

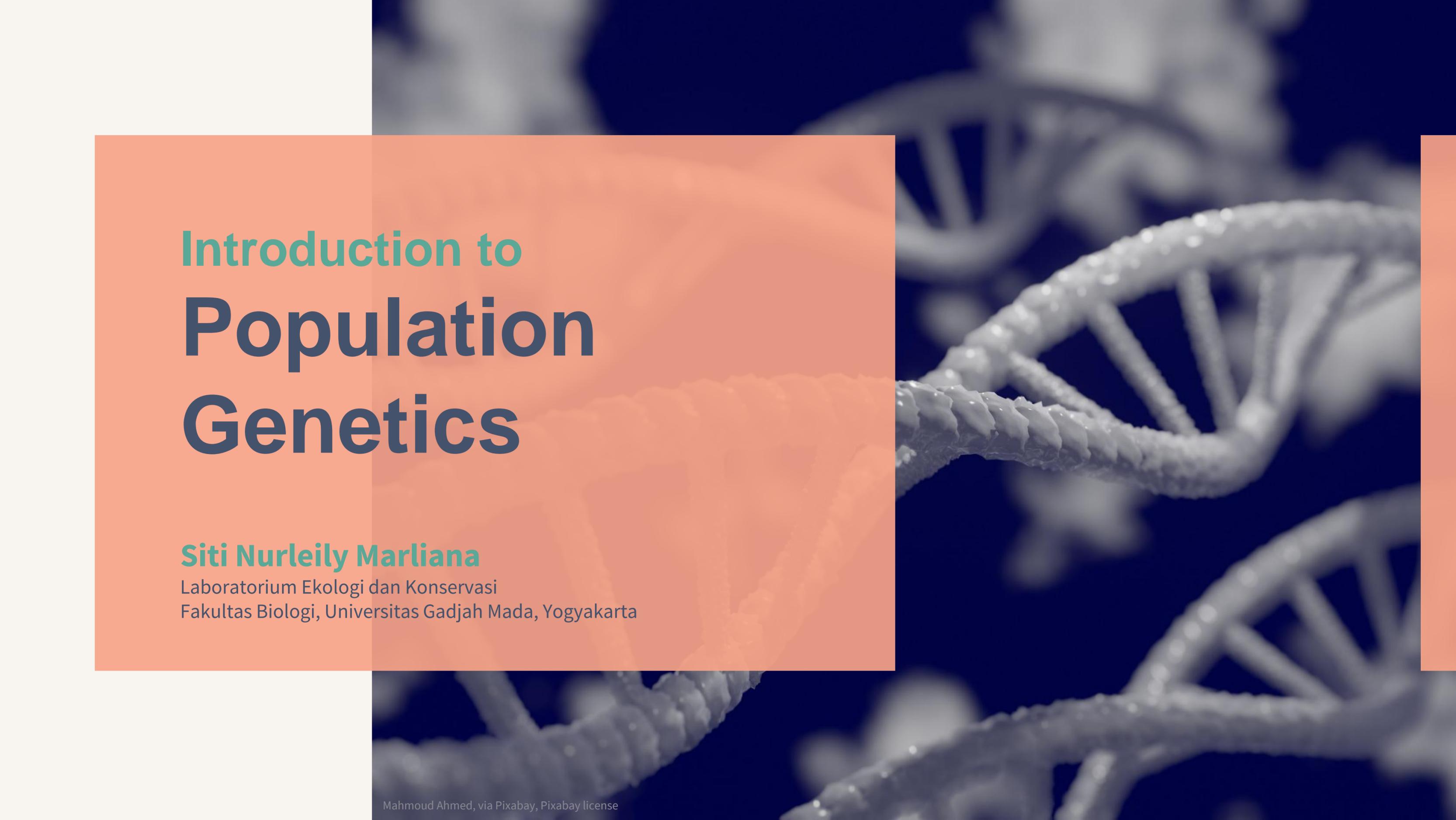


# PERNYATAAN SANGKALAN

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A background image of a DNA double helix structure, rendered in a light blue/white color against a dark blue background. The helix is shown in a perspective view, curving across the frame. The text is overlaid on a semi-transparent orange rectangular area.

# Introduction to Population Genetics

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# Population genetics

The study of "the genetic composition of biological populations, and the changes in genetic composition that result from the operation of various factors, including natural selection." (Stanford Encyclopedia of Philosophy 2012)



Change of allele, genotype, and phenotype frequencies over time.



Patterns of genetic variation in populations and their causes.

Understanding the variation in a population to determine how evolution takes place in response to different evolutionary pressures.

Bridging gap between gene and species evolution (i.e. microevolution).

# Evolution revisited

Individuals are selected;  
populations evolve.

Natural selection works on the  
existing variation.

Differential reproductive  
success: who bears more  
offspring, more likely to  
survive  
("survival of the fittest").

The more variation, the more choice for the selection, the more  
adaptable a population will be.

Genetic makeup of population changes over time.

Individuals with an adaptive trait leave more surviving offspring.

Favorable traits (greater  
fitness) become more  
common.

# Microevolution

Microevolution is change in the traits of a population.

Occurs through change of gene pools.

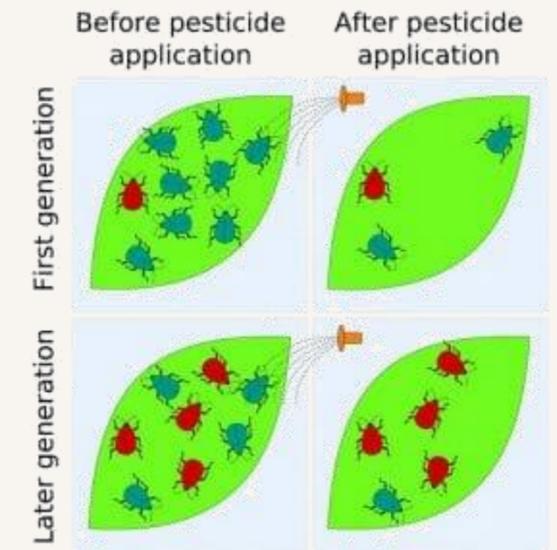
Results in more or less variation amongst individuals in a population.

The focus of study in ecological population genetics.

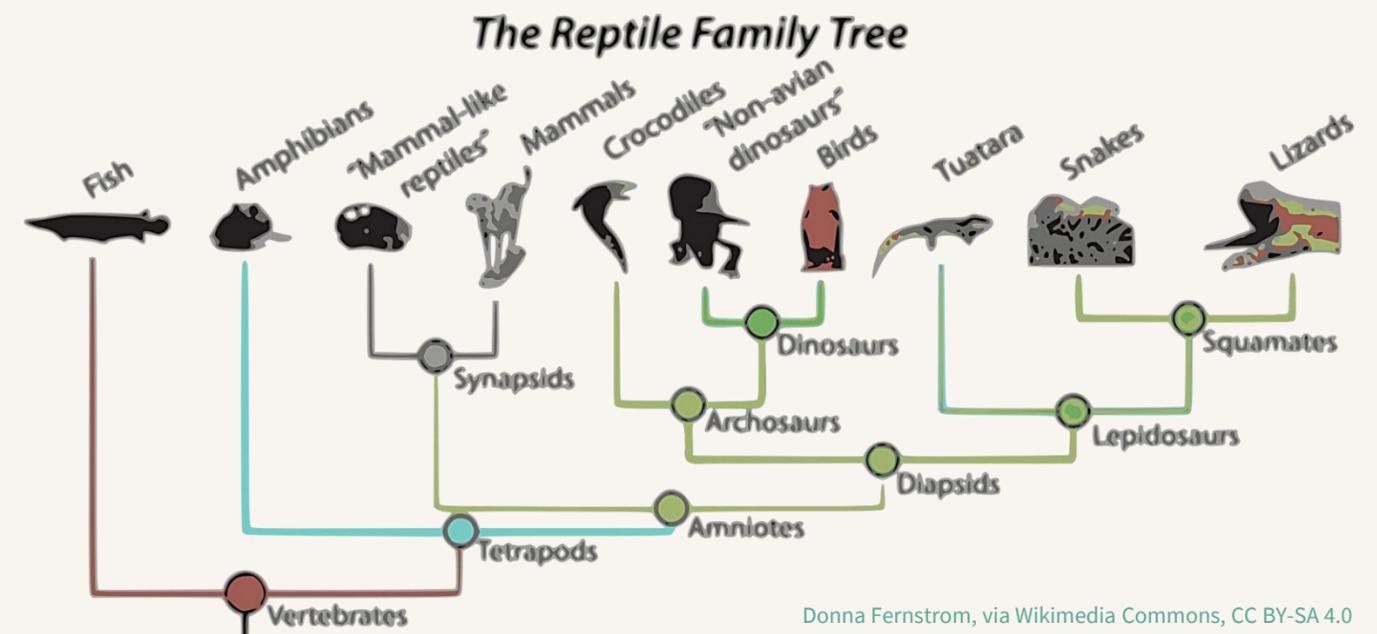
Microevolution

vs.

Macroevolution

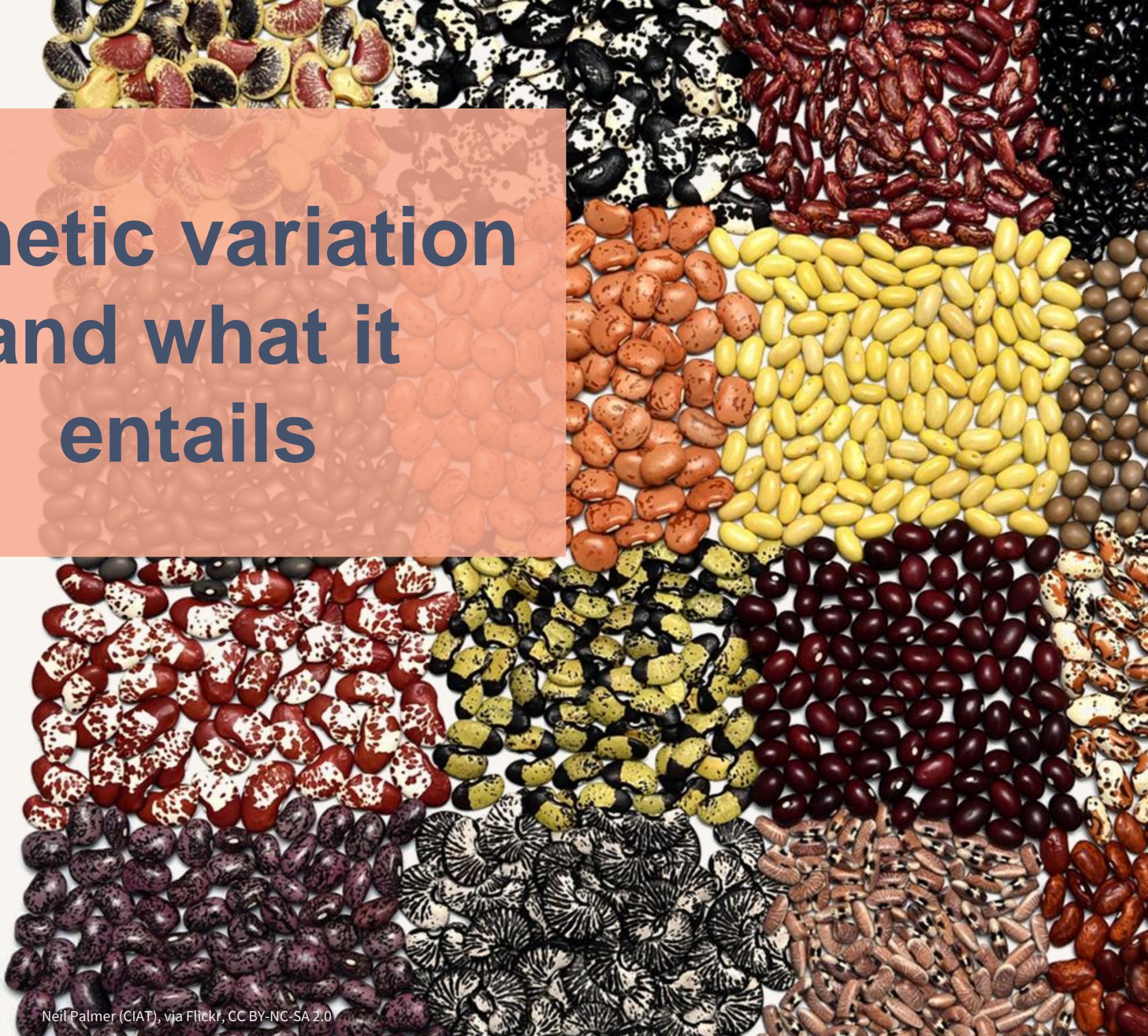


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Donna Fernstrom, via Wikimedia Commons, CC BY-SA 4.0

# Genetic variation and what it entails



Neil Palmer (CIAT), via Flickr, CC BY-NC-SA 2.0

01

# The importance of genetic variation

Increases a species' ability to evolve.

