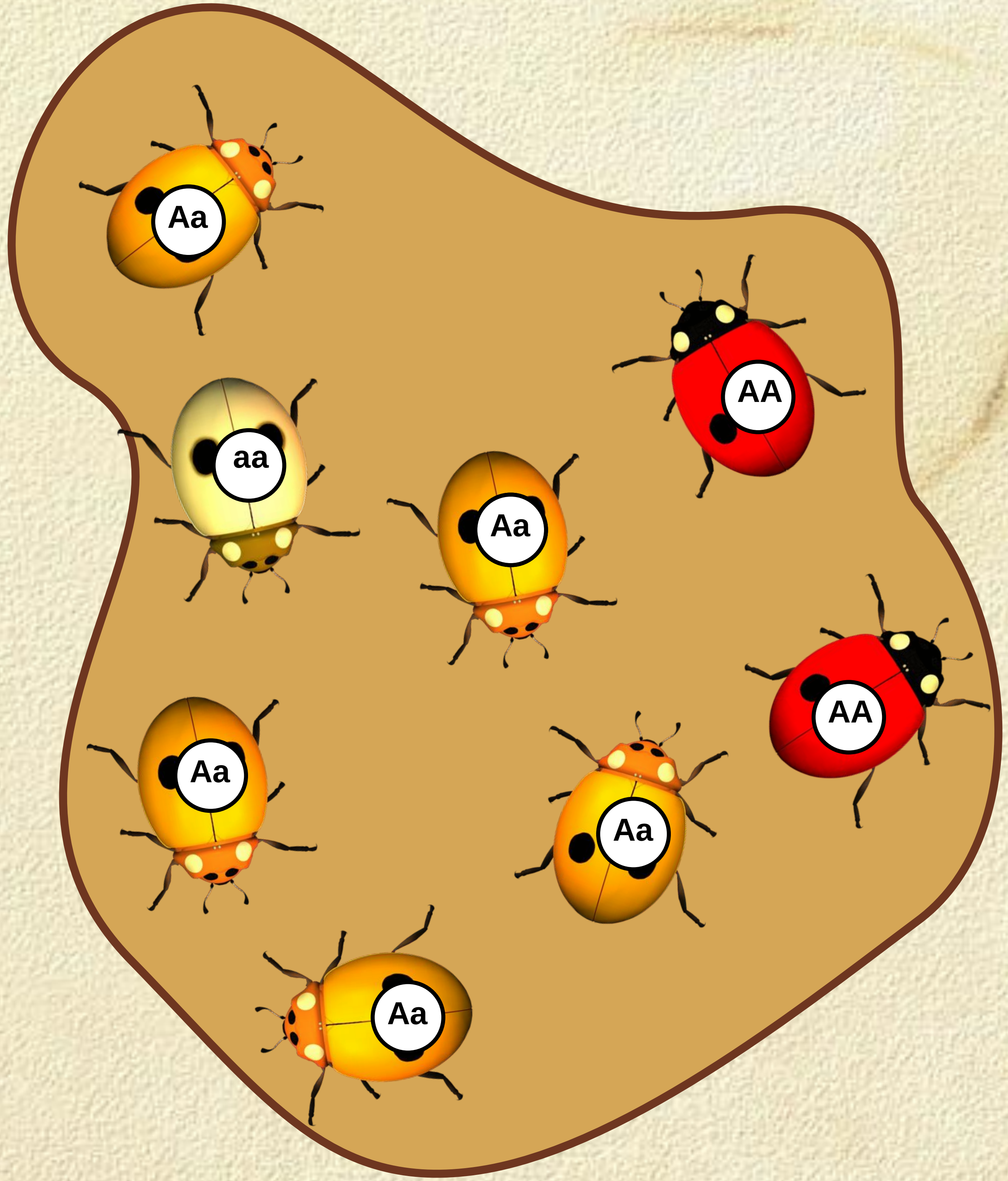
- To determine the frequencies of alleles in the population, count up the numbers of dominant and recessive alleles, regardless of the combinations in which they occur.
- Convert these to percentages by a simple equation:

$$\frac{\text{No. of dominant alleles}}{\text{Total no. of alleles}} \times 100$$



Determining Genotype Frequencies

- To determine the frequencies of different genotypes in the population, count up the actual number of each genotype in the population:
  - homozygous dominant (AA)
  - heterozygous (Aa)
  - homozygous recessive (aa).

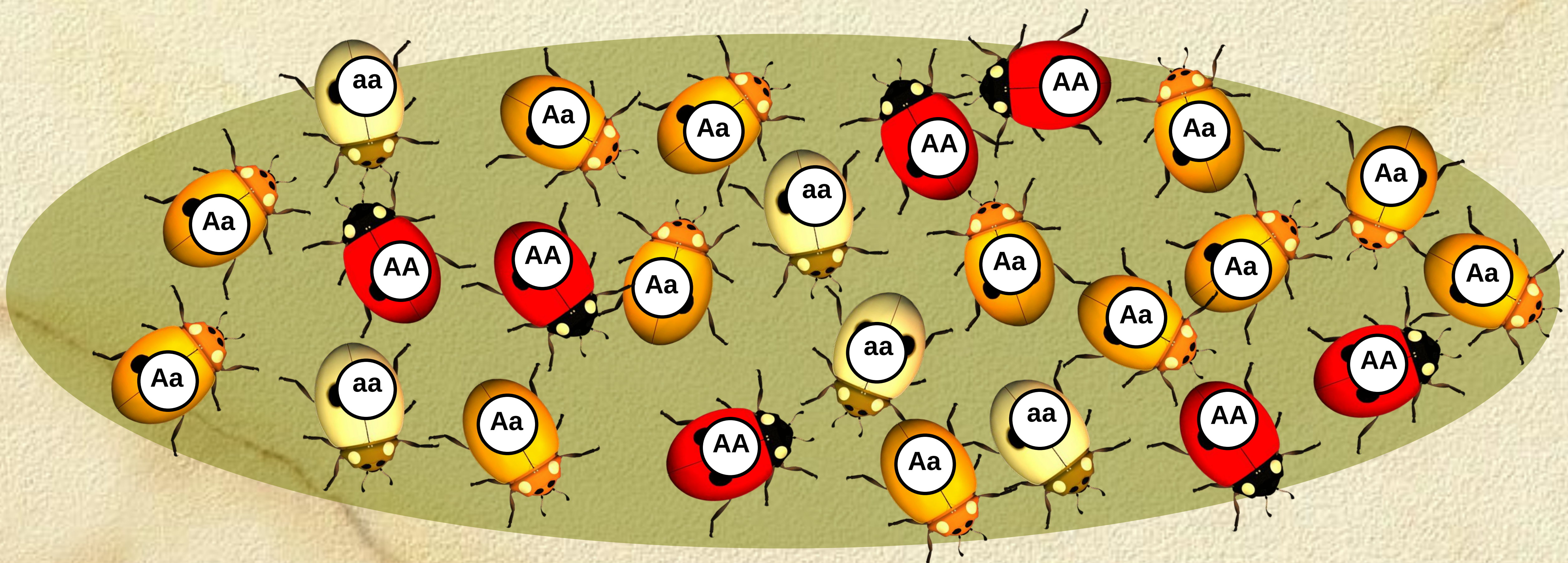


Changes in a Gene Pool 1

- **Phase 1: Initial gene pool**
  - In the gene pool below there are 25 individuals, each possessing two copies of a gene for a trait called A.
  - This is the gene pool before changes occur:

A	a	AA	Aa	aa
27	23	7	13	5
54	46	28	52	20

Allele types *Allele combinations*



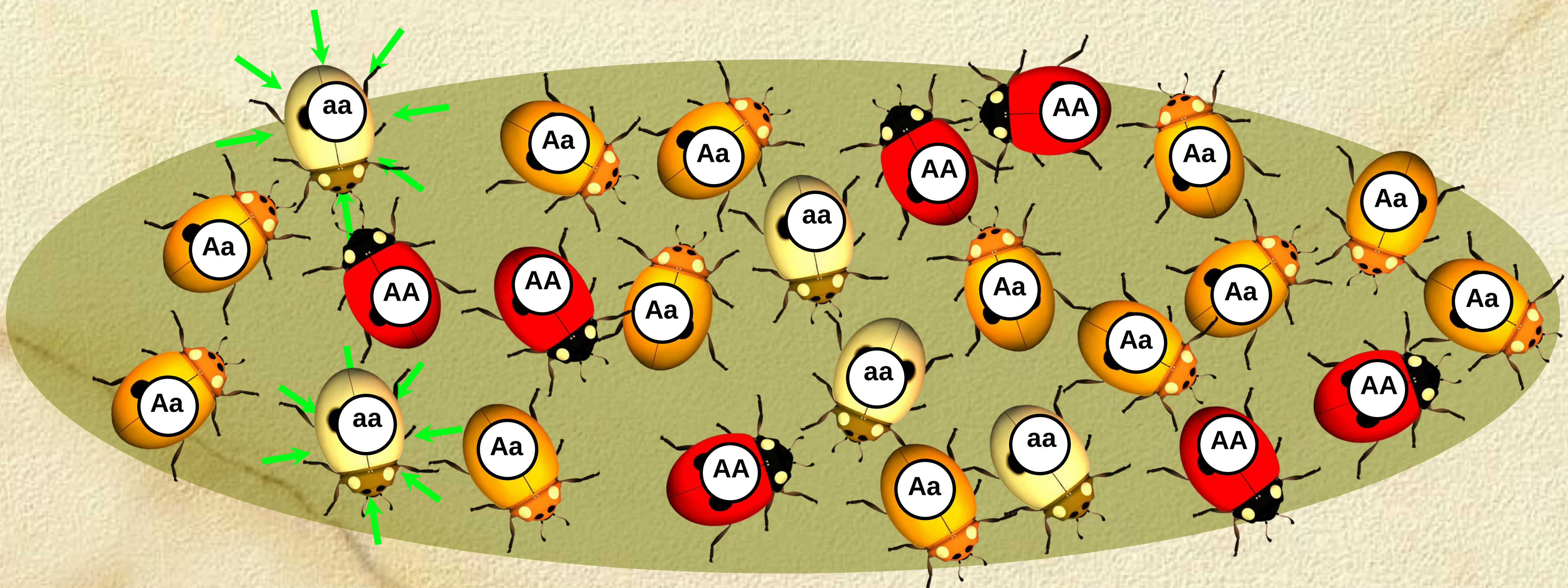
Changes in a Gene Pool 2

- **Phase 2: Natural selection**
  - In the same gene pool, at a later time, two pale individuals die due to the poor fitness of their phenotype.

Two pale individuals died and therefore their alleles are removed from the gene pool

	A	a	AA	Aa	aa
No.	27	19	7	13	3
%	58.7	41.3	30.4	56.5	13.0

Allele types (A, a) — *Allele combinations* (AA, Aa, aa)

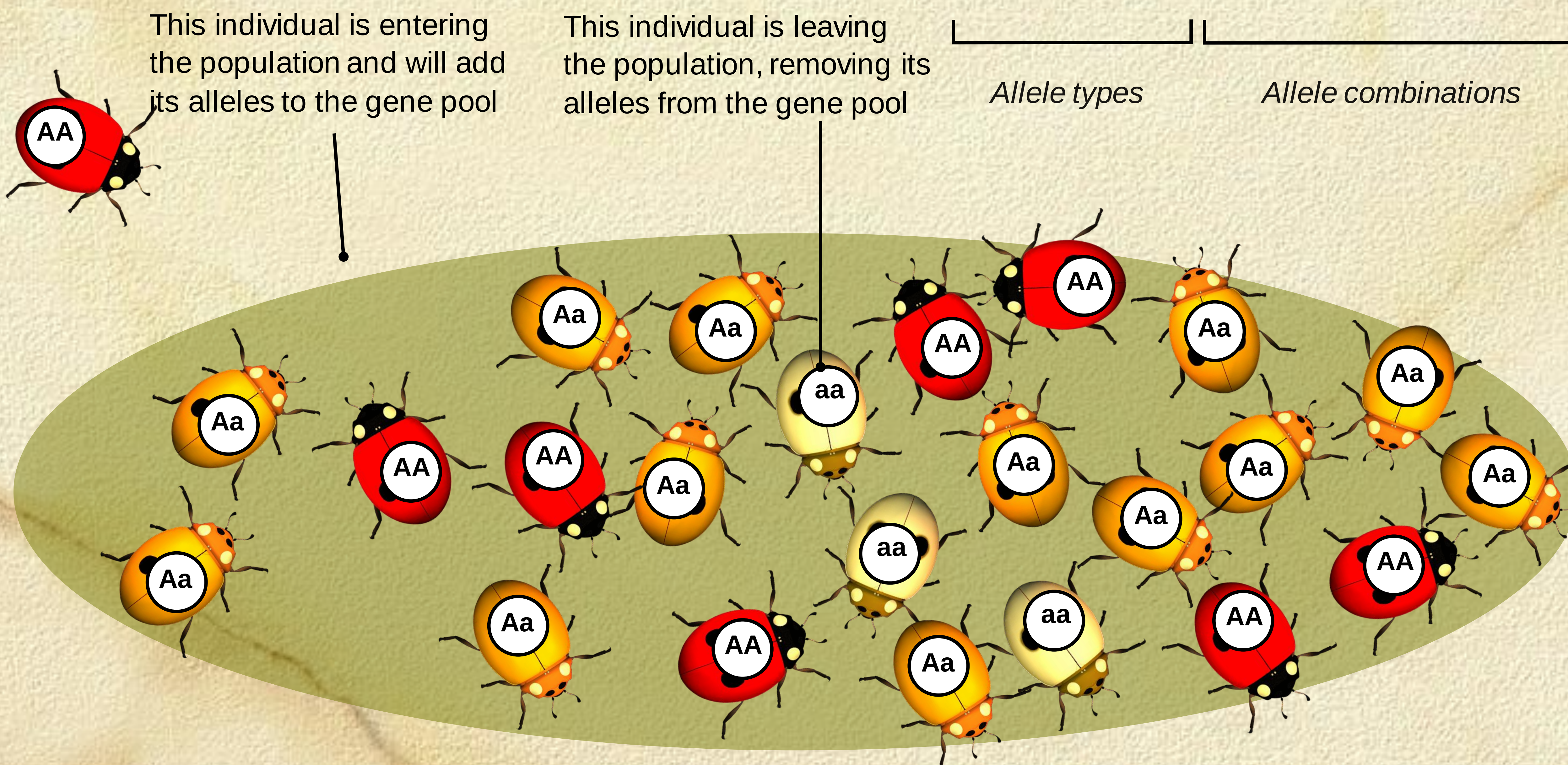


Changes in a Gene Pool 3

- **Phase 3: Immigration/Emigration**
  - Later still, one beetle (AA) joins the gene pool, while another (aa) leaves.