➤ The higher the variation, the higher a species' adaptability.

➤ Without adaptation ability, a population may become extinct.

The role of genetic variation:

➤ **Biodiversity maintenance:** by allowing a population to diverge.

➤ **Conservation of species:** by allowing adaptation to environmental changes.

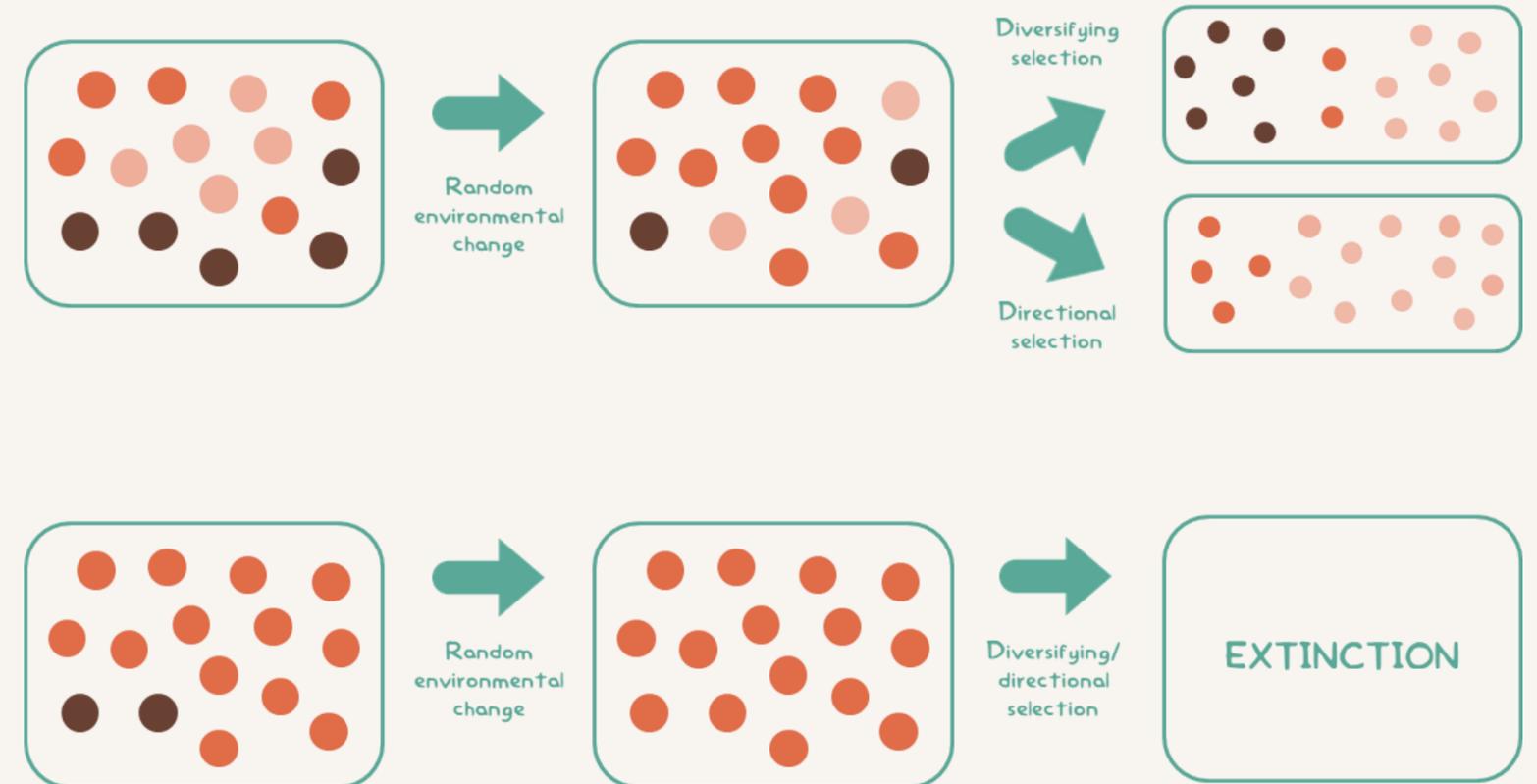
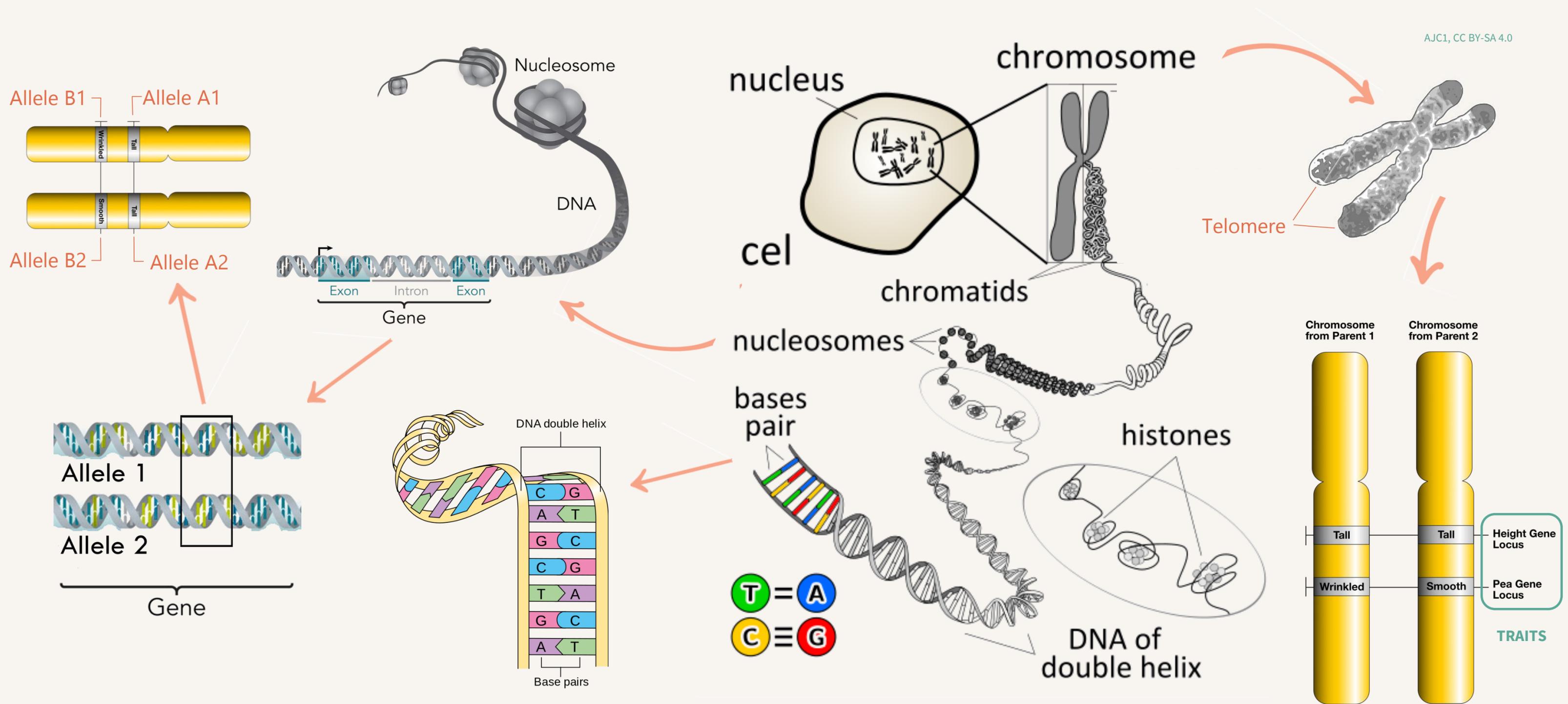


Illustration of the role of genetic variation in maintaining a long-term survival of a population. Top, with more diversity; bottom: with less diversity.

# Structure of a chromosome



AJC1, CC BY-SA 4.0

# Genotype, phenotype, and alleles

Genotype

an organism's complete set of genes, or the alleles related to a single trait (e.g. Aa), or to a number of traits.

Phenotype

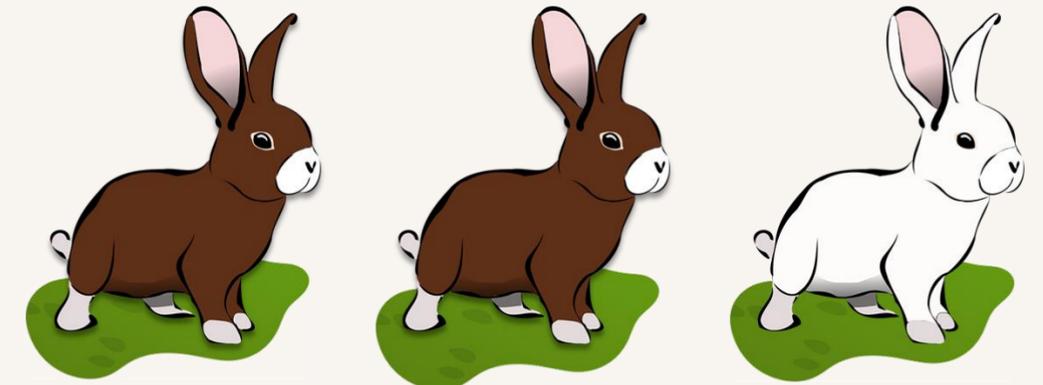
the observable physical properties of an organism; is determined by its genotype.

Alleles

variant forms of a gene.

Can differ from one population to another.

Phenotype



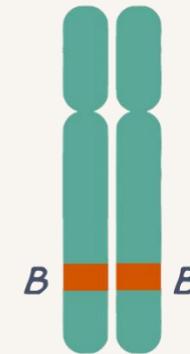
Genotype

*BB*

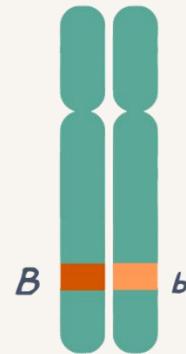
*Bb*

*bb*

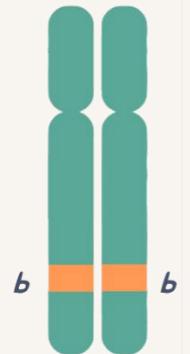
Chromosomes



*Homozygous*



*Heterozygous*



*Homozygous*

Alleles

**B** Allele for brown fur; dominant

**b** Allele for white fur; recessive

# Gene pool

A complete set of genetic information in *all* individuals within a *population*.