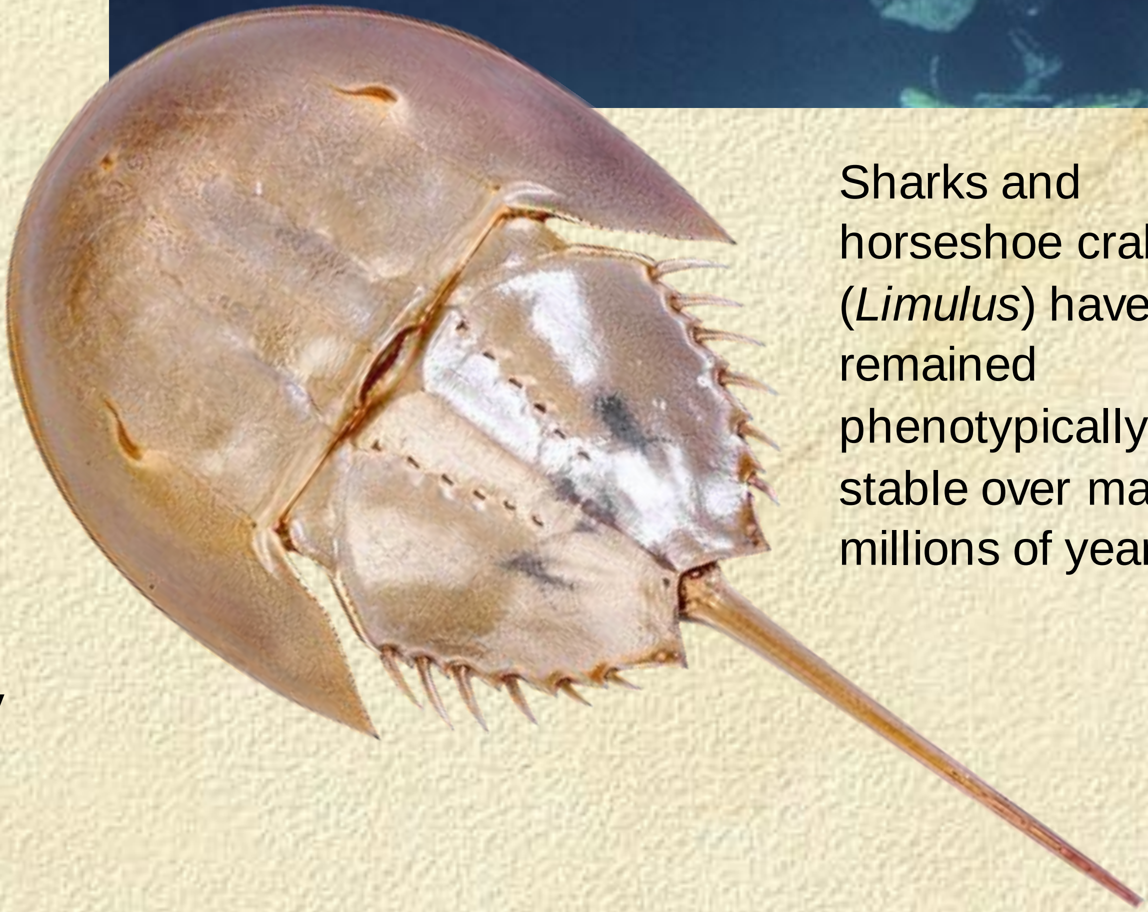
A	a	AA	Aa	aa
29	17	8	13	2
63	37	34.8	56.5	8.7

Allele types — *Allele combinations*



Hardy-Weinberg Equilibrium

- Populations that show no phenotypic change over many generations are said to be stable. This stability over time was described mathematically by two scientists:
  - G. Hardy: an English mathematician
  - W. Weinberg: a German physician
- The **Hardy-Weinberg law** describes the **genetic equilibrium** of large sexually reproducing populations.
  - The **frequencies of alleles** in a population will remain constant from one generation to the next unless acted on by outside forces.



Sharks and horseshoe crabs (*Limulus*) have remained phenotypically stable over many millions of years.

Conditions Required for Hardy-Weinberg Equilibrium

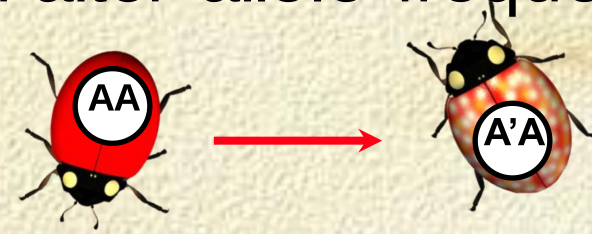
- The genetic equilibrium described by the Hardy-Weinberg law is only maintained in the absence of destabilizing events; all the **stabilizing** conditions described below must be met:

1	**Large population**: The population size is large.
2	**Random mating**: Every individual of reproductive age has an equal chance of finding a mate.
3	**No migration**: There is no movement of individuals into or out of the population (no gene flow).
4	**No selection pressure**: All genotypes have an equal chance of reproductive success.
5	**No mutation**: There are no mutations, which might create new alleles in the population.

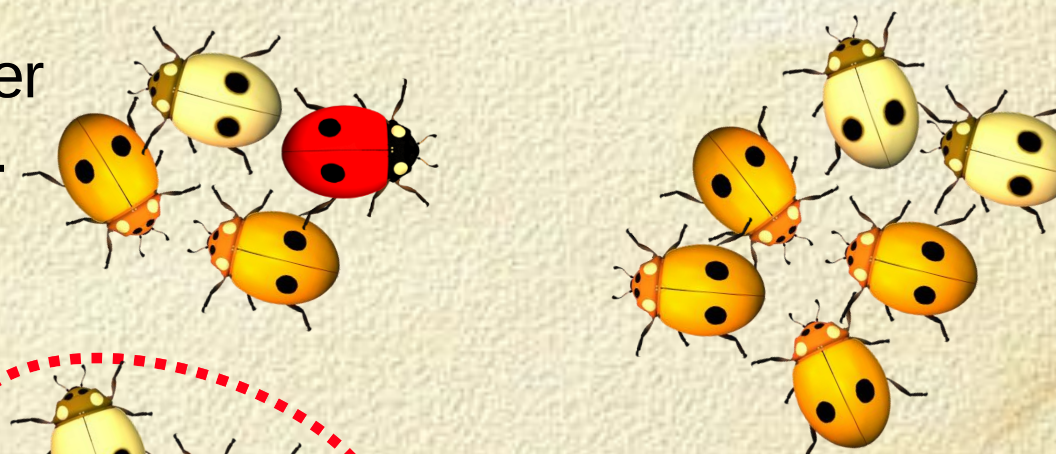
- Natural populations **seldom** meet all these requirements....
- .....therefore allele frequencies will change
- A change in the allele frequencies in a population is termed **microevolution**.

Changing Allele Frequencies

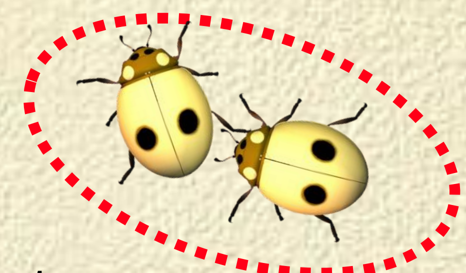
- Gene pools are subjected to processes that can alter allele frequencies:
  - **Mutation**: Spontaneous mutations can alter alleles frequencies and create new alleles.



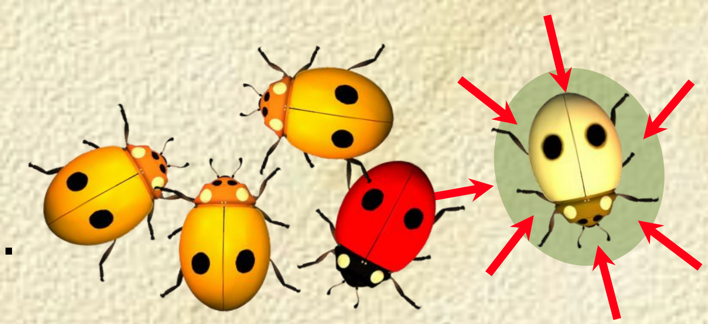
  - **Gene flow**: Genes can be exchanged with other gene pools as individuals move between them.



  - **Small population size and genetic drift**: In small populations, allele frequencies can change randomly from generation to generation; alleles may be lost or fixed.



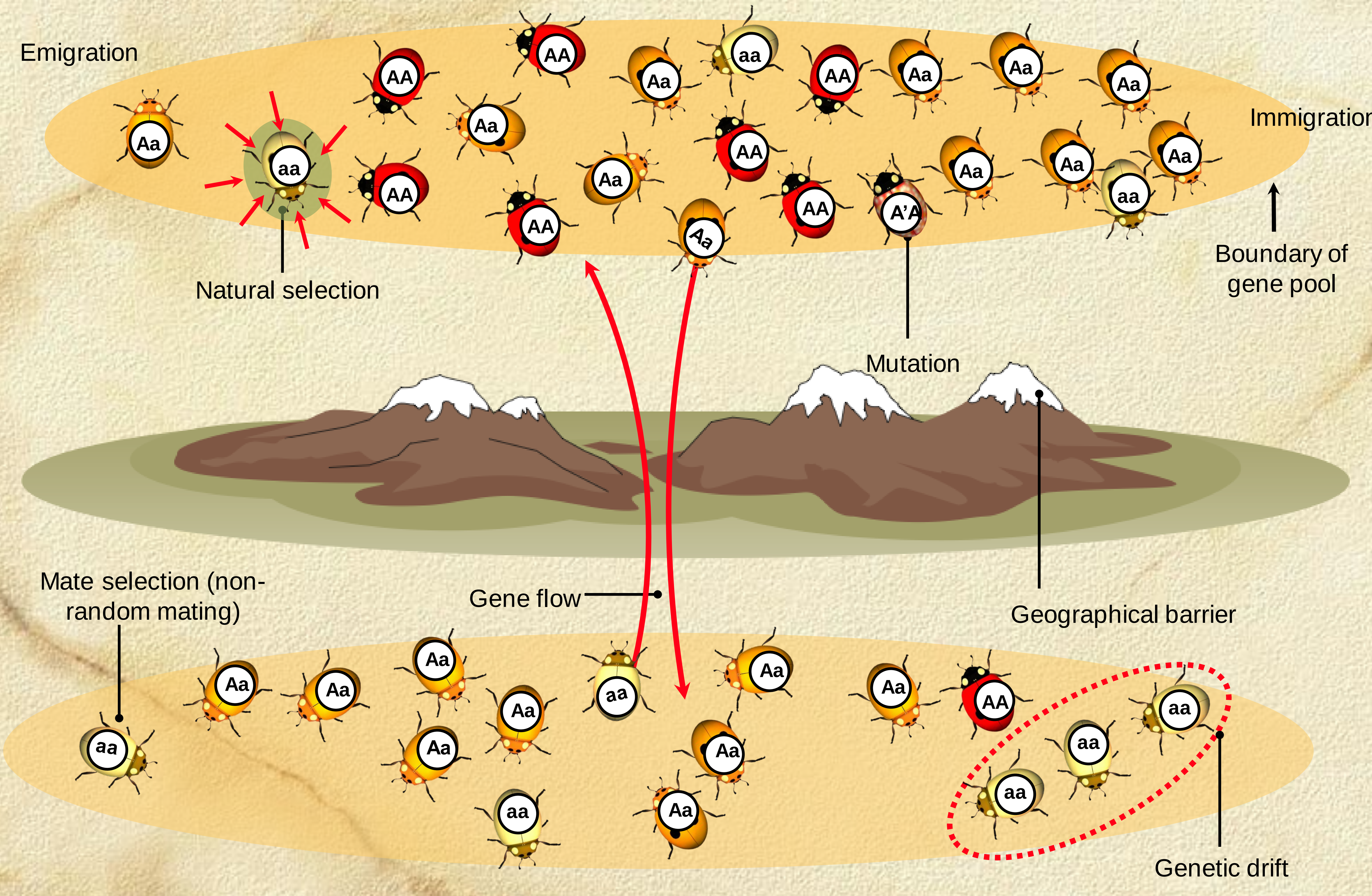
  - **Natural selection**: Selection pressure against certain alleles combinations may reduce reproductive success.



  - **Non-random mating**: Individuals seek out particular phenotypes with which to mate.

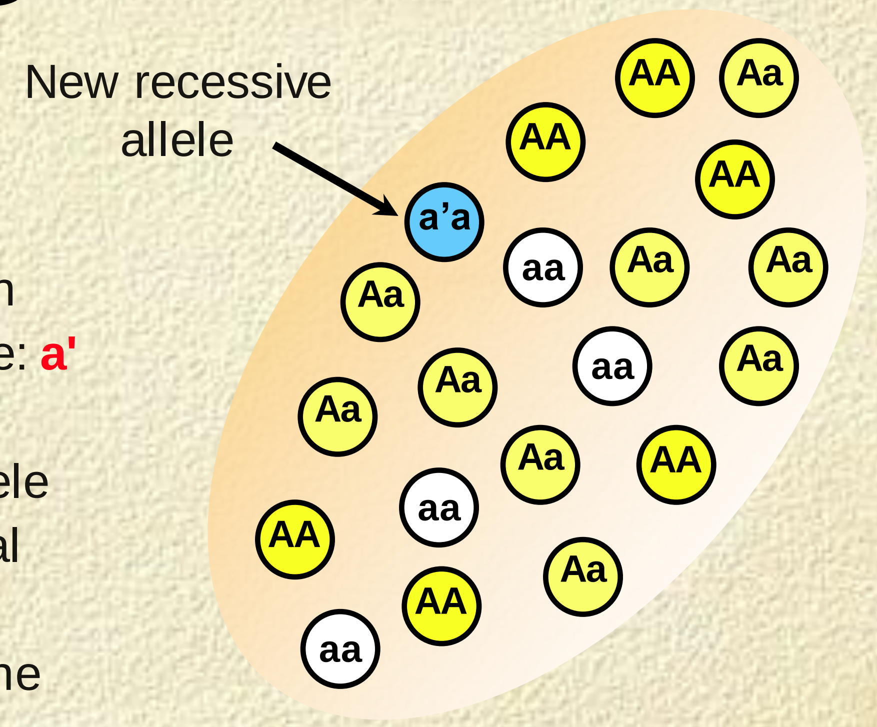
- Some of these processes cause random changes, others may be directional (i.e. they favor some alleles at the expense of others).

Changing Allele Frequencies



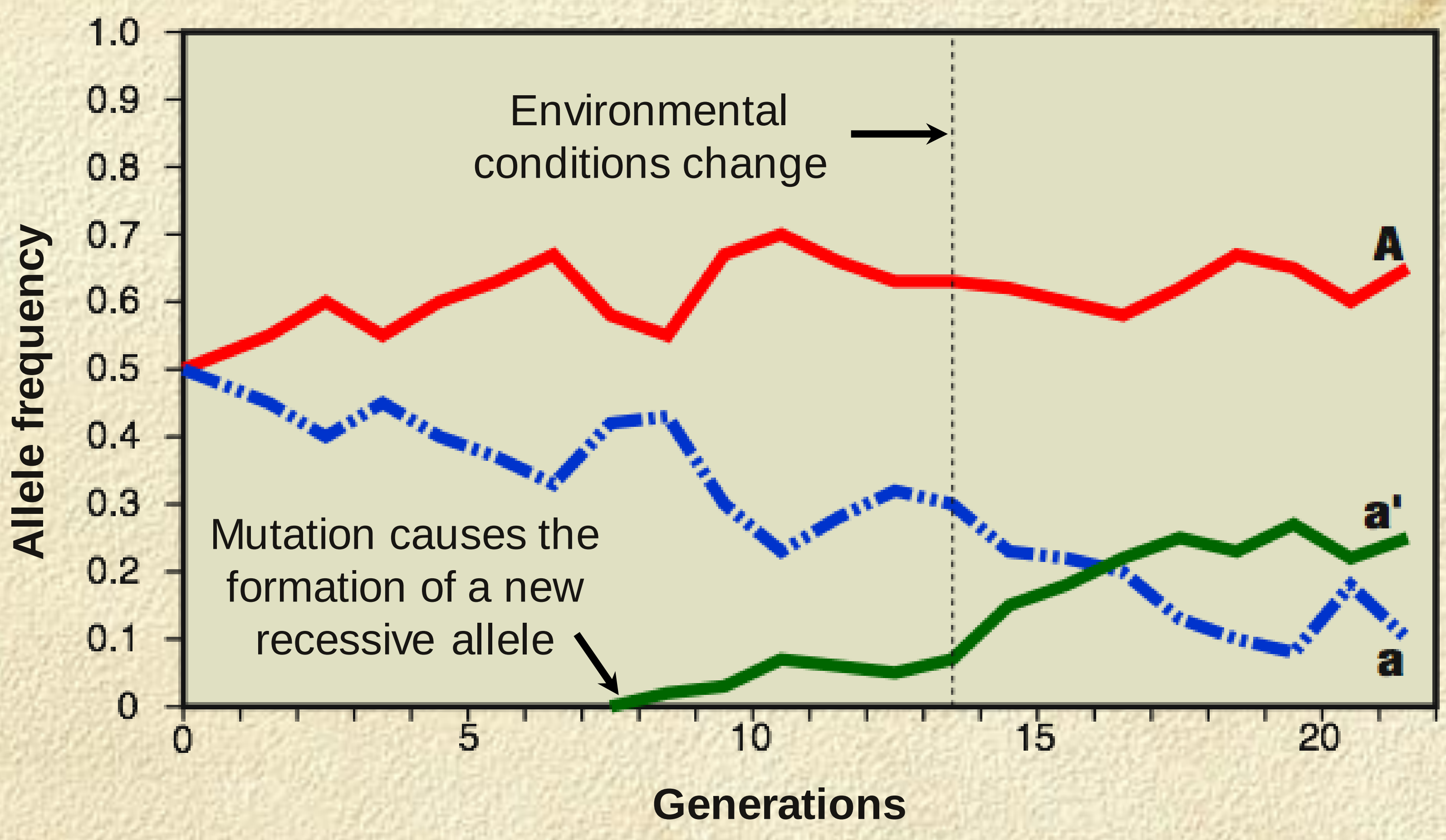
Mutations

- Mutations are the source of all new alleles.
- Mutations can therefore change the frequency of existing alleles by competing with them.
- Recurrent spontaneous mutations may become common in a population if they are not harmful and are not eliminated.