Contains all of the alleles of a population.

Species have a shared gene pool.

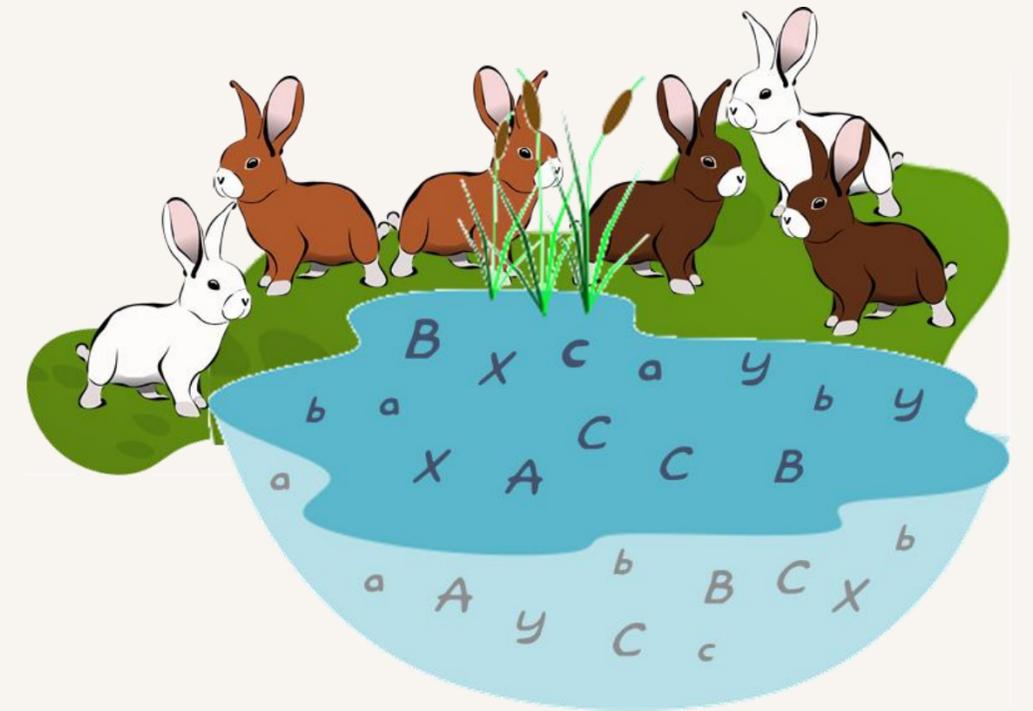
Effect on breeding:



Members of the same species can interbreed and produce fertile offspring.



Members of different species can interbreed, but offspring is usually sterile, & genes are not shared.



All alleles in the population

Vectors: [Rabbit] by Gold\_Wolf and [Lake] by Jerzy Górecki, via Pixabay, Pixabay license

# Frequencies of genotype and allele

Genotype frequencies



proportion of individuals in a population with a specific genotype.

Allele frequencies

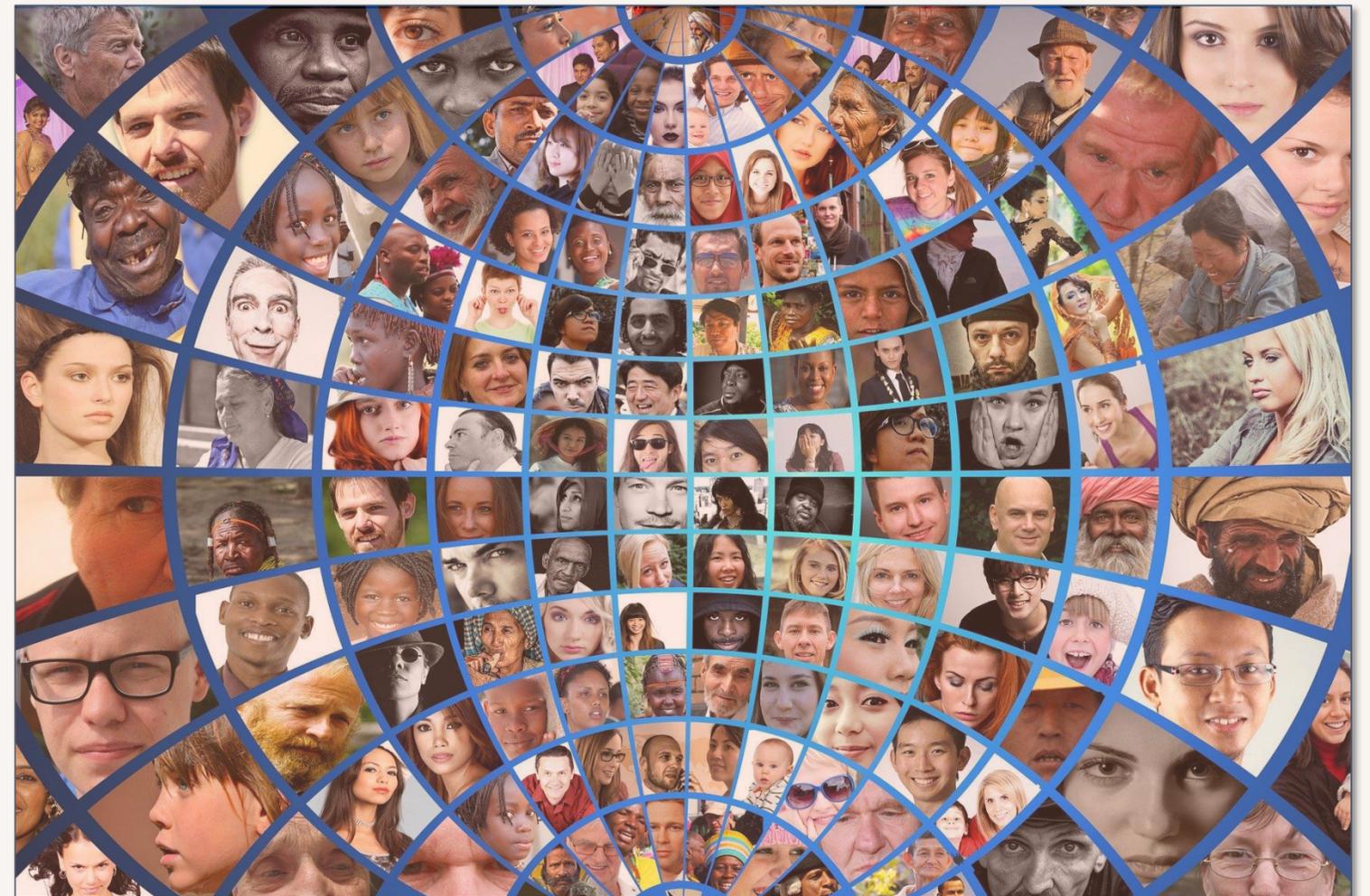


proportion of any specific allele in a population.



estimated from genotype frequencies.

Gerd Altmann, via Pixabay, Pixabay license



Human characteristics around the world represent the diversity of alleles in the human population.

# Frequencies of genotype and allele



$$\text{Frequency of allele } A = \frac{\text{No. of copies of allele } A \text{ in population}}{\text{Total number of } A/a \text{ gene copies in population}}$$

**GENOTYPE FREQUENCY:** How often we see each allele combo

Freq. of  $WW$  =  $6/9 = 0.67$   
 Freq. of  $Ww$  =  $1/9 = 0.11$   
 Freq. of  $ww$  =  $2/9 = 0.22$

**PHENOTYPE FREQUENCY:** How often we see white vs. purple

Freq. of purple =  $7/9 = 0.78$   
 Freq. of white =  $2/9 = 0.22$

**ALLELE FREQUENCY:** How often we see each allele

$p$  = Freq. of  $W$  =  $13/18 = 0.72$   
 $q$  = Freq. of  $w$  =  $5/18 = 0.28$

ORIGINAL GENERATION

**ORIGINAL GENERATION**

$p$  = Frequency of  $W$  =  $13/18 = 0.72$   
 $q$  = Frequency of  $w$  =  $5/18 = 0.28$

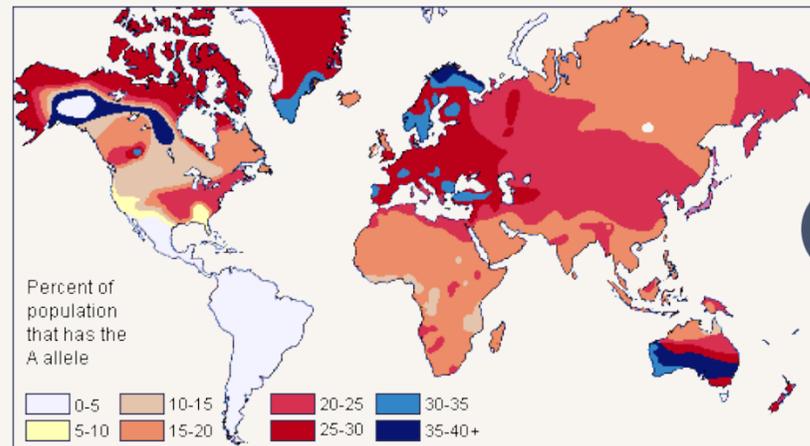
Old plants die → Their offspring grow up

**NEW GENERATION**

$p$  = Frequency of  $W$  =  $8/18 = 0.44$   
 $q$  = Frequency of  $w$  =  $10/18 = 0.56$

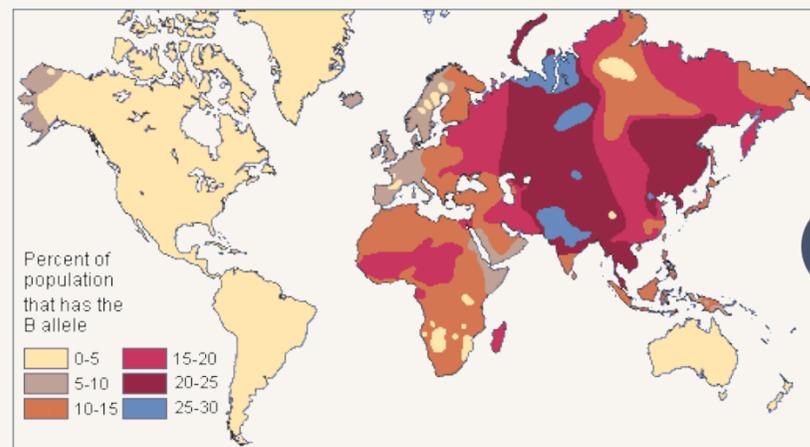
Allele frequencies change → population evolves

# Example: ABO blood type allele frequency



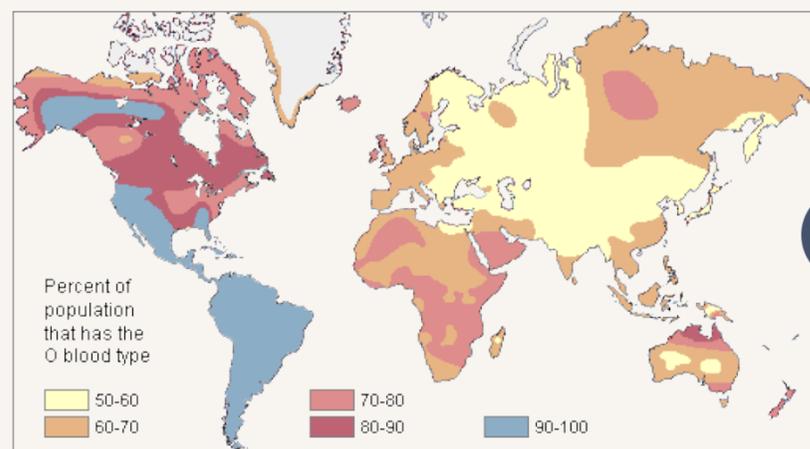
**I<sup>A</sup> allele frequency: 21%**  
 Commonly found among Blackfoot people of Montana, the Australian Aborigines, Saami people (Scandinavia); absent among Central and South American Indians.

The distribution of the ABO blood type in native populations of the world.



**I<sup>B</sup> allele frequency: 16%**  
 Most common in Central Asia; lowest among the indigenous peoples of the Americas and Australia. Rarest ABO blood allele.

ABO blood groups alleles, phenotypes, and genotypes.



**i allele frequency: 63%**  
 Most common in the world, particularly among the indigenous populations of Central and South America (almost 100%). The lowest frequency is found in Eastern Europe and Central Asia.

$I^A$ (co-dominant)  $I^B$ (co-dominant)  $i$ (recessive)	phenotype (blood type)	genotype
	Type A	$I^A I^A$ $I^A i$
	Type B	$I^B I^B$ $I^B i$
	Type AB	$I^A I^B$
Type O	$i i$	

Maliayee, via Wikimedia Commons, CC BY-SA 4.0

Sources: [A and B blood type] by Muntuwandi, CC BY-SA 3.0; [O blood type] by anthro palomar, CC BY-SA 3.0, all via Wikimedia Commons.

# Genetic variation and evolution

Evolution involves changes in allele and/or genotype frequencies over time.

These mechanisms can increase or decrease genetic variation.

## Main mechanisms

### Natural selection

Increases frequency of favored alleles



### Mutation

Creates new alleles



### Gene flow (migration)

Increases similarity of different populations



### Genetic drift

Causes random change of allele frequencies



### Genetic recombination

Forms new combinations of different alleles



### Nonrandom mating

Changes genotype, but not allele, frequencies



## Ancillary mechanisms

# Hardy-Weinberg equilibrium



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# The Hardy-Weinberg principle

Allele and genotype frequencies in a population will remain *constant* when *no evolutionary mechanisms* influence it.

Evolution will not occur in a population under a **Hardy-Weinberg equilibrium** for a gene, as allele frequencies will stay the same across generations.

## The Hardy-Weinberg equilibrium for 2 alleles

For example: A locus has two alleles, **A** and **a**. The **A** allele frequency is **p** and the **a** allele frequency is **q**.

### EVOLUTION:

a change in frequencies of alleles in the gene pool of a population over time.

Remember...

## The role of the Hardy-Weinberg principle

Characterizes the distributions of genotype frequencies in an unevolving population.

Provides a theoretical benchmark to evaluate real-life populations.

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

Thus, the genotype frequency distribution is:

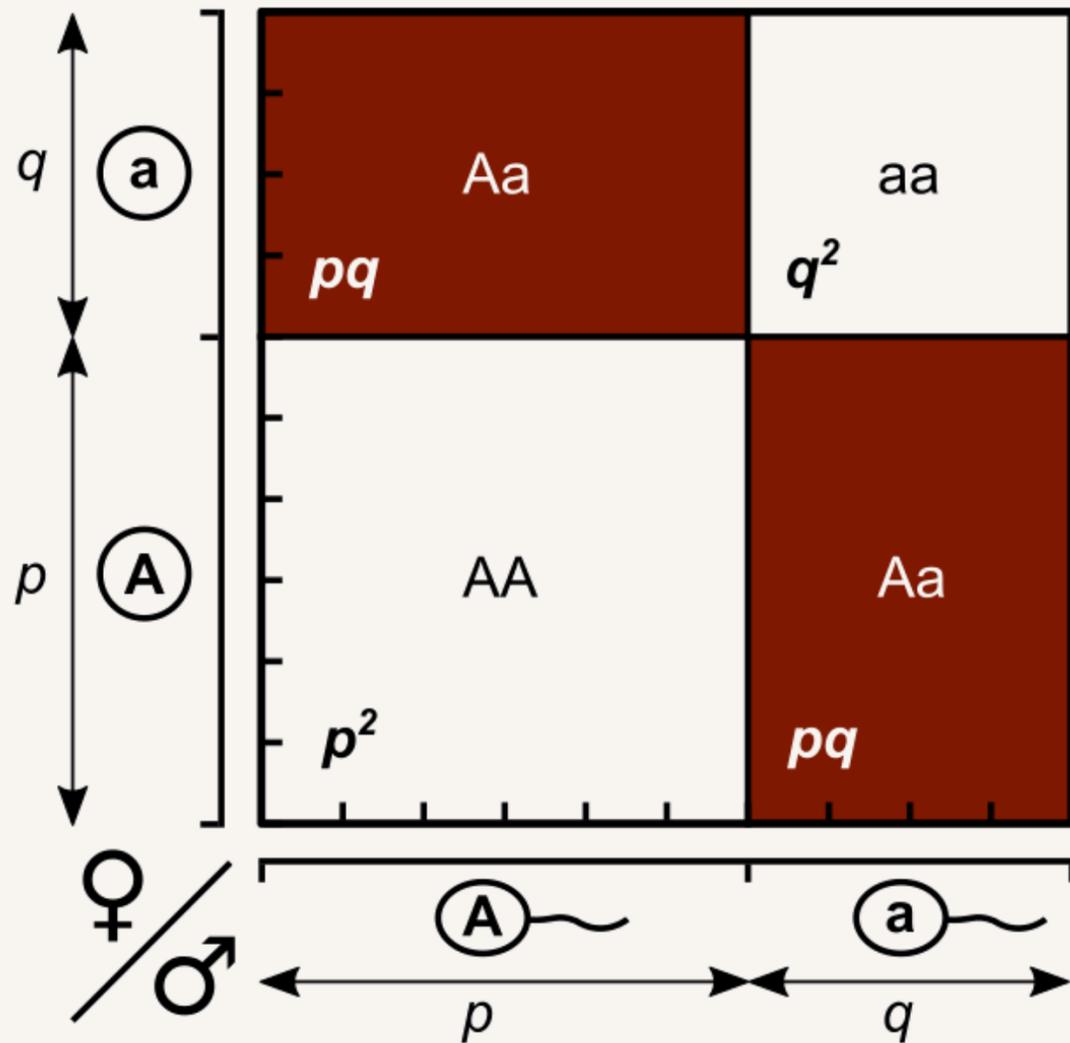
$$AA = p^2$$

$$Aa = 2pq$$

$$aa = q^2$$

For loci with  $k$  alleles:  $(p_1 + p_2 + \dots + p_k)^2 = 1$

# The Hardy-Weinberg equilibrium



Punnet square of Hardy-Weinberg equilibrium. Length of  $p$  and  $q$  corresponds to allele frequencies (here  $p = 0.6$ ,  $q = 0.4$ ). Then area of rectangle represents genotype frequencies (thus  $AA : Aa : aa = 0.36 : 0.48 : 0.16$ ).

The Parent Generation-1,000 individuals

	360	480	160
Genotypes			
Frequency of Each Genotype in the Population	0.36	0.48	0.16
Frequency of Each Allele in the Population	$0.36 + 0.24$ $0.6$		$0.24 + 0.16$ $0.4$

Simple overview of Hardy-Weinberg equilibrium in a hypothetical beetle population.

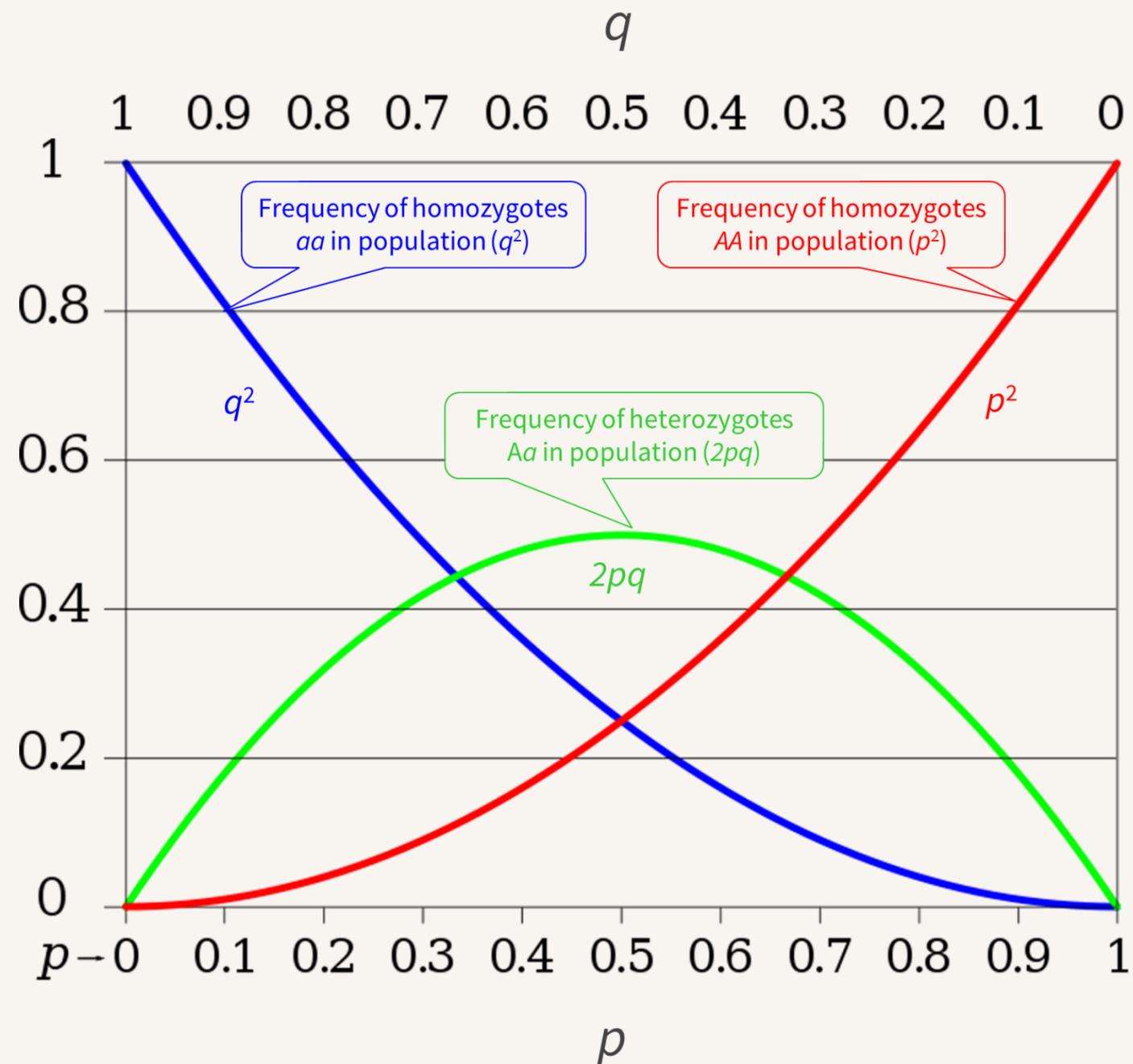
Angelahrtsock, via Wikimedia Commons, CC0

Offspring

	$p=0.6$ 	$q=0.4$ 	
	Father		
$p=0.6$ 	 $(p^2)=0.36$	 $(p^2)=0.24$	
Mother			
$q=0.4$ 	 $(pq)=0.24$	 $(q^2)=0.16$	
Genotypes			
Frequency of Each Genotype in the Offspring Population	0.36	0.48	0.16

When evolutionary influences are absent from a population, the offspring will have the exact same genotype and allele frequencies as the parent population.