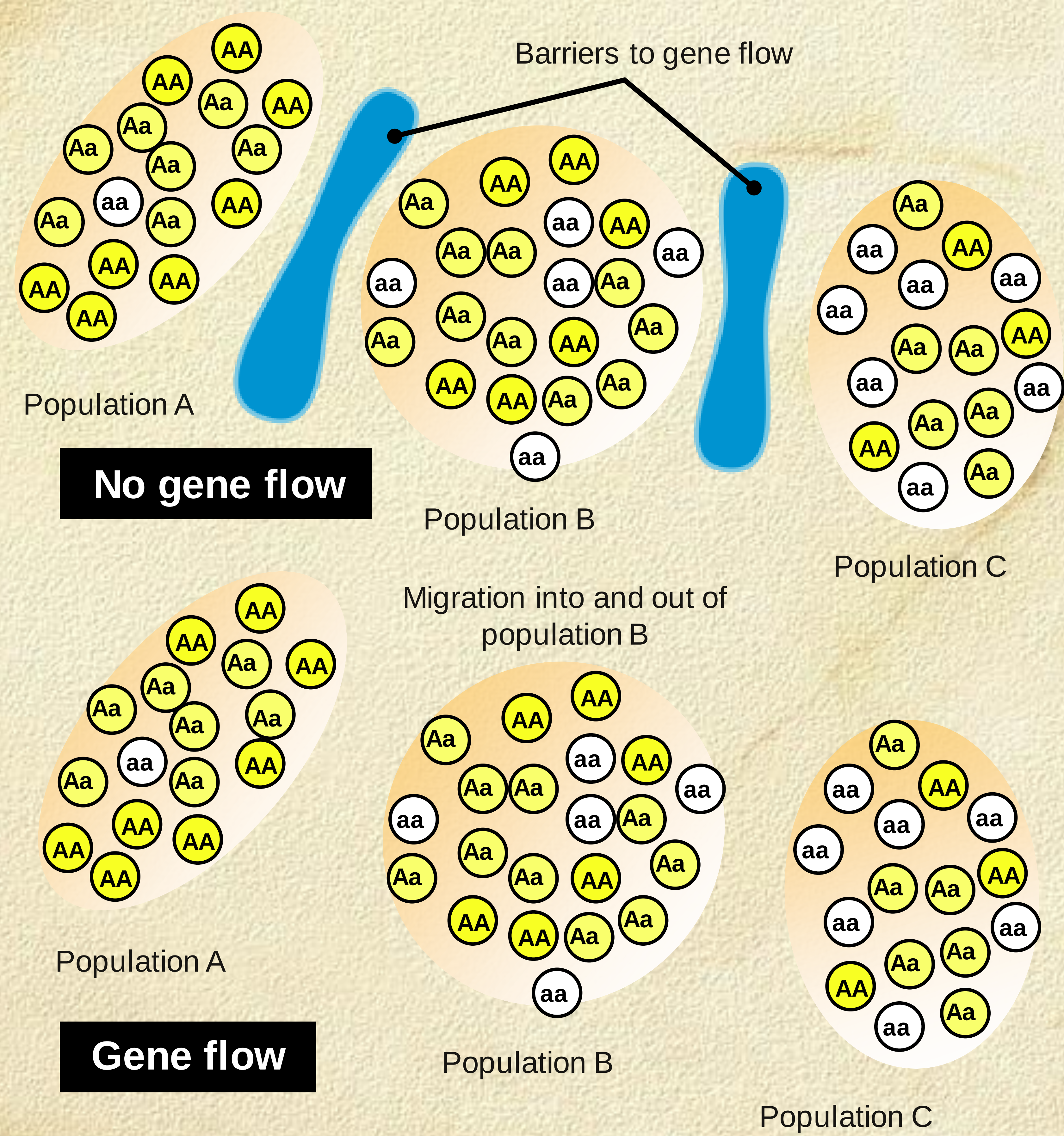
In the graph below, a mutation creates a new recessive allele: **a'**

The frequency of this new allele increases when environmental conditions change, giving it a competitive advantage over the other recessive allele: **a**



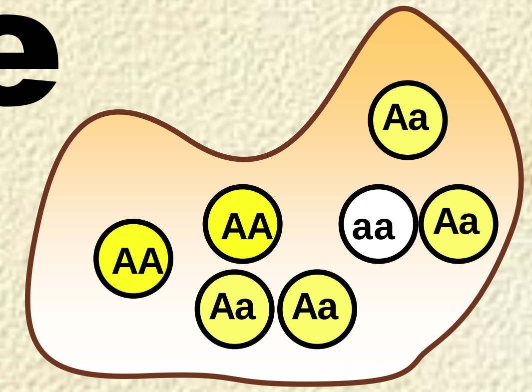
Gene Flow

- Gene flow is the movement of genes into or out of a population (immigration and emigration).
- A population may gain or lose alleles through gene flow.
- Gene flow tends to **reduce the differences between populations** because the gene pools become more similar.

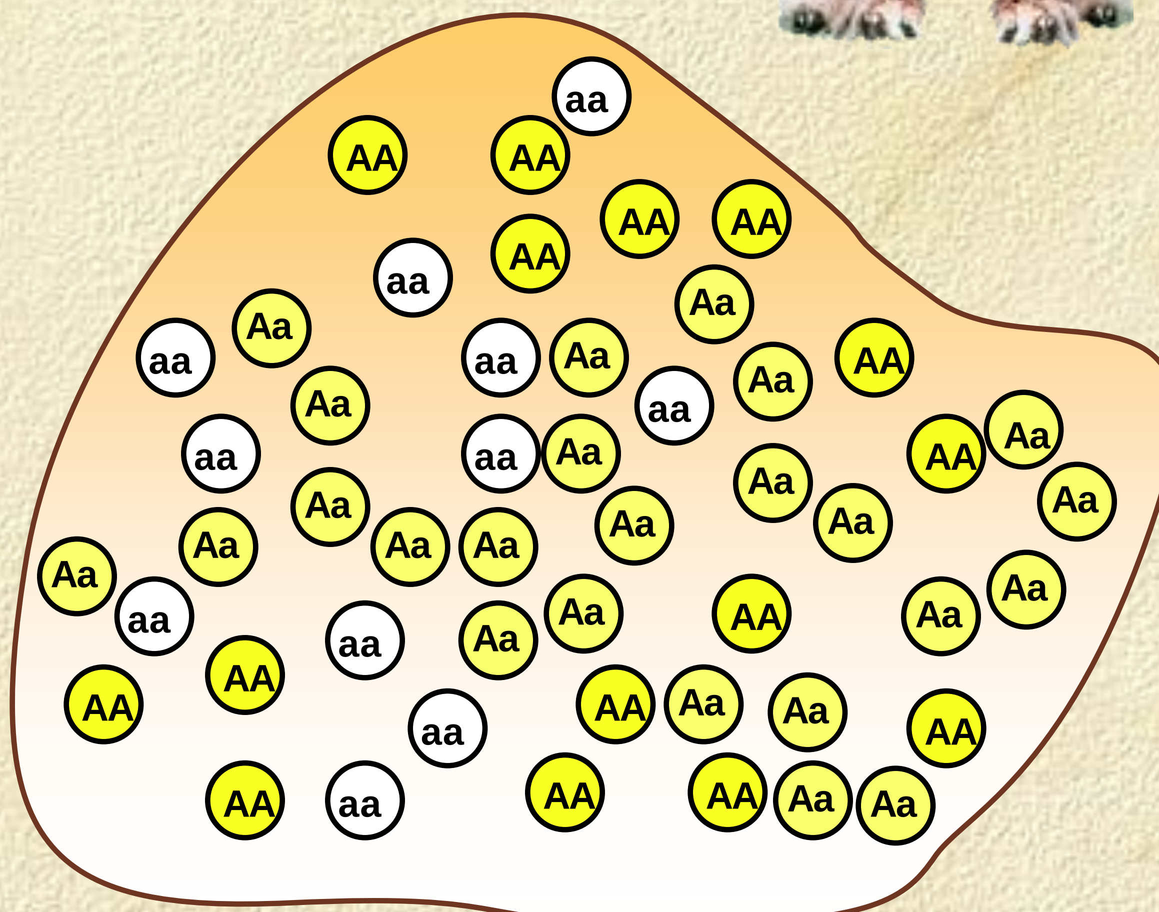


Allele Frequencies and Population Size

- The allele frequencies of large populations are more stable because there is a greater reservoir of variability and they are less affected by changes involving only a few individuals.
- Small populations have fewer alleles to begin with and so the severity and speed of changes in allele frequencies are greater.
- Endangered species with very low population numbers or restricted distributions may be subjected to severe and rapid allele changes.



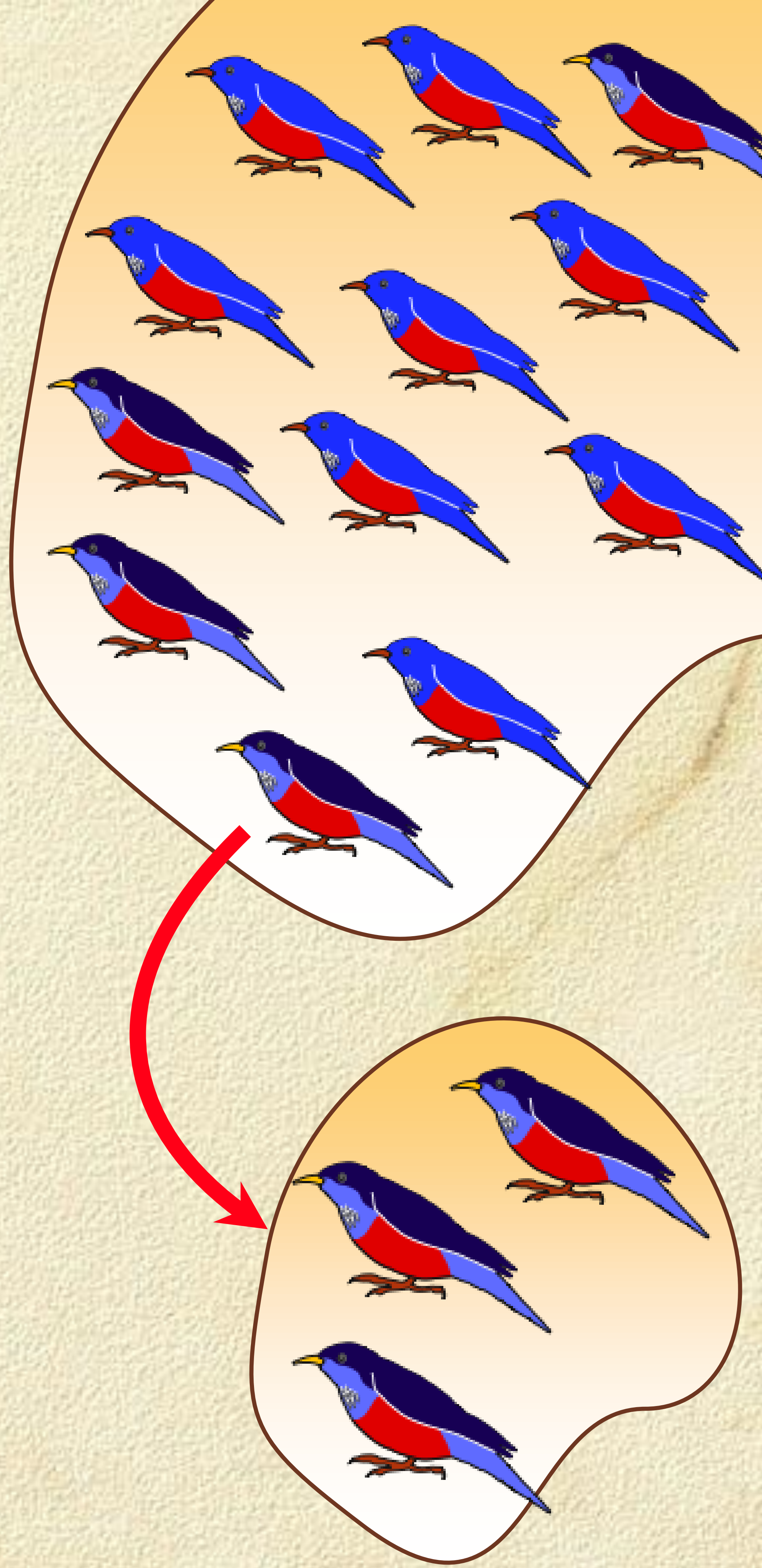
Small population
Cheetahs have a small population with very restricted genetic diversity



Large population

Genetic Drift

- For various reasons, not all individuals will be able to contribute their genes to the next generation. As a result, random changes occur in allele frequencies in all populations.
- These random changes are referred to as **genetic drift**. In small, inbreeding populations, genetic drift may have pronounced effects on allele frequencies. Alleles may become:
  - **Lost** from the gene pool (frequency = **0%**)
  - **Fixed** as the only allele present in the gene pool (frequency = **100%**)
- Genetic drift is often a feature of small populations that become isolated from the larger population gene pool, as with island colonizers (right).