# The Hardy-Weinberg equilibrium



Hardy-Weinberg proportions for two alleles

Frequency of heterozygotes  $Aa$  (heterozygosity) in population ( $2pq$ ) is highest when  $p = q = 0.5$ .

Rare alleles are found primarily in heterozygotes ( $p^2$  or  $q^2$  is much smaller than  $2pq$ ); the impact of rare, recessive alleles to the population is thus masked by the heterozygosity.

Hardy-Weinberg equilibrium is a *neutral equilibrium* (perturbation leads to a new equilibrium), not a stable one.

# Assumptions underlying the HW equilibrium

## MAIN ASSUMPTIONS

Population size infinitely large

Random mating

No migration

No mutations

No natural selection



## ADDITIONAL ASSUMPTIONS

Diploid organisms

Sexual reproduction

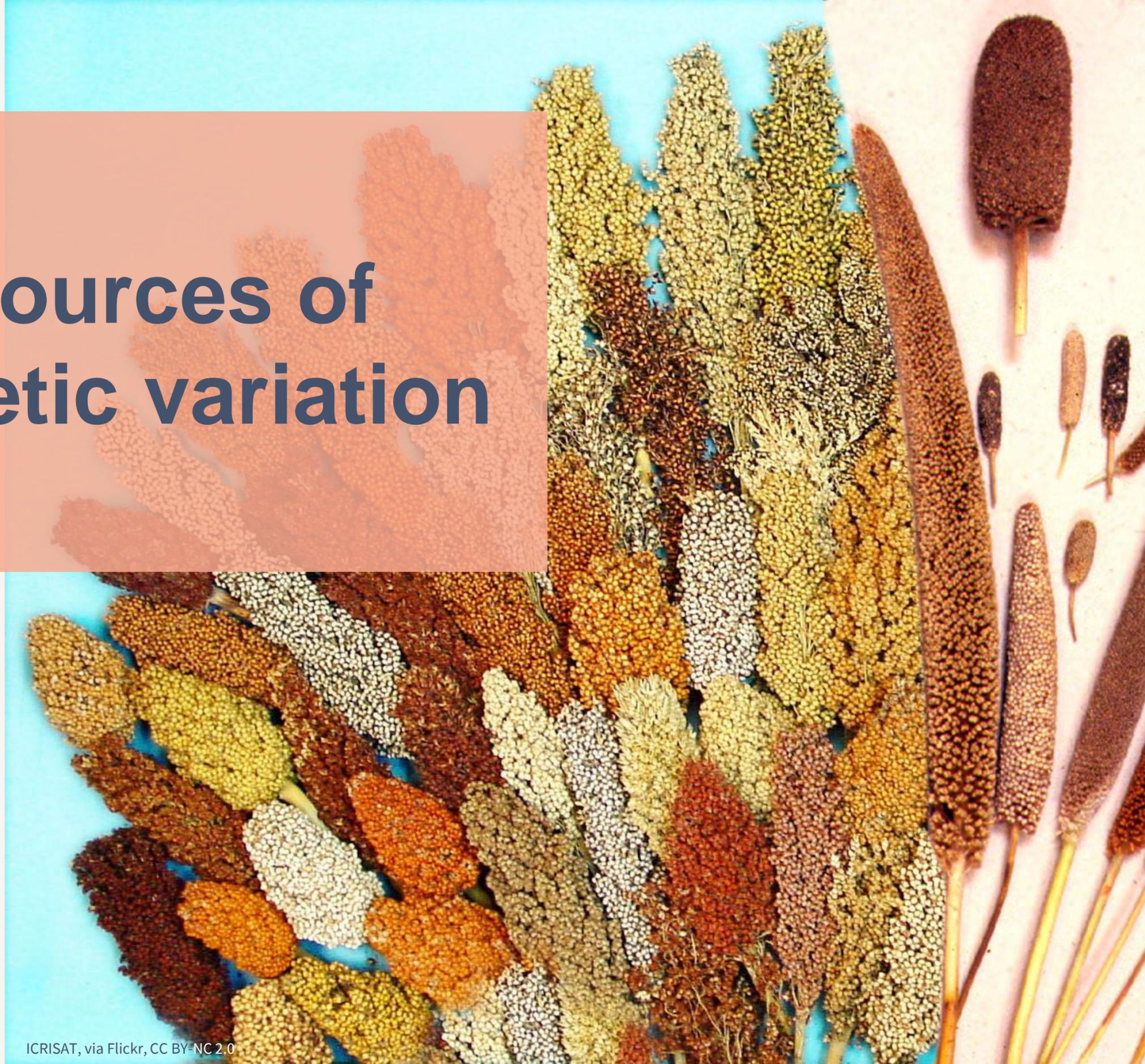
Non-overlapping generations

Same allele frequencies  
between sexes



If any one of the assumptions is not met, equilibrium is not reached; the population may evolve.

# Sources of genetic variation



ICRISAT, via Flickr, CC BY-NC 2.0

03

# Mechanisms that increase genetic variation

Several processes can produce new genes and alleles, thus increasing genetic variation.

➤ Formation of new alleles

➤ Alteration of gene number/position

➤ Rapid reproduction

➤ Sexual reproduction

Mechanisms producing genetic variation in a population

➤ Mutation

➤ Gen flow (migration)

➤ Genetic recombination



LepoRello, Wikimedia Commons, CC BY-SA 3.0



A red Darwin hybrid tulip "Apeldoorn" with a mutation resulting in half of one petal being yellow.



Joe King, Wikimedia Commons, CC BY-SA 3.0



Hybrid red-sika deer, a result of interbreeding between sika deer introduced into Western Europe with the native red deer.



Staff at NIMH, Wikimedia Commons, Public Domain



A mouse with the agouti coat color gene introduced into its DNA via gene targeting.

# 1. Mutation

Spontaneous changes in the genetic material (DNA sequence, amino acid sequence, and proteins).

➔ Rate is very low.

➔ Provides the raw material for evolutionary change (new alleles).

➔ Does not have a measurable effect on allele or genotype frequencies.

Radiation in the environment (cosmic rays, radioactive elements).

CAUSES

Mutagenic chemical compounds.

Chance events inherent in the process of DNA replication.

Result: new alleles

BENEFICIAL;  
spread through the population

DELETERIOUS;  
eliminated by natural selection

# Mutation types

## Types of mutation

Location of occurrence

Germline mutations  
(occur in gametes)

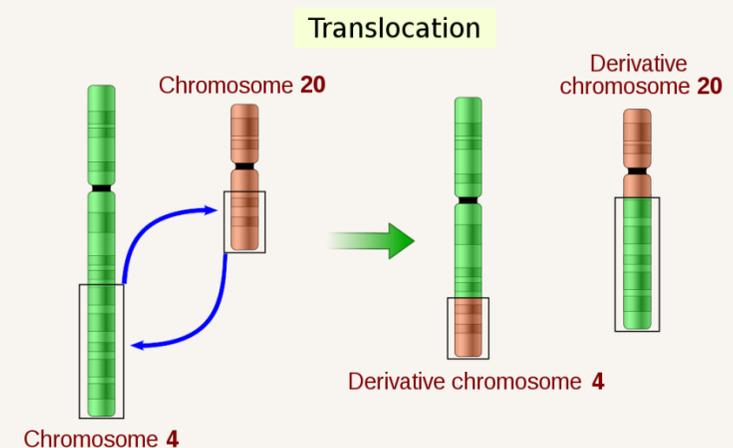
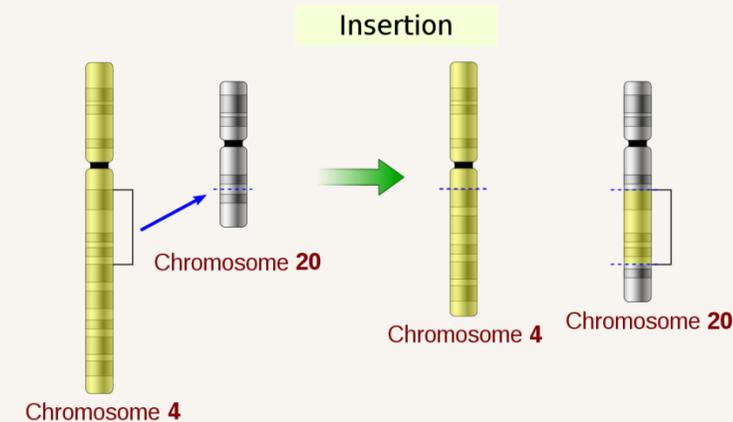
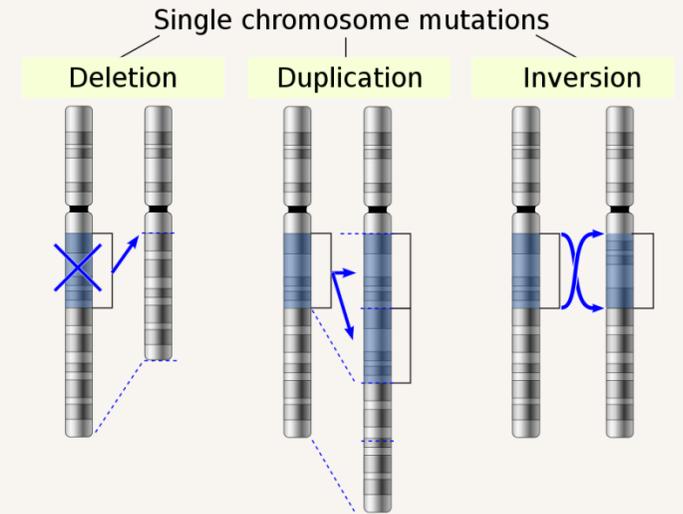
Somatic mutations  
(occur in other cells)

Types of changes made

Point mutations

Frameshift mutations

Chromosomal alterations



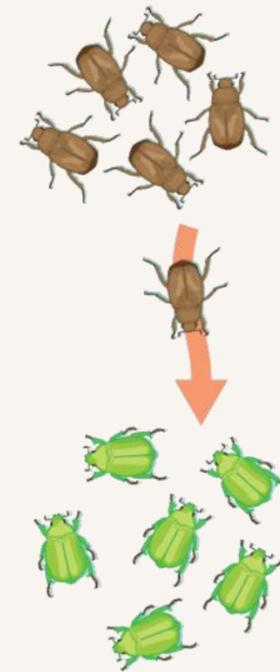
- Possible ways of chromosomal mutations:
- deletion,
  - duplication,
  - insertion,
  - inversion,
  - translocation.

## 2. Gene flow (migration)

Movement of individuals and their alleles in & out of populations.

➔ Individuals move to another population then interbreed, transferring new alleles to the population's gene pool.

➔ Changes allele distribution among populations (allele/genotype frequencies).



OpenStax/Rice University, via Wikimedia Commons, CC BY 4.0

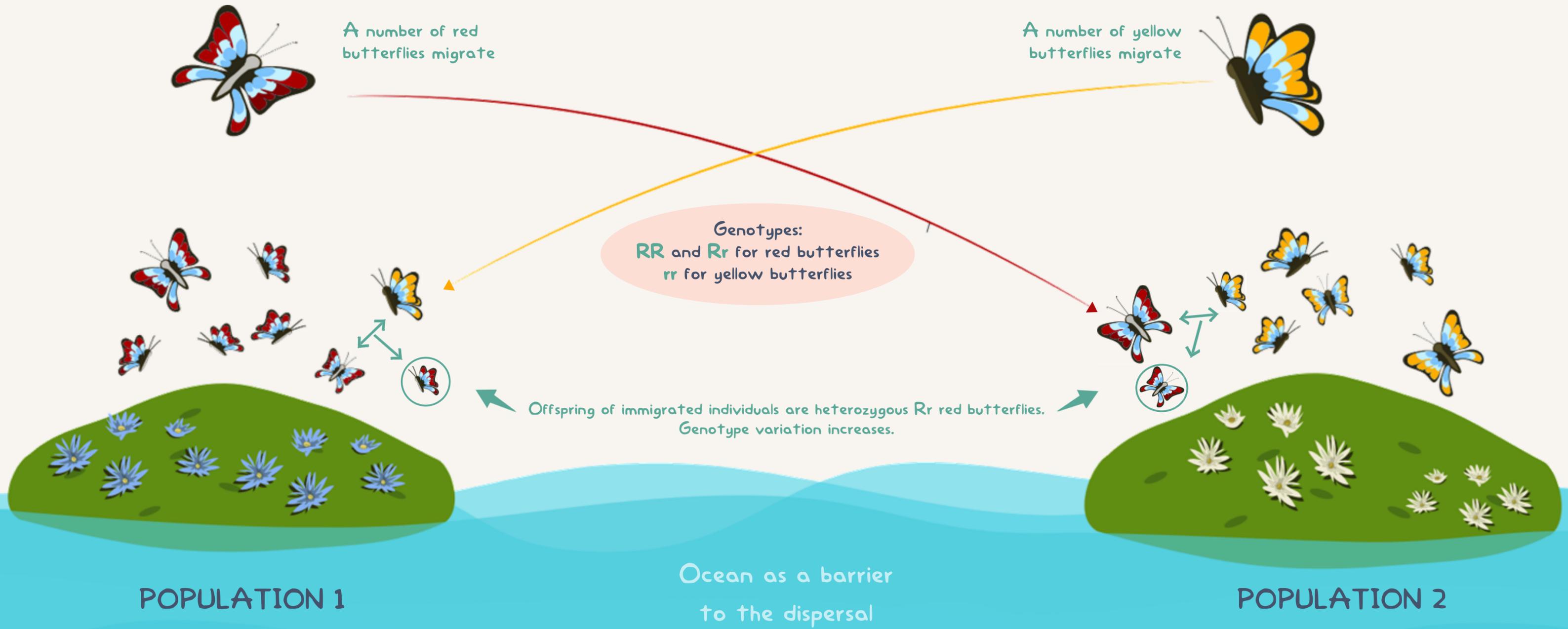
Result: genetic mixing

➔ Increases the genetic similarity among populations of a species.

➔ Prevents a species from fragmenting.

Occurs through dispersal (including migration in animals).

# Illustration of a gene flow

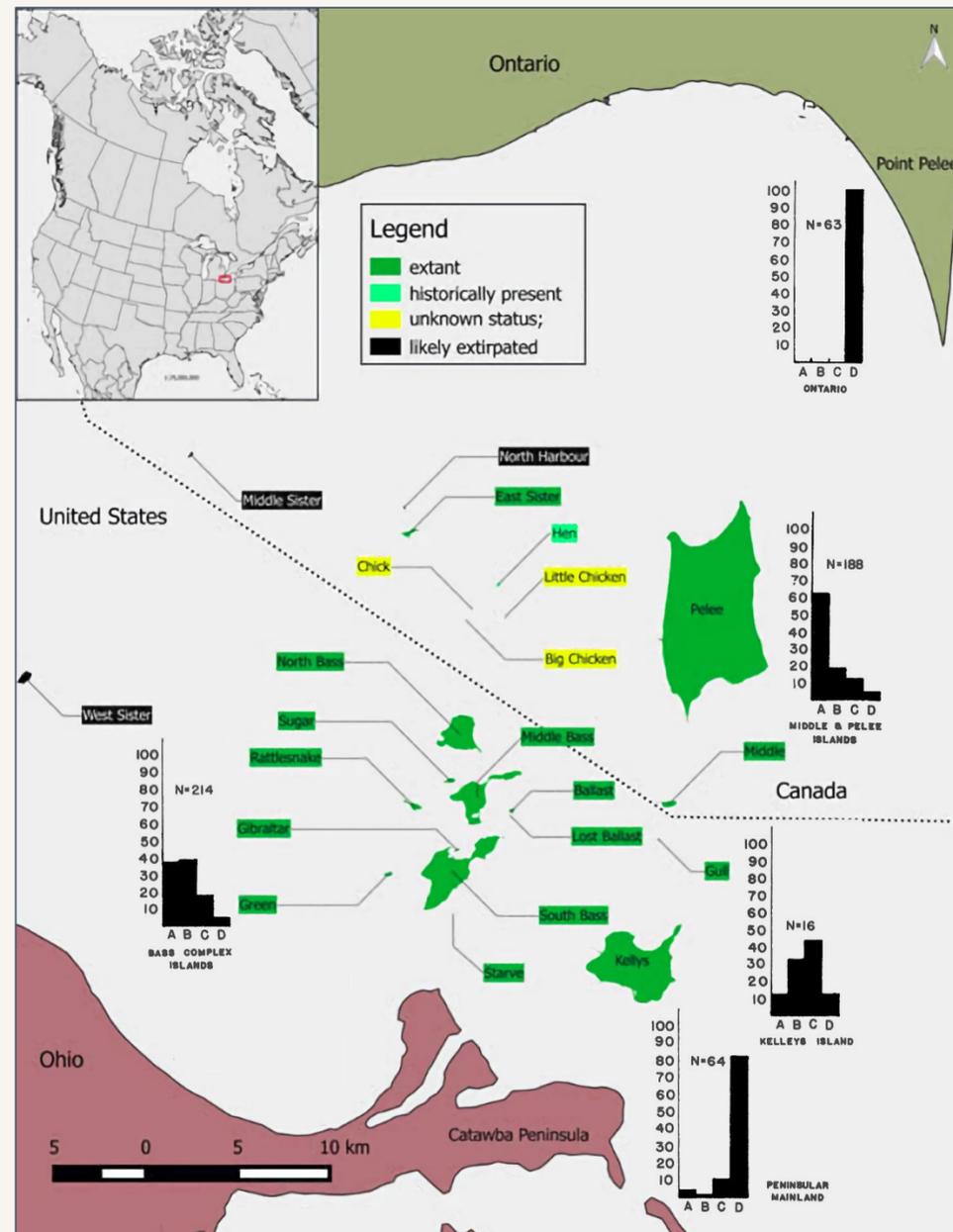


Selection pressure against recessive phenotype has created a homozygous **RR** red butterfly population.

Selection pressure against dominant phenotype has created a homozygous **rr** yellow butterfly population.

# Gene flow between *Nerodia sipedon* populations

COSEWIC, via Memorial University of Newfoundland, used under a Fair Use rationale



The Lake Erie water snakes (*Nerodia sipedon insularum*)

The classic Lake Erie islands' water snake color pattern is unbanded (selection on the islands favors unbanded snakes).

Some of the mainland snakes, which are strongly banded, migrated to the islands and bred with the local populations.

Frequency distribution of different coloration of *Nerodia sipedon* individuals in the Lake Erie islands and the mainlands (Ontario and Ohio), suggesting a gene flow event to the islands. Codes: **A**, unbanded; **B & C**, intermediate; **D**, strongly banded.

Donna Braig, via Flickr, CC BY-NC 2.0



Kristin Stanford, via Flickr, CC BY-NC 2.0

Results: equilibrium frequencies of the different phenotypes.

# 3. Genetic recombination

“...the rearrangement of DNA sequences by the breakage and rejoining of chromosomes or chromosome segments...a programmed feature of meiosis in most sexual organisms...” (Carroll 2013)

➤ Produced by chromosomal crossover and independent assortment process.

➤ Produces offspring with traits differing from parents' (preventing production of clones).

Source of genetic variation; promotes genetic diversity in a species.

Genetic variation through recombination can be much faster than through mutation.