Population Bottlenecks

- Populations may be reduced to low numbers through periods of:

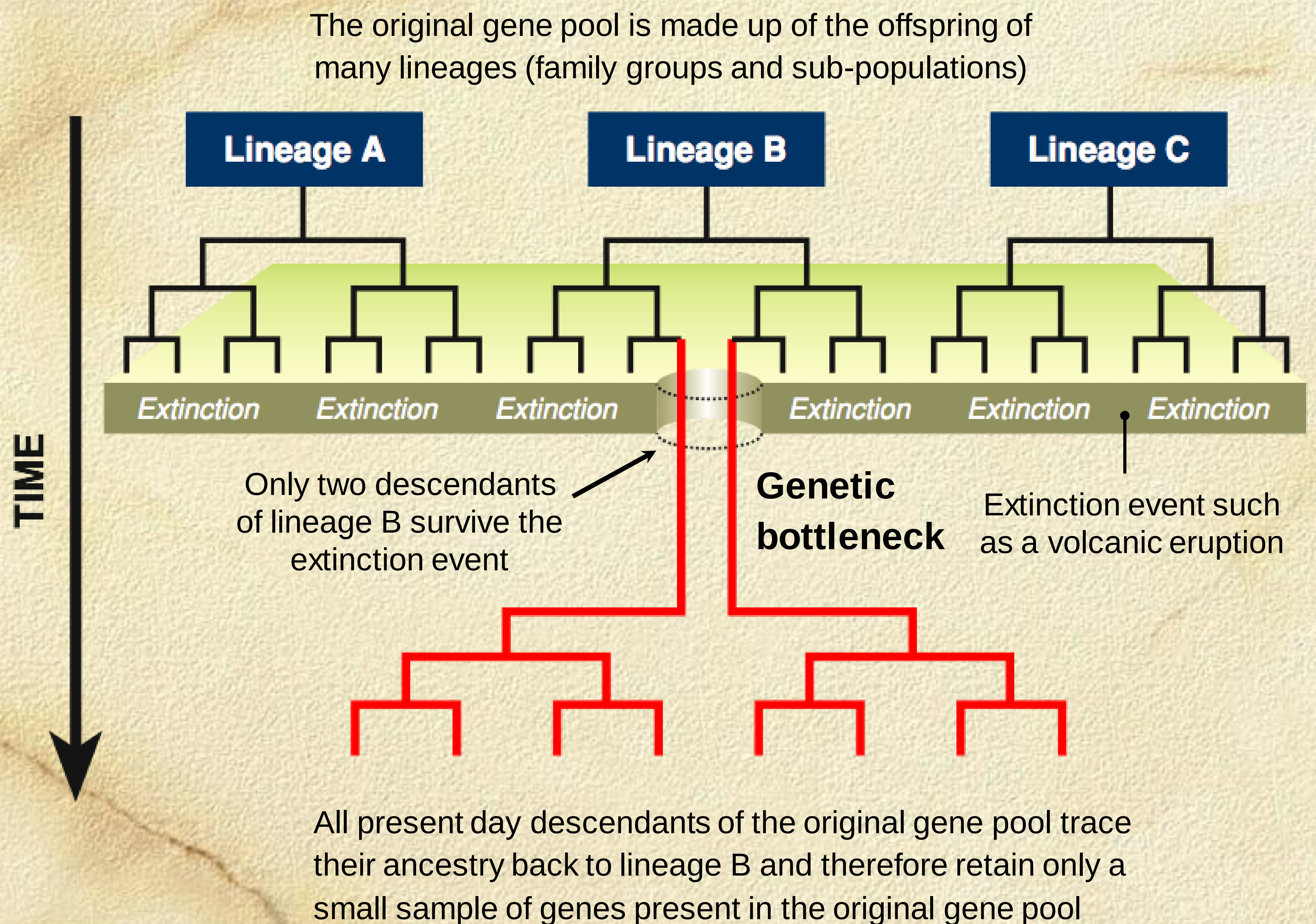
Seasonal climatic change

Heavy predation or disease

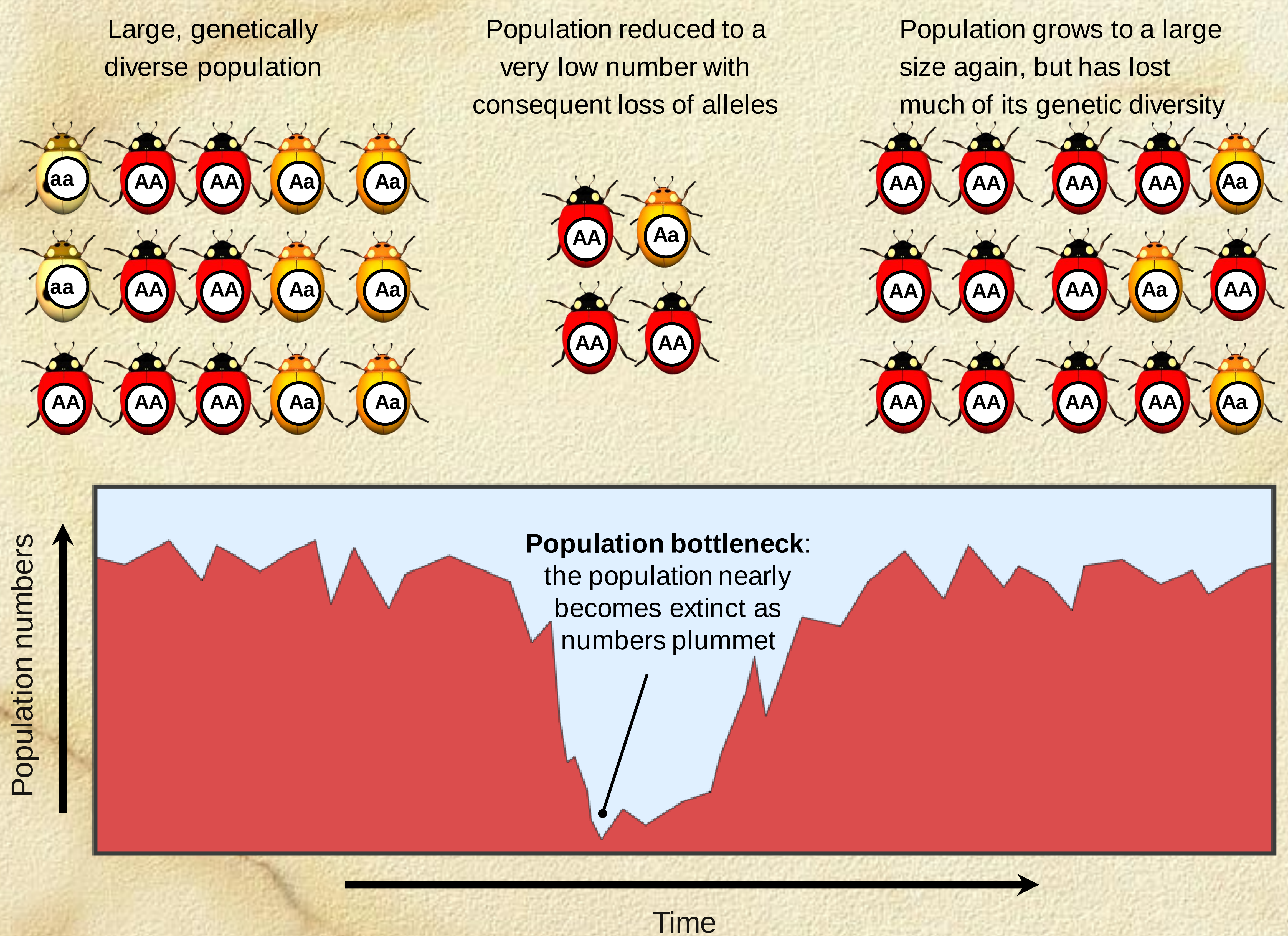
Catastrophic events (e.g. flood, volcanic eruptions, landslide)

- As a result, only a small number of individuals remain in the gene pool to contribute their genes to the next generation.
- The small sample that survives will often not be representative of the original, larger gene pool, and the resulting allele frequencies may be severely altered.
- In addition to this '**bottleneck' effect**, the small surviving population is often affected by inbreeding and genetic drift.

Population Bottlenecks



Population Bottlenecks



Genetic Bottlenecks & the Cheetah Population

- The world population of **cheetahs** has declined in recent years to fewer than **20 000**.
  - Recent genetic analyses has found that the total cheetah population has very little genetic diversity.
  - Cheetahs appear to have narrowly escaped extinction at the end of the last ice age: 10-20 000 years ago.
  - All modern cheetahs may have arisen from a single surviving litter, accounting for the lack of diversity.
  - At this time, 75% of all large mammals perished (including mammoths, cave bears, and saber-toothed cats).

Genetic Diversity in Cheetahs

- The lack of genetic variation has led to:
  - sperm abnormalities
  - decreased fecundity
  - high cub mortality
  - sensitivity to disease
- Since the **genetic bottleneck**, there has been insufficient time for random mutations to produce new genetic variation.

The Founder Effect

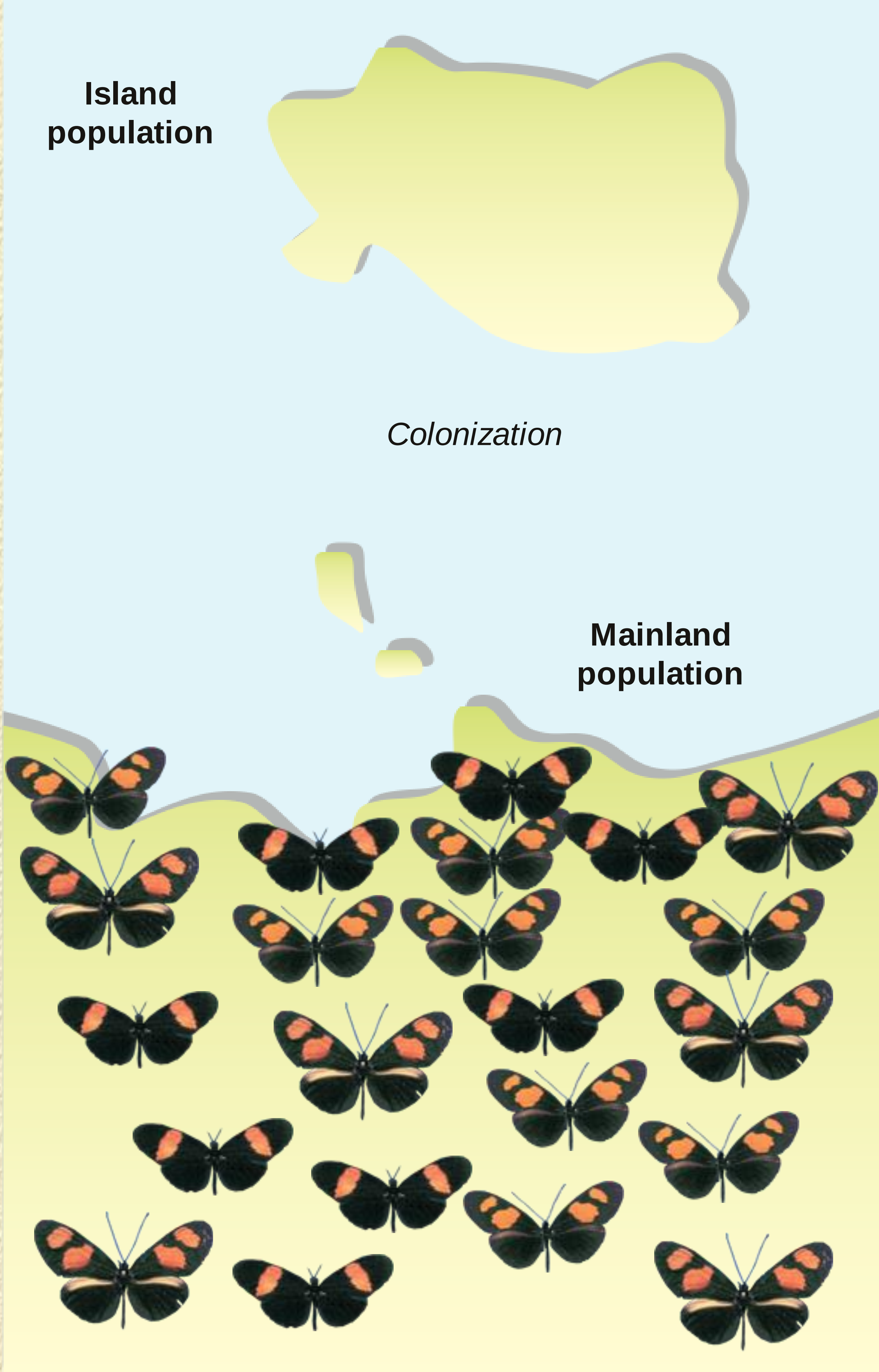
- Occasionally, a small number of individuals may migrate away or become **isolated** from their original population.
- This **colonizing** or **founder** population will have a small and probably non-representative sample of alleles from the parent population's gene pool.
- As a consequence of this **founder effect**, the colonizing population may **evolve** in a different direction than the parent population.

Offshore islands can provide an environment in which founder populations can evolve in isolation from the parental population.

The marine iguana of the Galapagos has evolved in an isolated island habitat

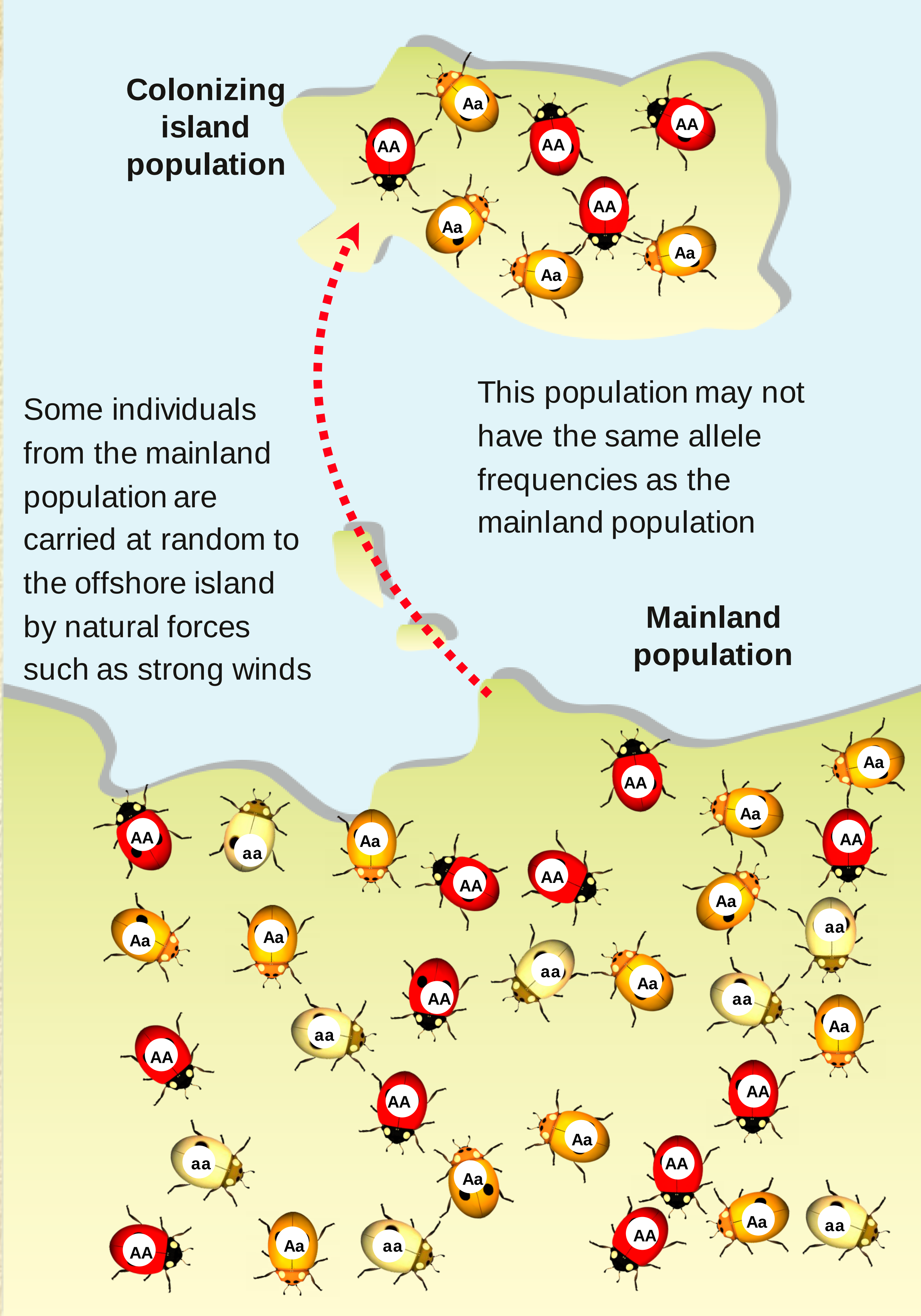
The Founder Effect

- Small founder populations are subject to the effects of **random genetic drift**.
- The founder effect is typically seen in the populations of islands which are colonized by individuals from mainland populations.
- Often these species have low or limited mobility; their dispersal is often dependent on prevailing winds (e.g. butterflies and other insects, reptiles, and small birds).



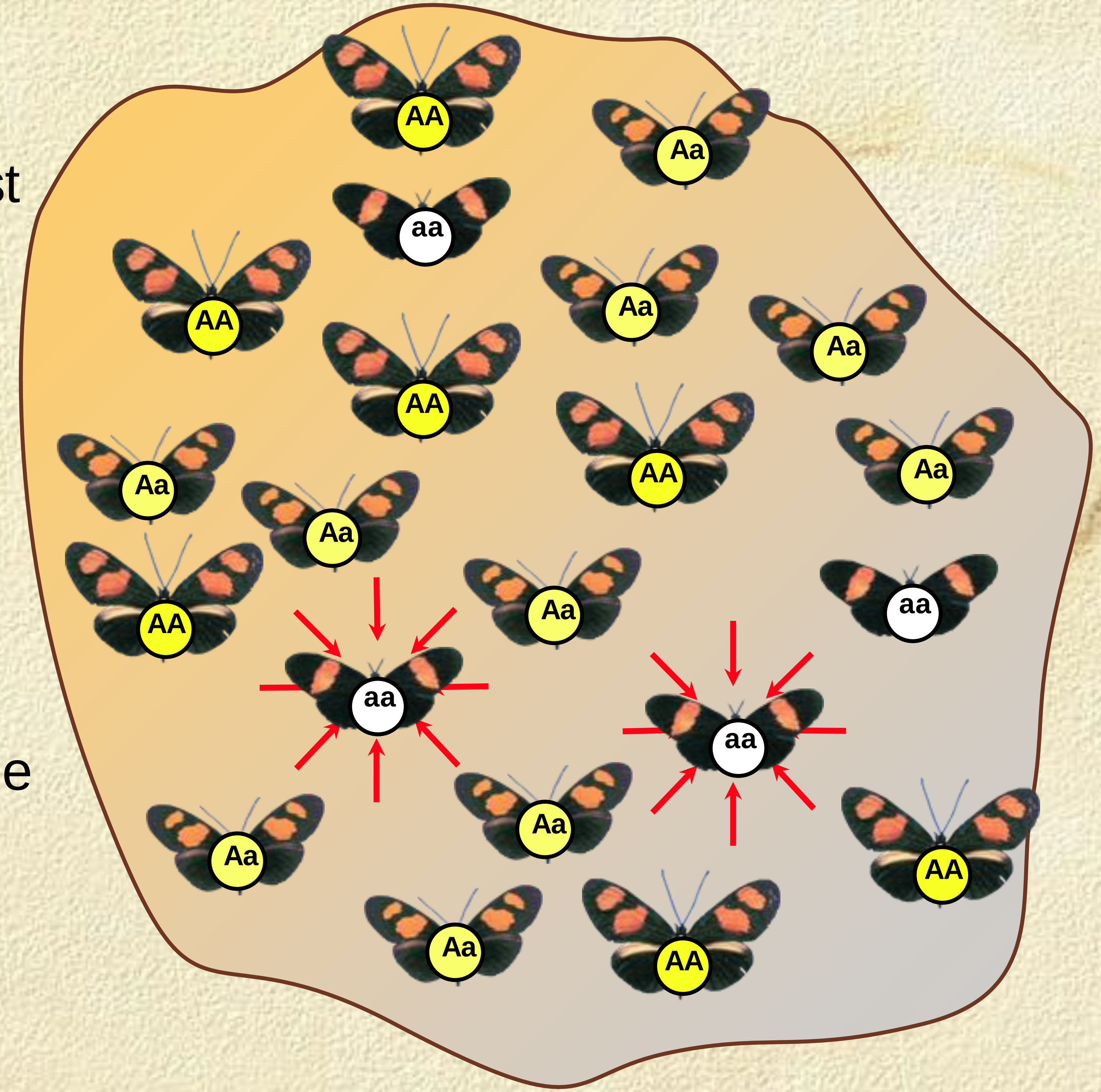
The Founder Effect

- In this hypothetical population of beetles, a small, randomly selected group is blown offshore to a neighboring island where they establish a breeding population.



Natural Selection

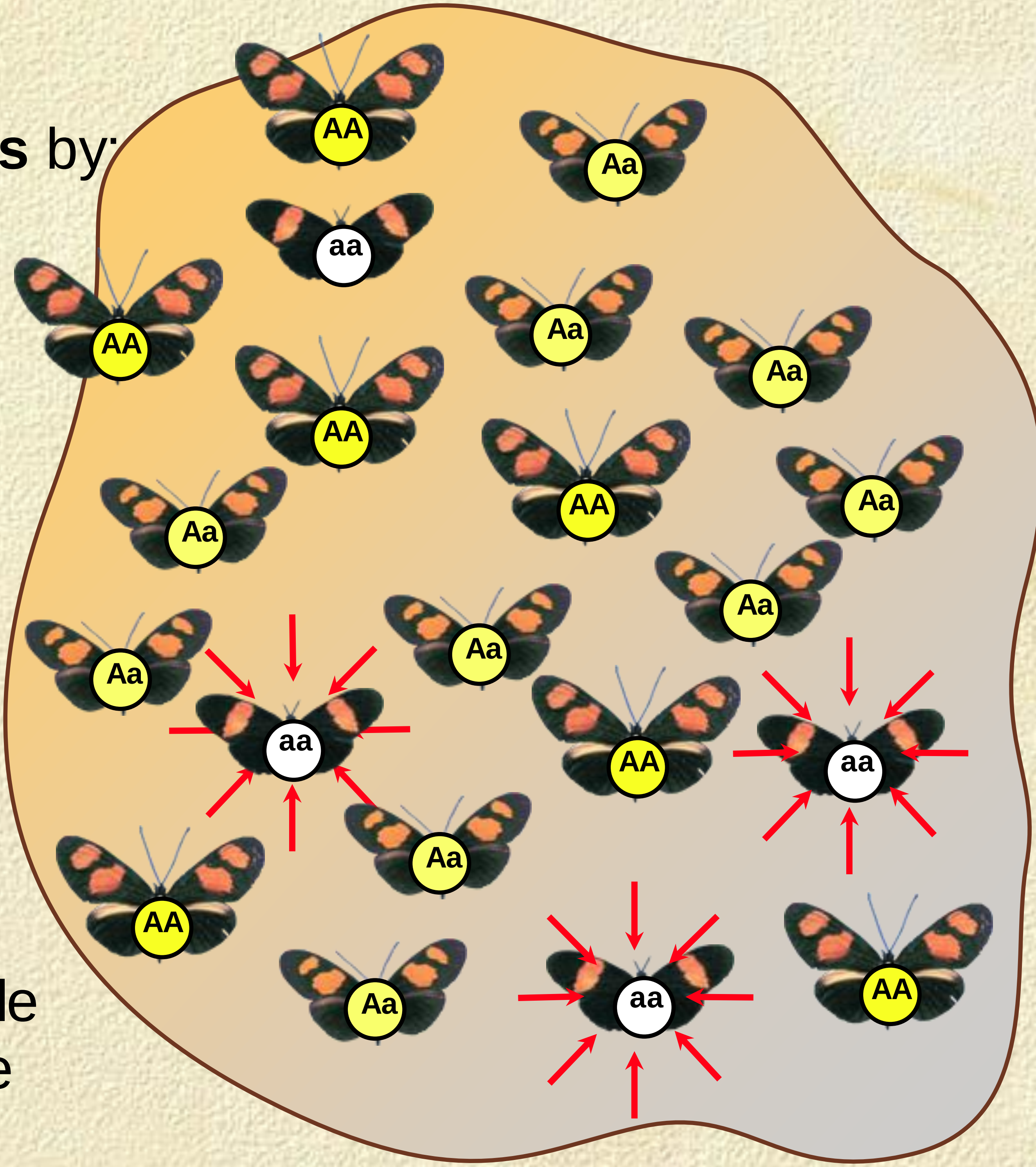
- Populations of sexually reproducing organisms consist of **varied individuals**, with some variants leaving more offspring than others.
- This differential success in reproduction (differential fitness) is called **natural selection**.
- Natural selection is responsible for most evolutionary change by selectively altering genetic variation through differential survival and reproduction.



Selection pressures may reduce the frequencies of certain alleles

Natural Selection

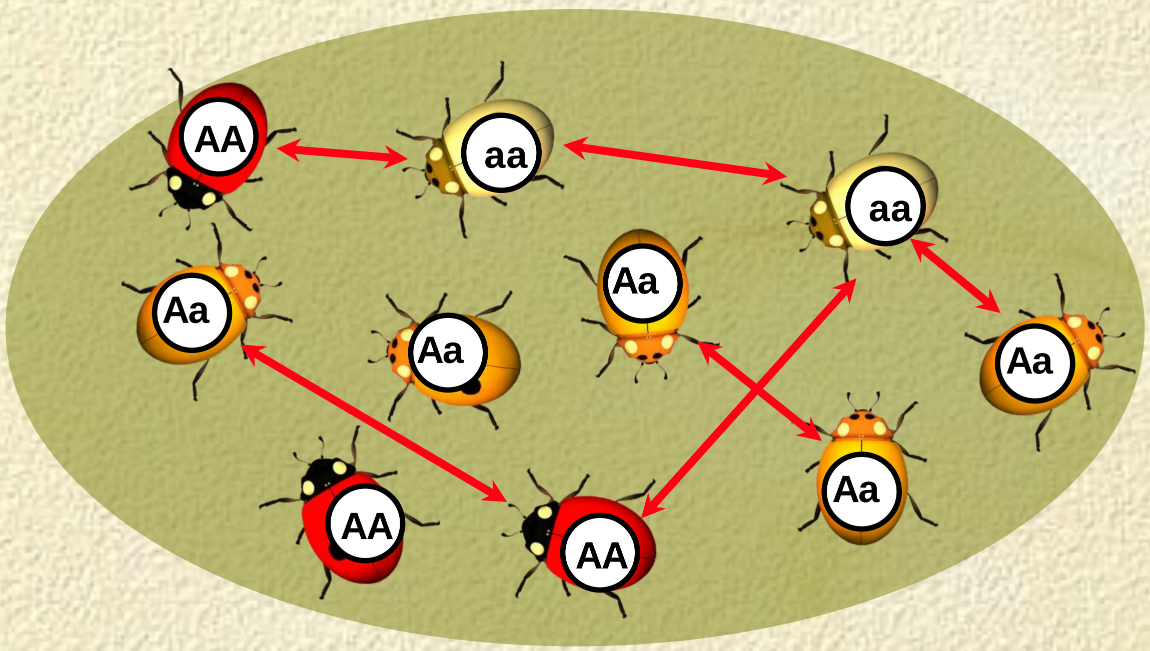
- Natural selection acts on **phenotypes** by:
  - Reducing the **reproductive success** of phenotypes poorly suited to the prevailing conditions; their alleles become less common in the gene pool.
  - Enhancing the survival and **reproductive success** of phenotypes well suited to the prevailing conditions; their alleles become more common in the gene pool.
- Natural selection therefore changes the composition of a gene pool and increases the probability that favorable alleles will come together in the same individual.



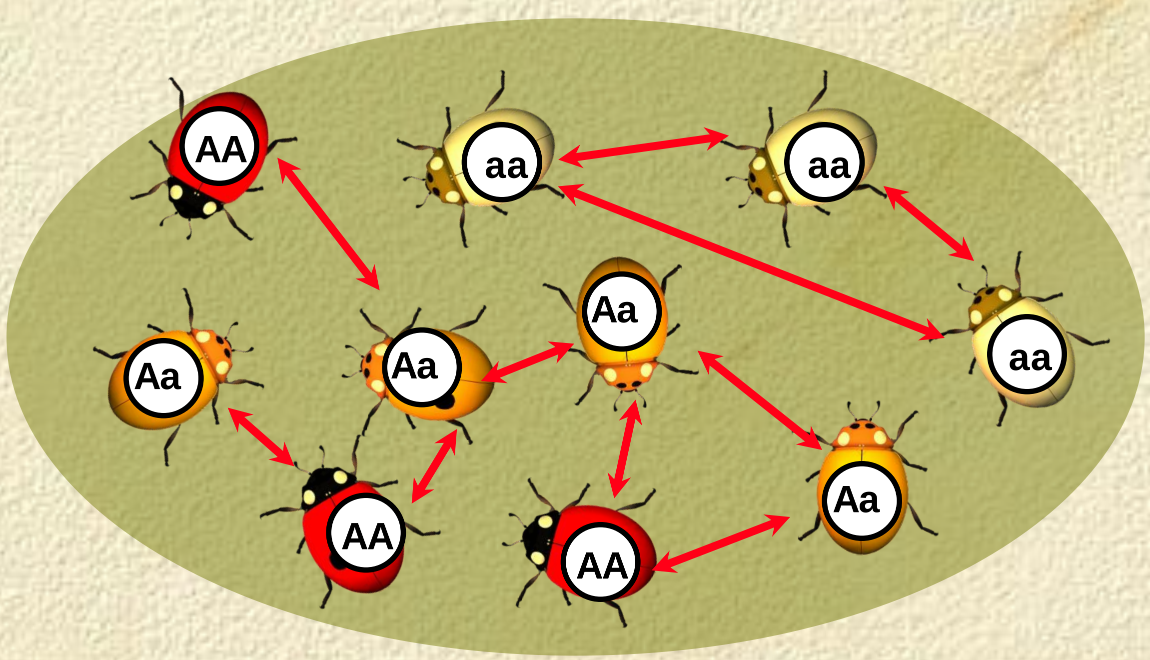
Phenotype with allele combination aa is more vulnerable to frost.

Assortative Mating

- Random (non-assortative) mating allows all genotypes to have an equal chance of reproductive success.
- Individuals may deviate from this by:
  - Mating more often with close neighbors than with distant members of the population.
  - Choosing mates that are most like themselves.
  - Such behavior is call **assortative mating**.



Random mating



Assortative mating

Assortative Mating

- The most extreme case of assortative mating is **self-fertilization** in some plant species, e.g. violets.
- Sometimes individuals may show random mating for some alleles but not others.
  - Humans exhibit assortative mating for racial features, but mate randomly for blood types.
- While assortative mating does not change the frequency of alleles in the overall gene pool, it does cause the ratio of genotypes to depart from that of random mating.

Violets can produce seed by both cross pollination or self pollination. Unlike cross-pollinated flowers (above), self-pollinated flowers are closed and are found near the soil surface.

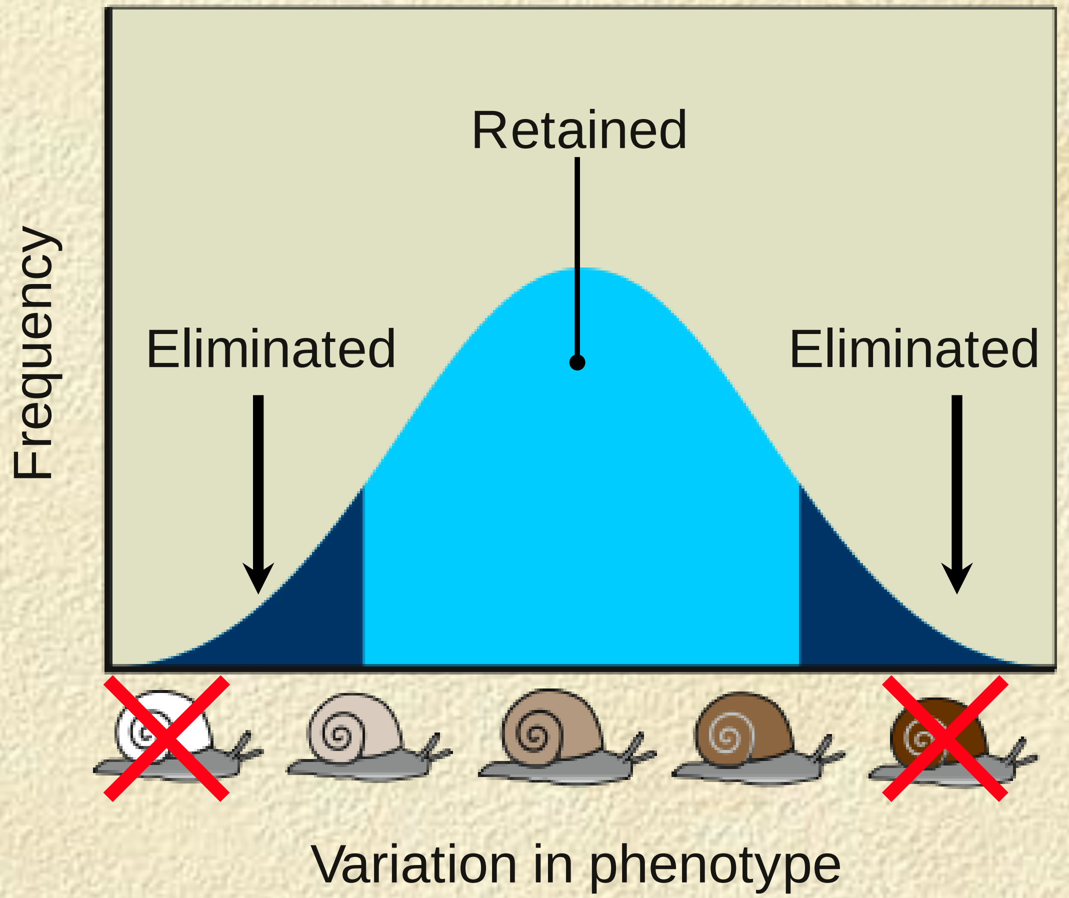
Humans often find partners within their racial group.