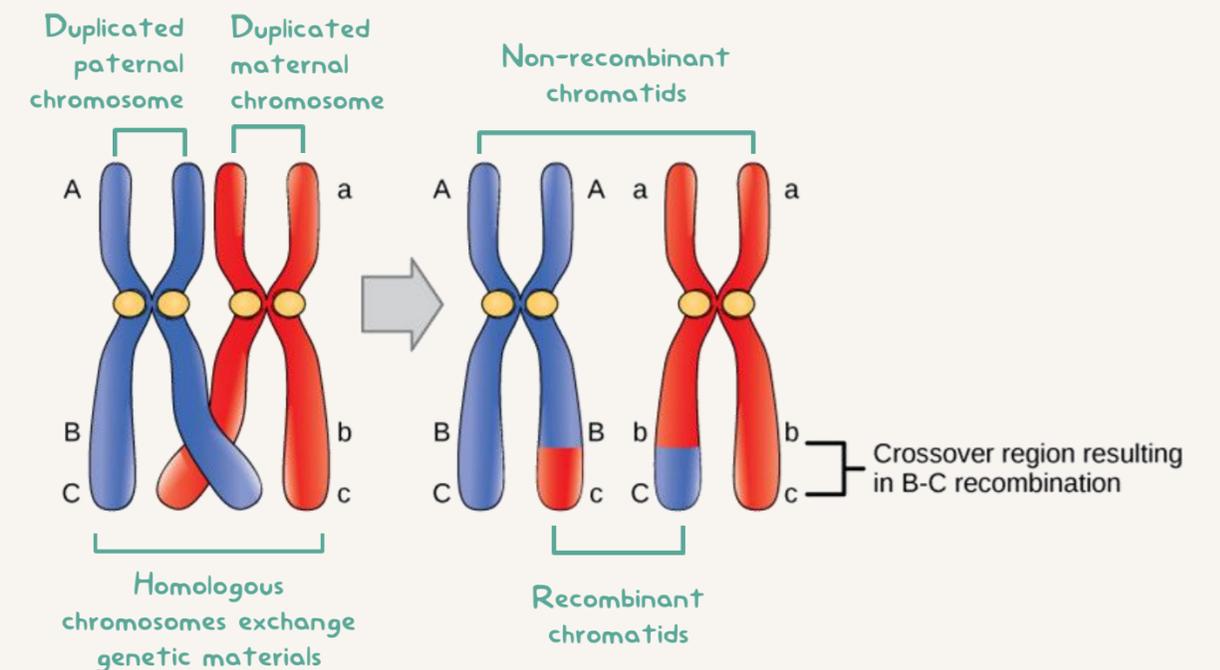
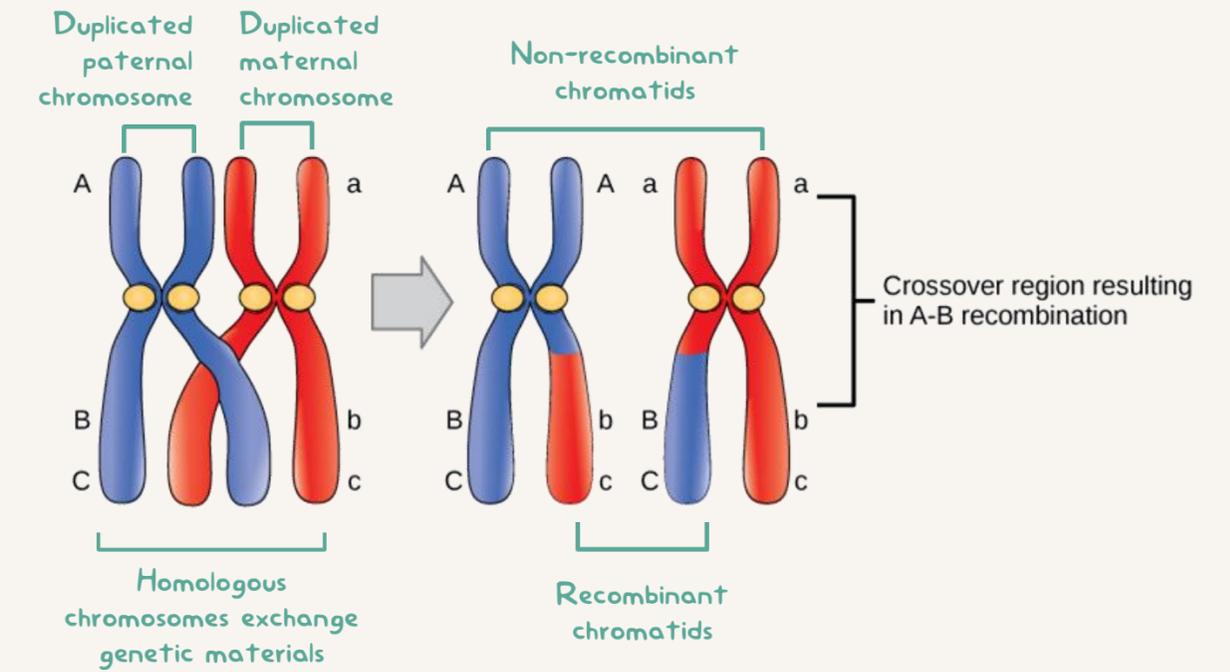
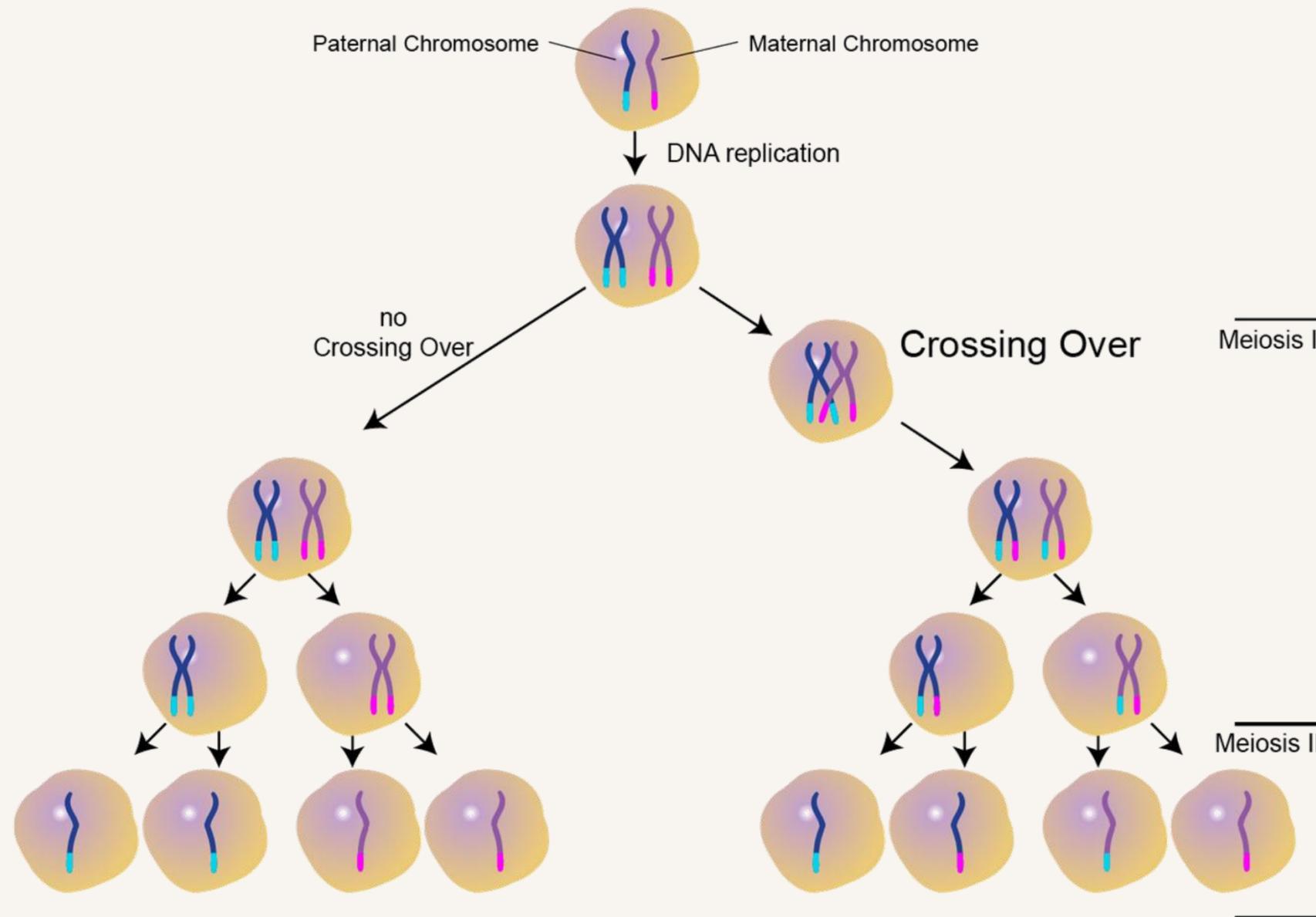
ARS-USDA, Wikimedia Commons, Public Domain



The silverleaf whitefly, *Bemisia tabaci*, experienced a rare gene crossover event between plant and animal. The gene it acquired from plants helps neutralize the anti-herbivory toxin produced by the plants it eats.

(Nature, 2021, <https://doi.org/10.1038/d41586-021-00782-w>)

# Schematic of genetic recombination



# Causes of loss in genetic variation

04

# Mechanisms that decrease genetic variation

Evolution: a change in the frequency of alleles in a population over time.

The change may or may not produce a better-suited population for its environment.

Alleles may be added into a population, but may also be lost from it during the process, reducing the variation.

Main causes of the loss of genetic variation in a population

Genetic drift

1. Random genetic drift

2. Bottleneck effect

3. Founder effect

4. Selection

Nonrandom mating

1. Inbreeding

2. Outbreeding

3. Assortative mating

# Genetic drift

The change in the composition of a gene pool (i.e. allele frequencies) as a result of chance or random events.

➤ Happens naturally in all populations of all sizes.

➤ Change in allele frequency is rapid in small populations, slower in larger populations.

Some changes can be “neutral”, have no immediate consequence to the biology of the population.

The effects can be magnified by natural events leading to the bottleneck effect, or formation of a new population in founder effect.

Figure 1, Jambari et al. 2017. J Wildl Park 32: 79-83, used under a Fair Use rationale



A melanistic Malayan tapir *Tapirus indicus* var. *brevetianus* showing the lack of “saddle blanket”, the white region normally found in a tapir’s body, an example of genetic drift occurring in an animal.

# 1. Random (natural) genetic drift

Occurs in all populations, but has largest effect in small populations.

May cause random fixation of alleles and loss of heterozygosity.



Fixation: the increase of the frequency of a gene by genetic drift until no other allele is preserved in a specific finite population.

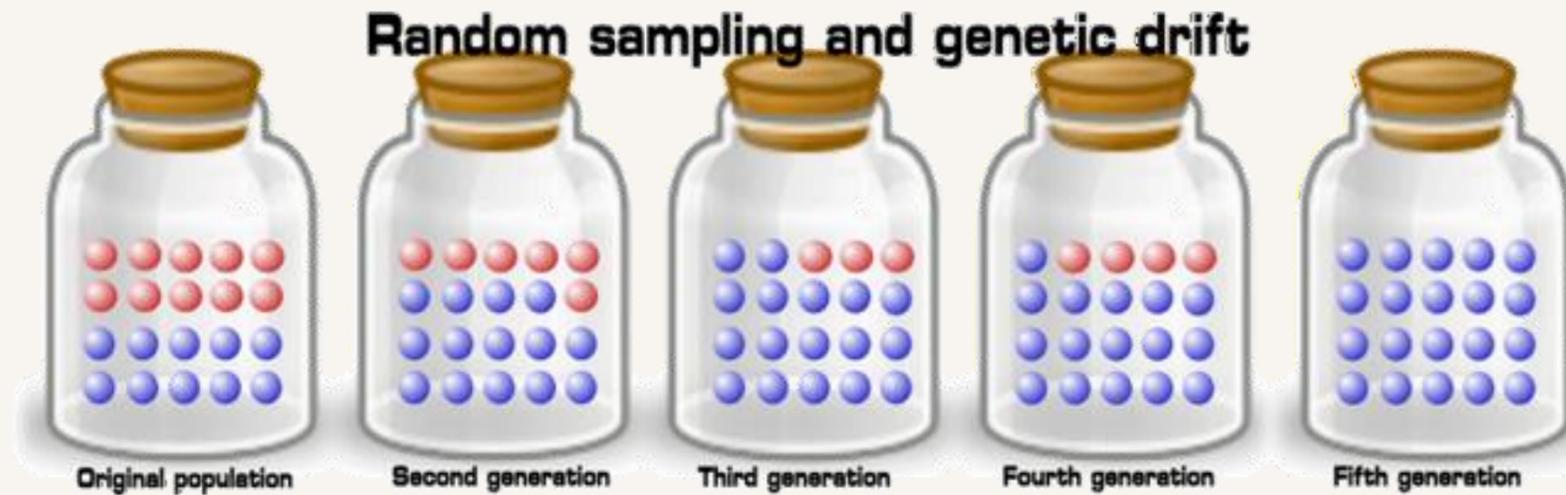
Does not differentiate between a deleterious and a beneficial allele.



Alleles of any trait have an equal chance of being lost or fixed.