Modes of Natural Selection

- **Natural selection** changes allele frequencies in populations, but it does not produce the “perfect organism”.
- Rather than developing new phenotypes, it reduces the frequency of phenotypes that are less suited to the prevailing conditions (e.g. increased frequecy of heavy frosts).
- Traits (e.g. skin color, height) that are under **polygenic** control show **quantitative variation** in the phenotype. Natural selection acts on this variation.

Stabilizing Selection

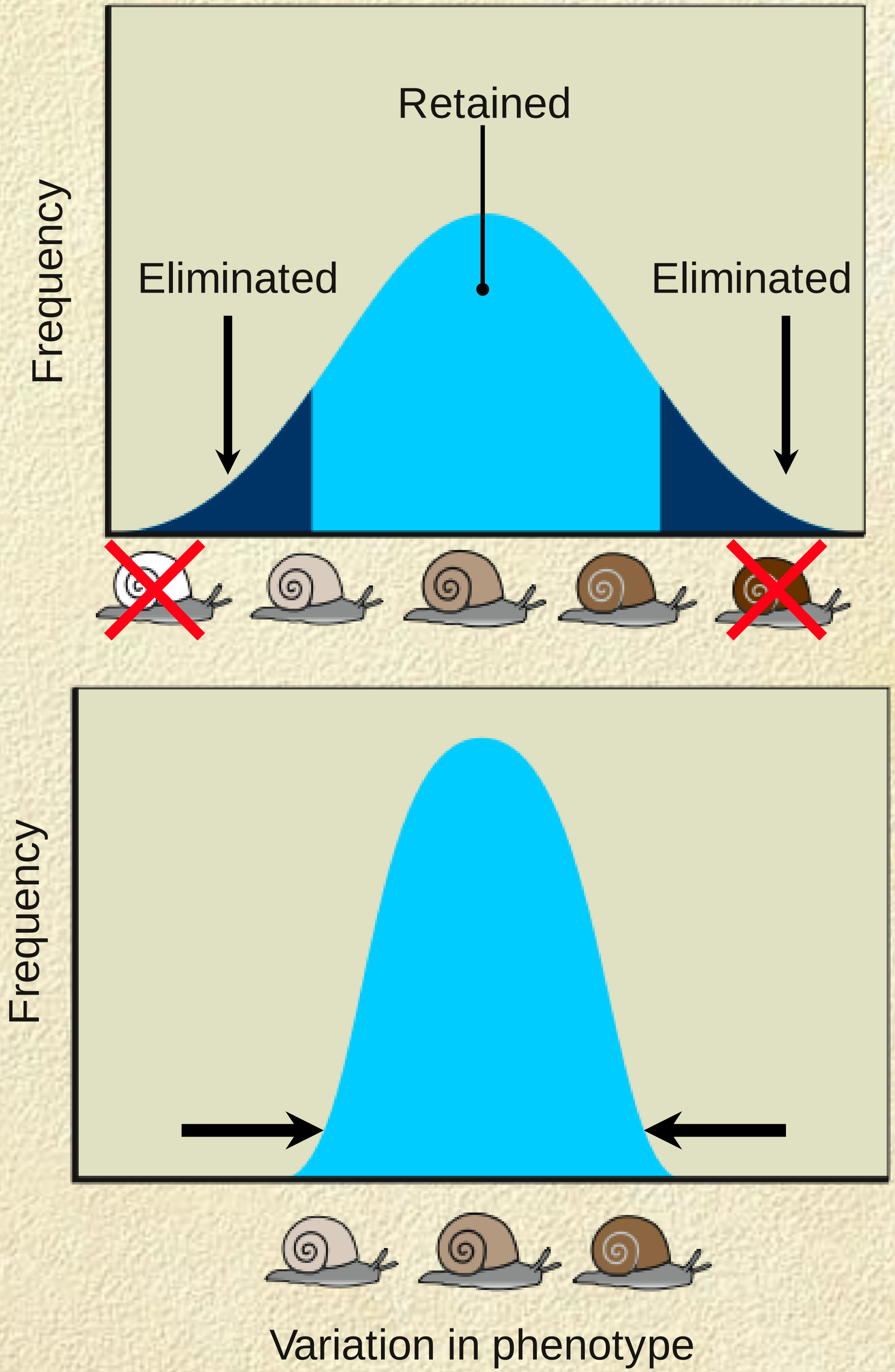
- Natural selection may produce phenotypic change over time. The **direction** of change will depend on the nature of the selection pressure.
- **Stabilizing selection** is probably the most common trend in natural populations; it favors the most common phenotype as the best adapted.
  - Stabilizing selection reduces variation by selecting against the extremes at each end of the phenotypic range.
  - The resulting bell shaped curve is narrower, about the same mean.
  - **EXAMPLE**: Human birth weights (right) are maintained in the 3-4 kg range by selection pressures at the extremes.



Stabilizing Selection

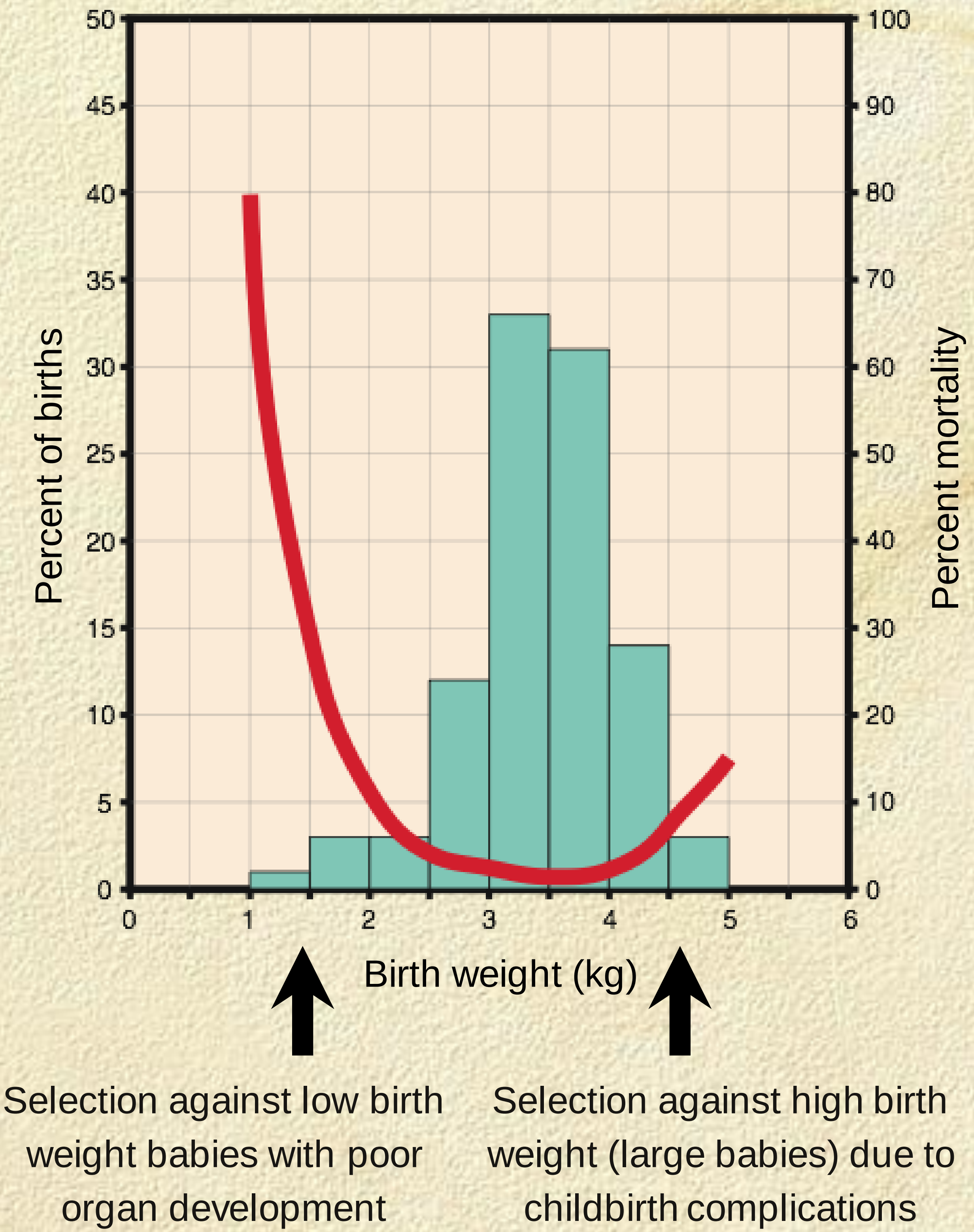
- Before selection there is a broad range of variation in the population:

- After selection, and for some generations later, there is a reduction in the amount of variation.



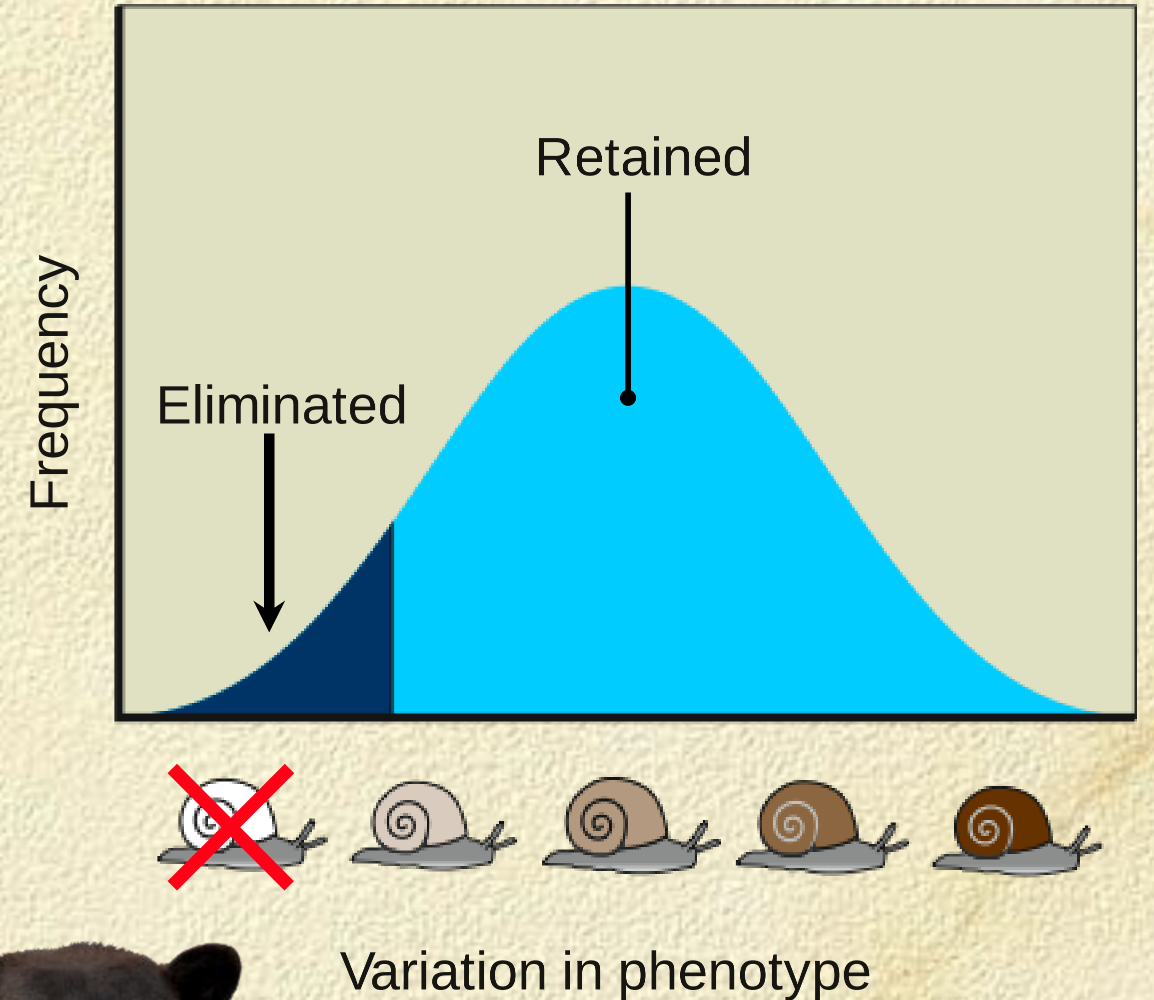
Stabilizing Selection in Human Birth Weights

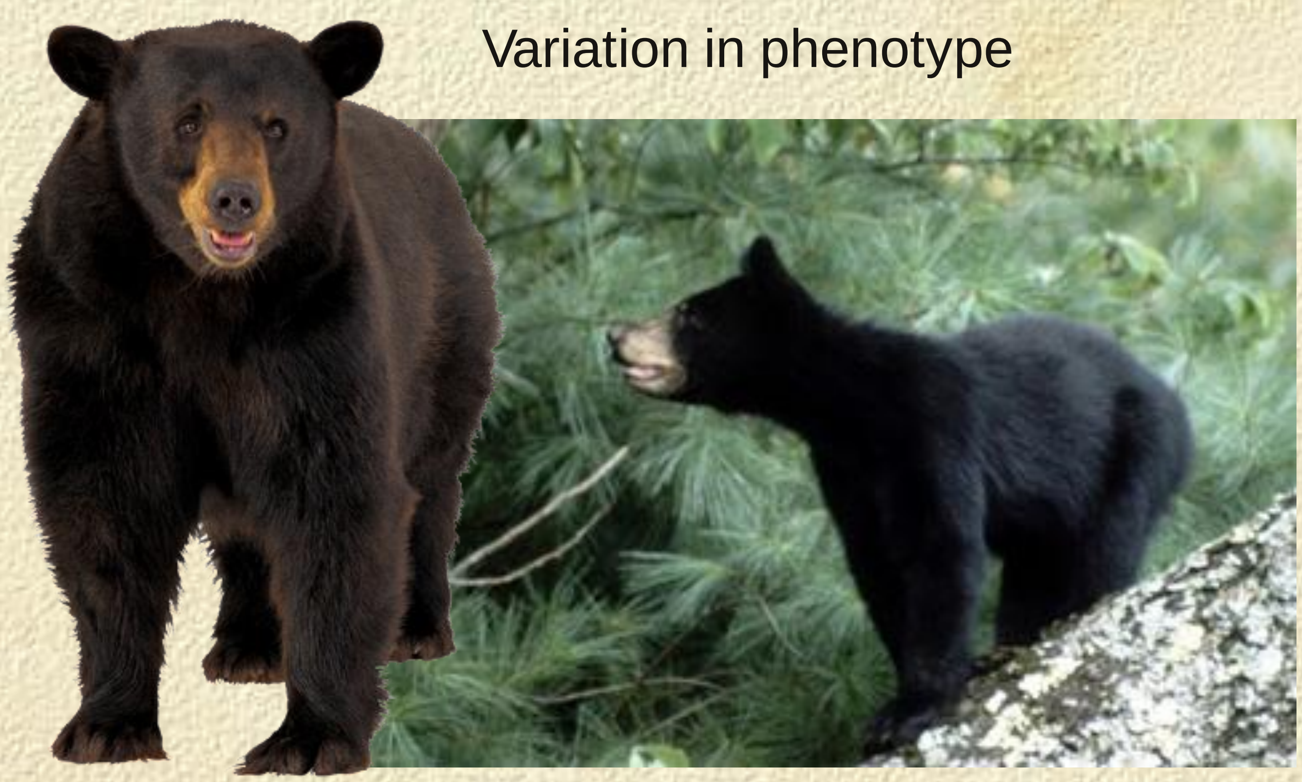
- **Stabilizing selection** against extremes in the birth weight range results in most births being between 3-4 kg.
- The histogram shows the percentage of births in each weight class. The red line shows the associated percentage mortality.
- Modern medical intervention is **reducing** this selection pressure by increasing the survival.



Directional Selection 1

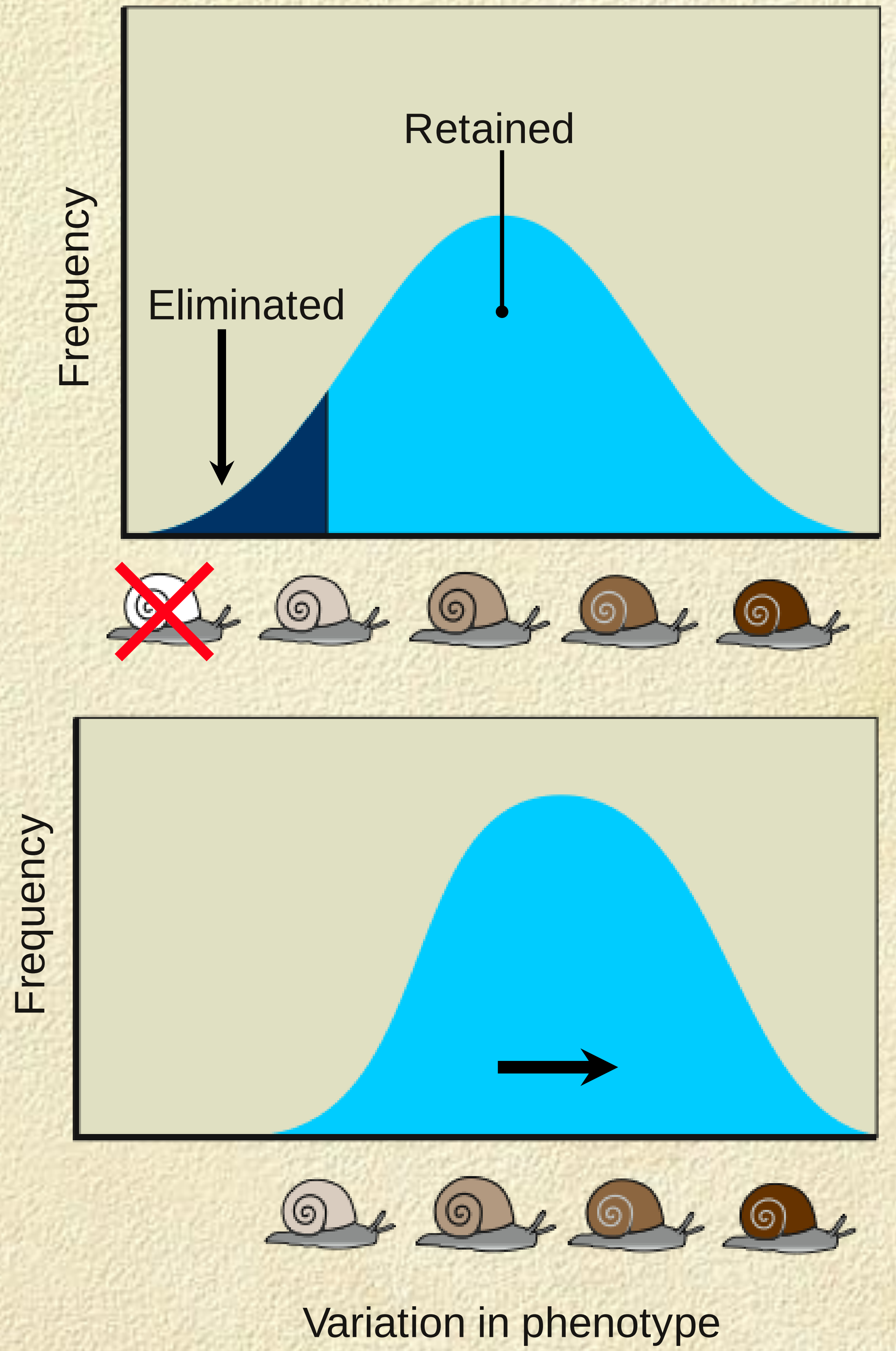
- Most common during periods of environmental change.
- Directional selection favors the phenotypes at one extreme of a phenotypic range.
- Selection reduces variation at one extreme of the range while favoring variants at the other end. The resulting bell shaped curve shifts in the direction of selection.
- **EXAMPLE**: Fossil evidence shows that the average size of black bears in Europe increased with each ice age, and decreased again during the interglacials.





Directional Selection 2

- Before selection (right) there is a broad range of variation in the population:
- After selection (right) and some generations later there is a reduction in variation at one extreme of the range while favoring variants at the other end.



Peppered Moths 1

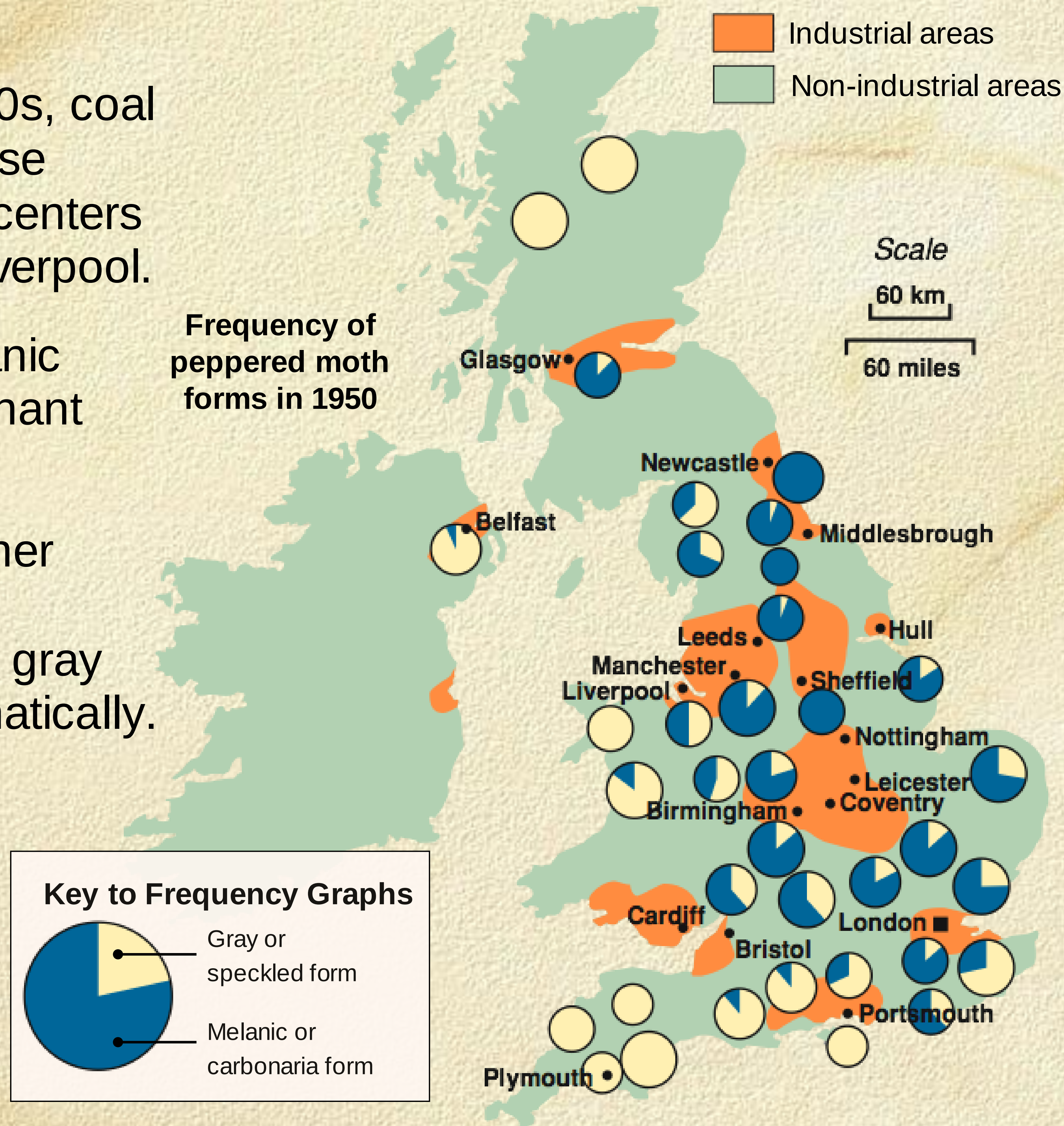
- The peppered moth, *Biston betularia*, occurs in two forms (or morphs):
  - The **mottled** or **gray** form is well camouflaged and less conspicuous (to predators) against the lichen-covered bark of trees in unpolluted regions.
  - The **dark melanic** forms are conspicuous in such environments as their body shape stands out against the background.
- With the onset of the Industrial Revolution in England, the air quality declined, killing off lichen and resulting in a marked increase in the relative frequency of the dark moths.
  - In a polluted environment, **directional selection** favored the melanic forms.

Gray or mottled form of the **peppered moth** *Biston betularia*; camouflaged on lichen

Melanic or carbonaria form of the **peppered moth** *Biston betularia* ; conspicuous on lichen, but camouflaged on soot-covered vegetation

Peppered Moths 2

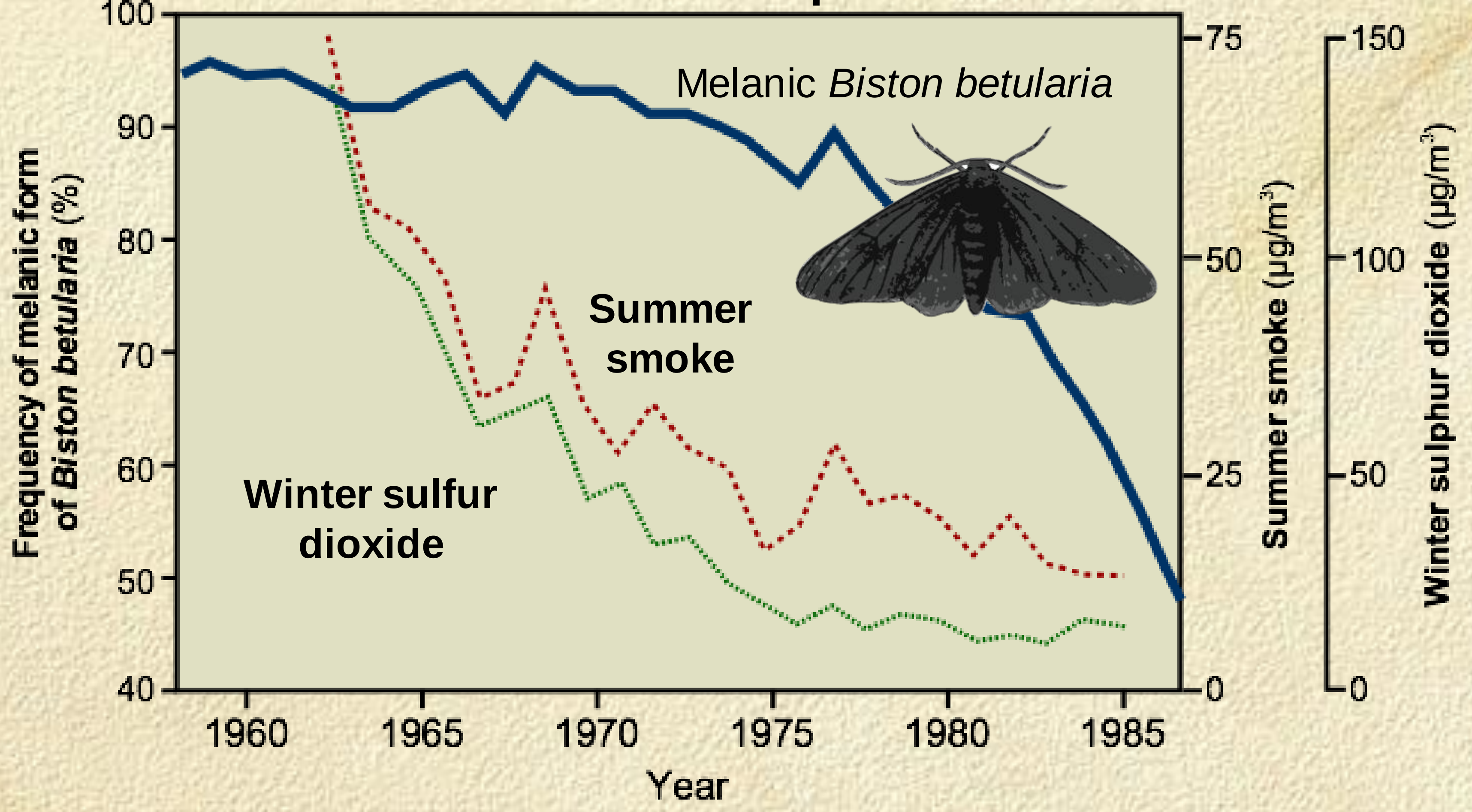
- In the 1940s and 1950s, coal burning was still intense around the industrial centers of Manchester and Liverpool.
- During this time, melanic forms remained dominant in these regions.
- In the rural areas further south and west of the industrial centers, the gray forms increased dramatically.



Peppered Moths 3

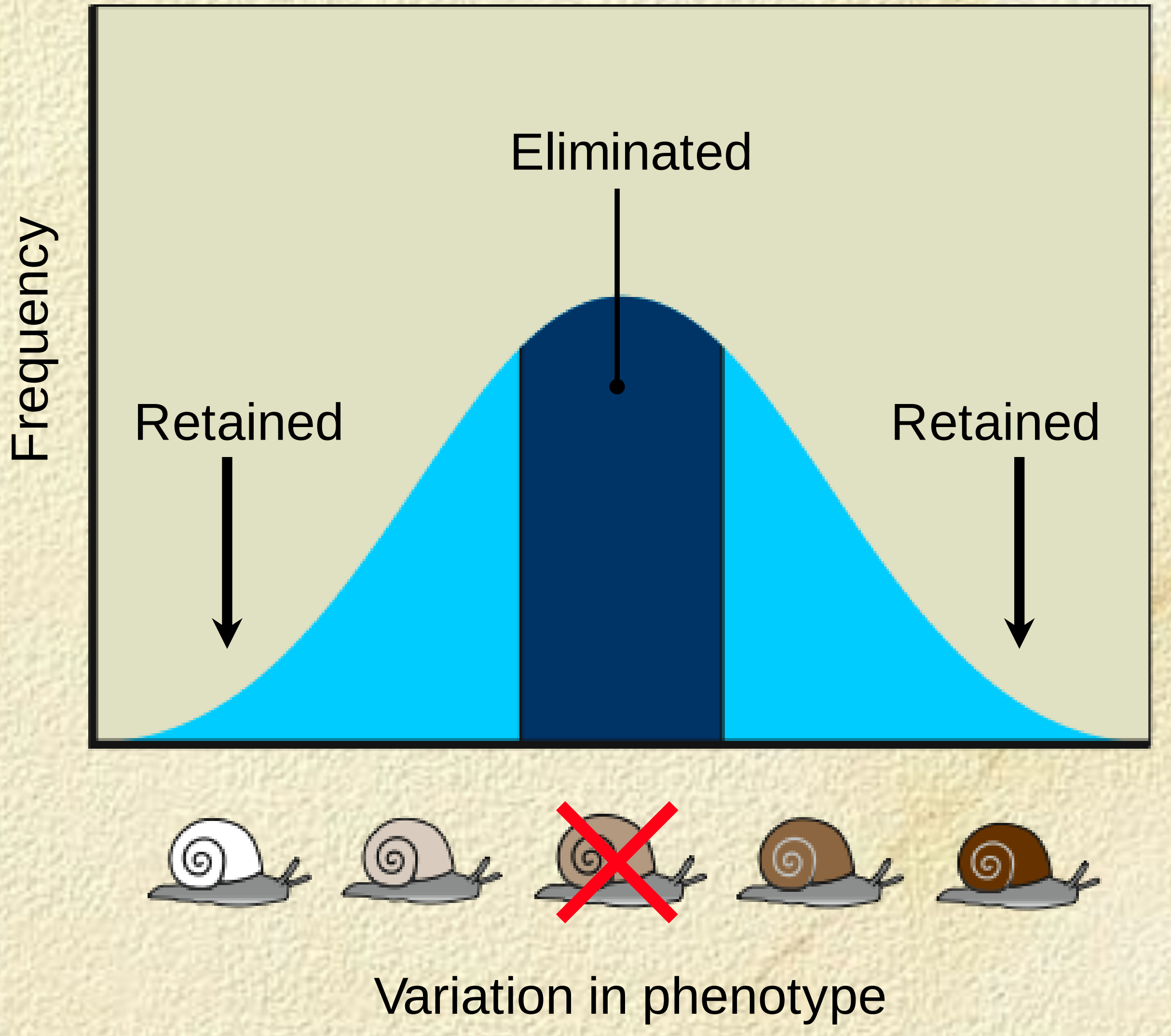
- With the decline of coal burning factories and the Clean Air Acts in cities, the air quality improved between 1960 and 1980.
  - Sulphur dioxide and smoke levels dropped to a fraction of their previous levels.
  - This caused the proportion of melanic peppered moths to plummet…
  - Now, with cleaner air, selection is increasingly in favor of the gray form.

Frequency of melanic peppered moth related to reduced air pollution



Disruptive Selection 1

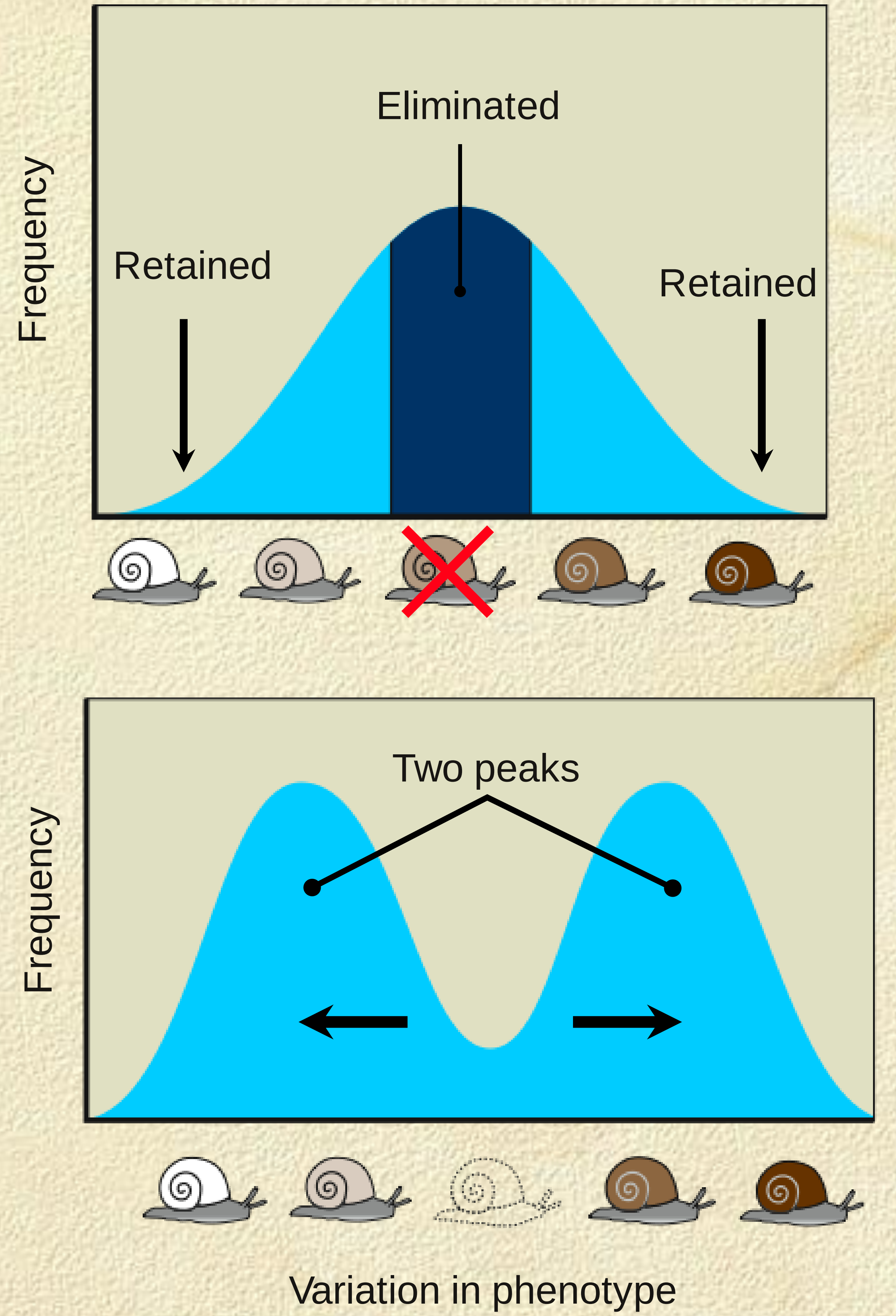
- Disruptive selection favors phenotypes at both extremes of a phenotypic range over intermediate variants. The bell shaped curve acquires two peaks (i.e. becomes bimodal).
- Disruptive selection may occur when environmental conditions are varied or when the environmental range of an organism is large.
- This type of selection can lead to the formation of clines or ecotypes (organisms of the same species that are slightly different in appearance), and **polymorphism**.



Disruptive Selection 2

- **Before selection** (right) there is a broad range of variation in the population:

- **After selection** (right) and some generations later individuals at both extremes of a phenotypic range are favored over intermediate variants (two peaks).



Disruptive Selection in Mimics

- An example of disruptive selection is seen in the **mimicry complexes** of African swallowtail butterflies.The African mocker swallowtail butterfly, *Papilio dardanus*, avoids predators by resembling poisonous butterfly species (it is a Batesian mimic).
- It has a large range and, in various regions, it looks very different, depending on which species of poisonous butterfly it mimics. These **Batesian mimics** look like completely different species but are one.
- Disruptive selection favours the extreme color patterns and nothing in between.
- In Madagascar, where no noxious species are found, it exists as one morph.

Artificial Selection

- Humans have controlled the breeding of domesticated animals and plants for centuries.
- This practice, called artificial selection or selective breeding, involves breeding (selecting) from individuals with the most desirable phenotypes. It can result in an astounding range of phenotypic variation over relatively short time periods.
- Selection imposed by humans is often more rapid and intense than that occurring in nature.
  - Strong selection pressures can be applied to produce extremes of diversity and development.

Brassicas

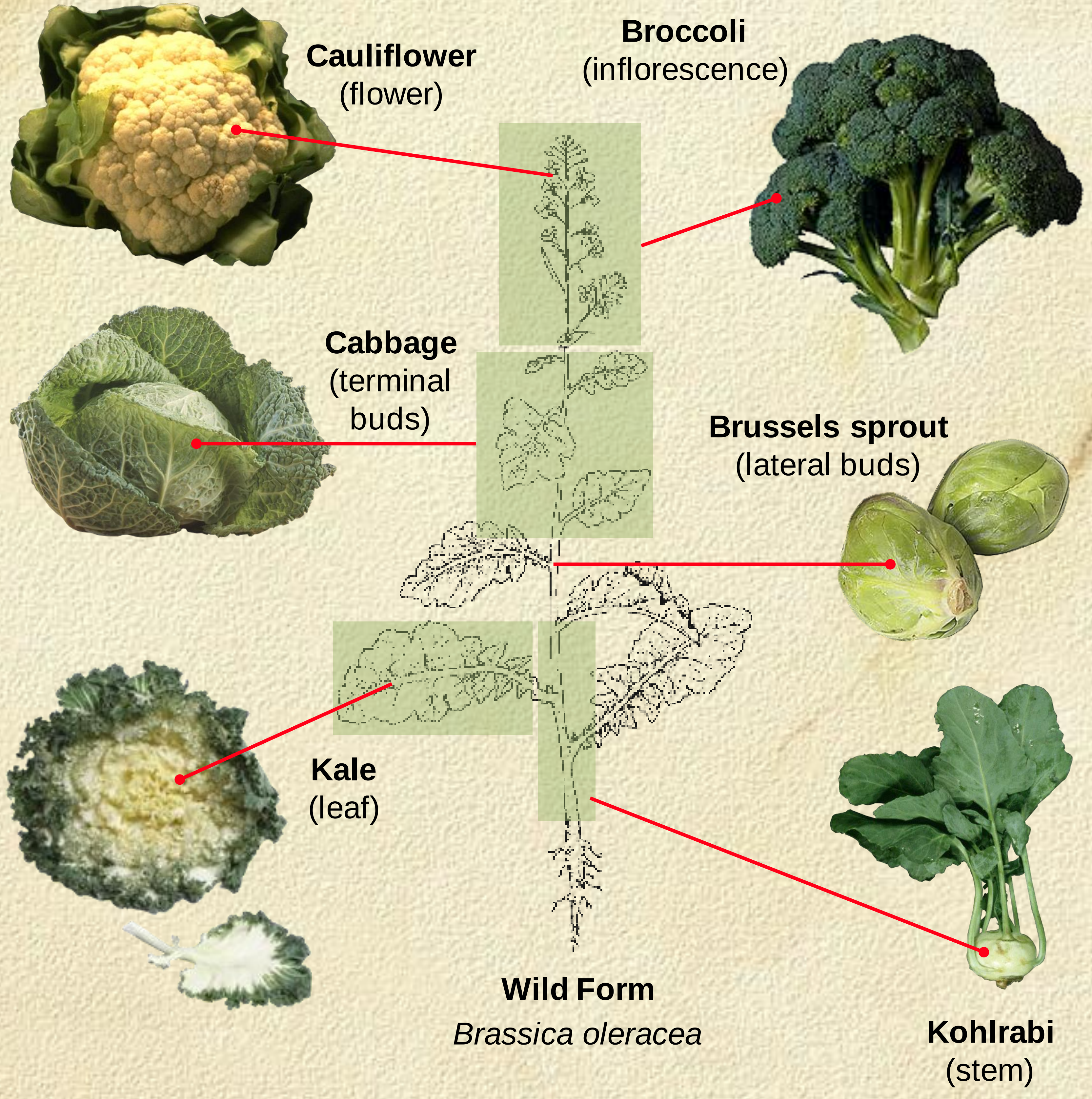
Wheat and other grain crops

Domestic pigeons and doves

Domestic dogs (pug)

Artificial Selection in Brassica

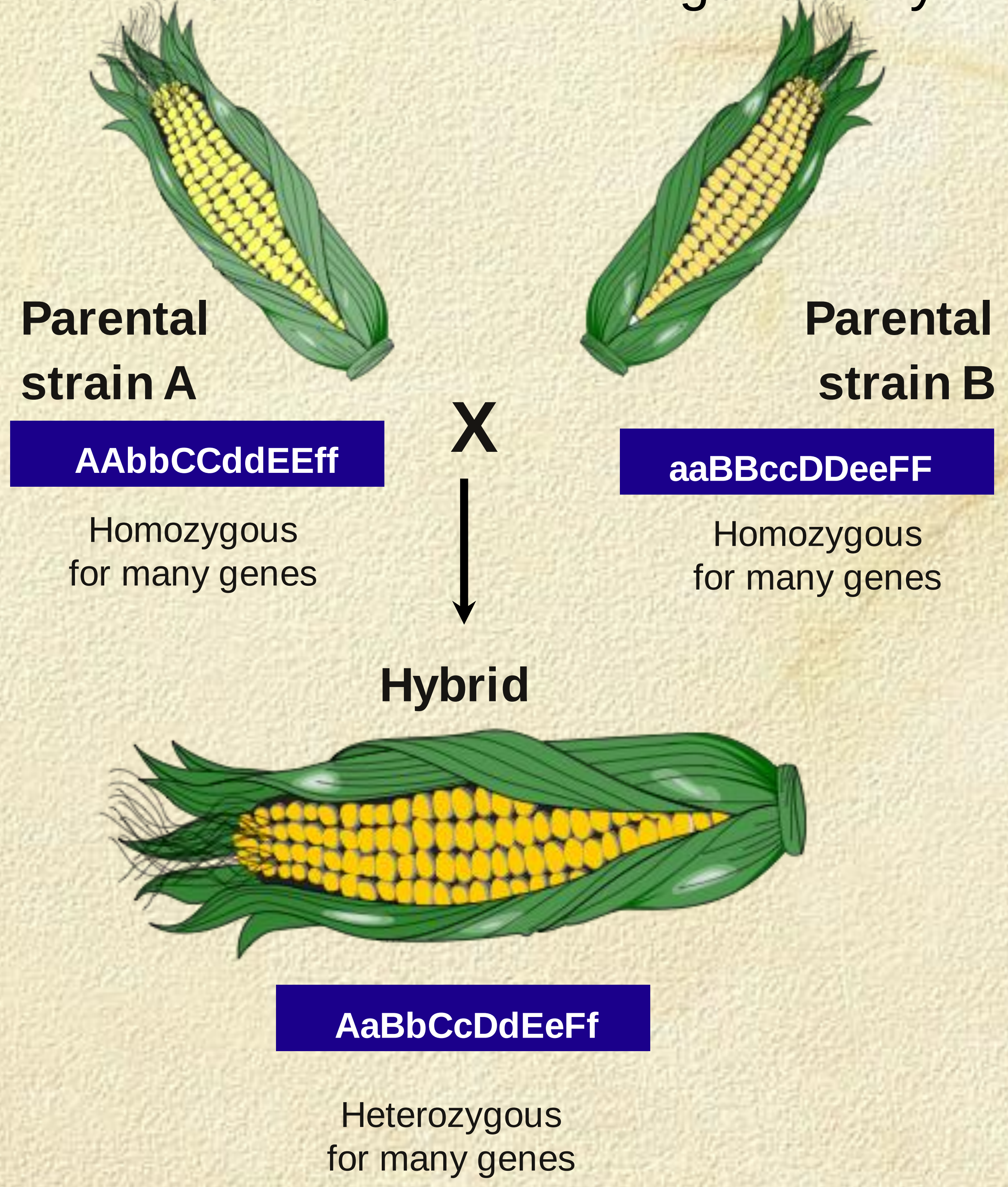
- Different parts of the wild brassica have been developed by human selection to produce at least six distinctly different vegetables.
- All these vegetables form a single species and will interbreed if allowed to flower.
  - **Example**: The new “broccoflower” is a cross between broccoli and cauliflower.



Hybrid Vigor

- Hybrids are the progeny resulting from a cross between two genetically different individuals.

- Hybrids maybe identical to the parents in some traits but not all.
- Hybrids recombine traits of (often inbred) parental lines and show **increased heterozygosity**.
- This is associated with greater growth survival, and fertility in the offspring; a phenomenon known as **hybrid vigor** or heterosis.
- The reasons for hybrid vigor are not always clear. Heterozygotes may benefit from the effects of a number of different interactions expressed in the phenotype.



Hybrid Corn

- **EXAMPLE**: Hybrid corn is valued for its high productivity. It is produced by crossing inbred **parental** strains with a high degree of **homozygosity**.

Maize breeding program, Texas A&M University

Inbred A **Hybrid** **Inbred B**

Artificial Selection in Dogs

- Dogs were probably first domesticated at least **14 000** years ago from a **gray wolf** ancestor.
- Some **400 breeds** have been bred from this single wild species as a result of selective breeding by humans.
  - Example: The staffordshire bull terrier was produced by breeding bulldogs and terriers. From each litter, breeders selected pups with the characteristics they desired.

Gray wolf

Bulldog

Terrier

Staffordshire bull terrier

Staffordshire bull terriers combine characteristics of both bulldogs and terriers

Artificial Selection in Dogs

- The gray wolf is distributed throughout Europe, North America and Asia. Amongst this species, there is a lot of phenotypic variation.
- Selection is based on both physical and behavioral characteristics. In this way, different breeds have been suited to different tasks.
- Five ancient dog breeds are recognized, from which all other breeds are thought to have descended by artificial selection.

Mastiff-type
Originally from Tibet, this breed dates back to the Stone Age

Pointer-type
Bred for the purpose of hunting small game.

Greyhound
One of the oldest breeds, originating the Middle East.

Sheepdog
Originated in Europe and bred for stock protection.

Wolf-type
Developed in snow-covered habitats in Alaska, northern Europe, and Siberia.