# Random sampling in genetic drift



Professor marginalia, via  
Wikimedia Commons, CC BY-SA 3.0



5 rabbits reproduce

2 rabbits reproduce

White phenotype disappears completely from the population

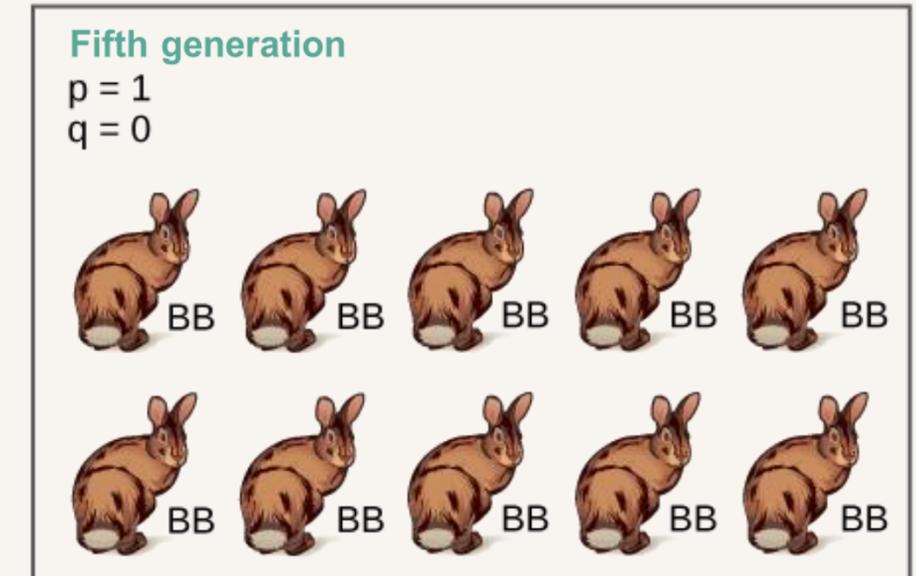
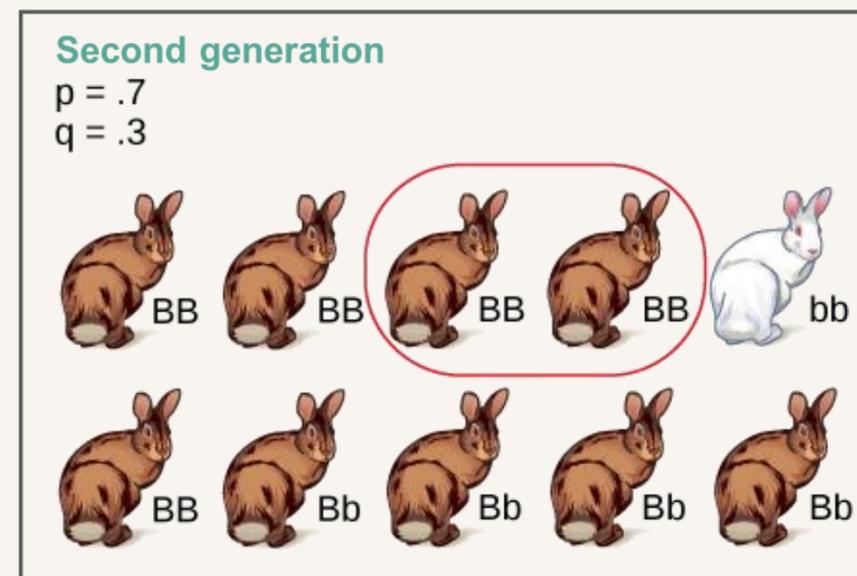
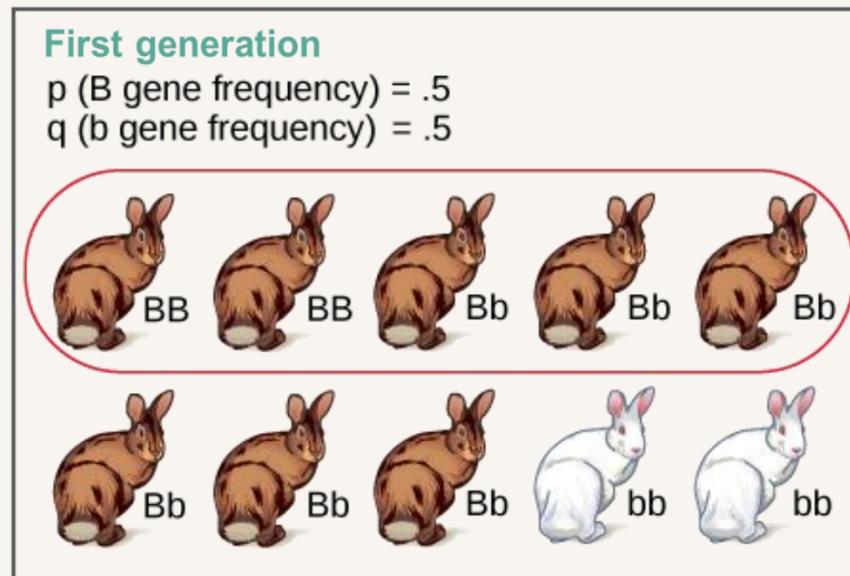
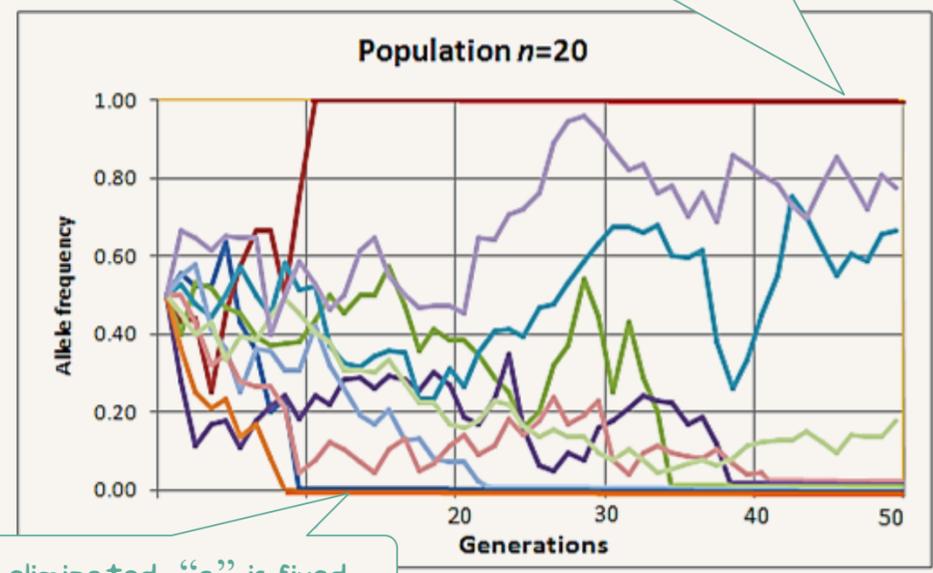


Figure 19.4 (with modifications), Rye et al., via OpenStax, CC BY 4.0

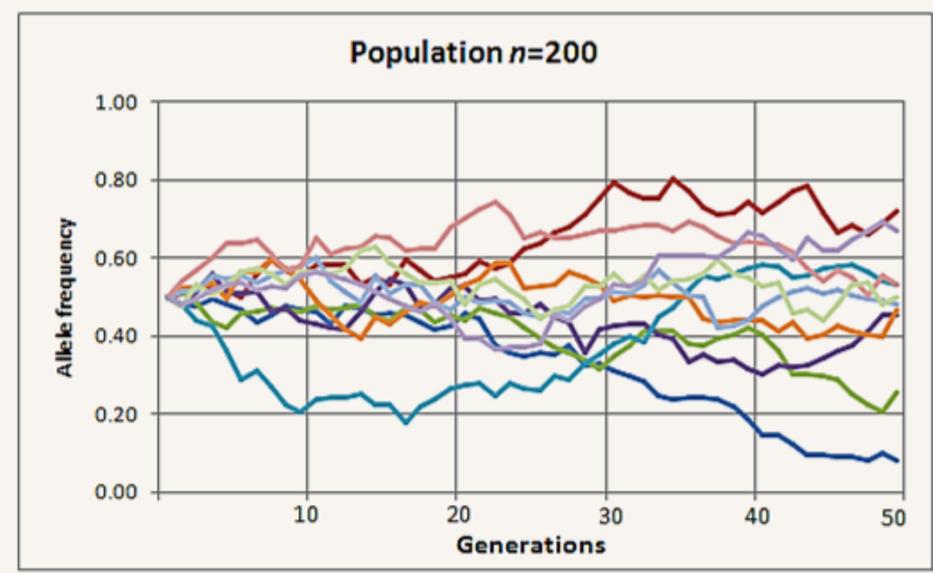
# Effect of population size on genetic drift

“a” is eliminated, “A” is fixed.

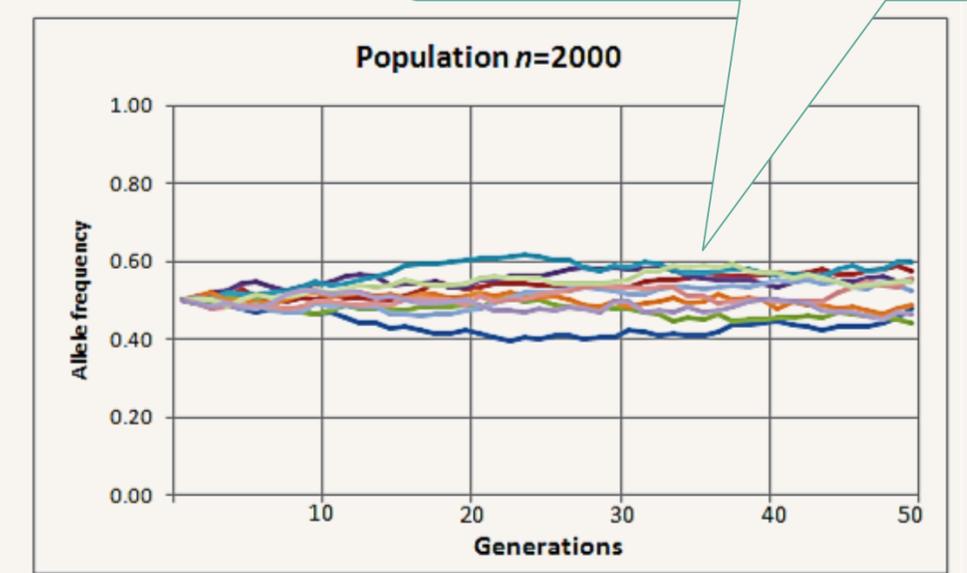


“A” is eliminated, “a” is fixed.

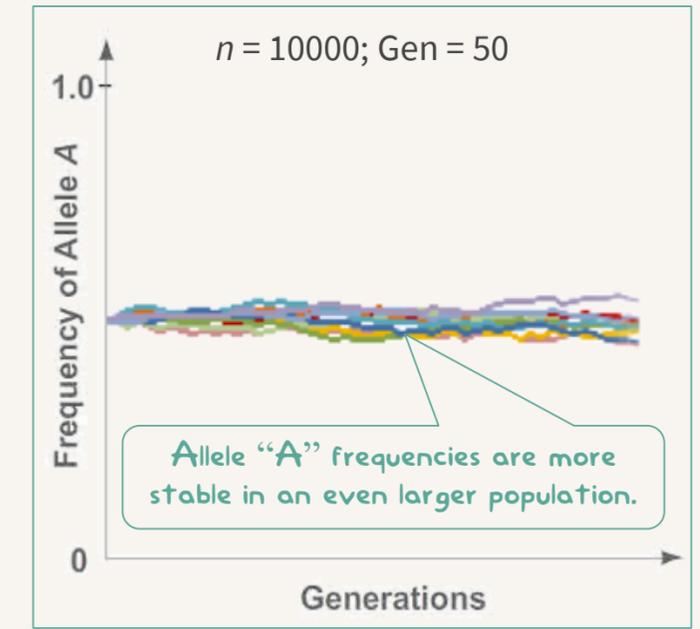
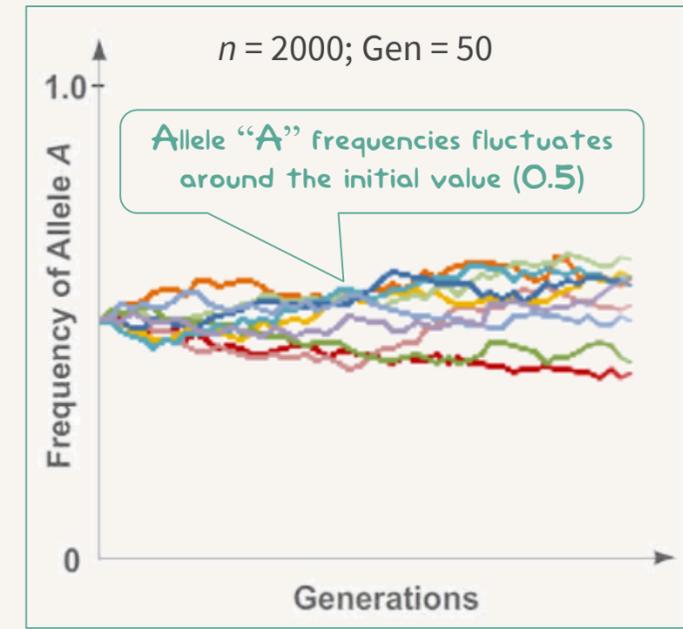
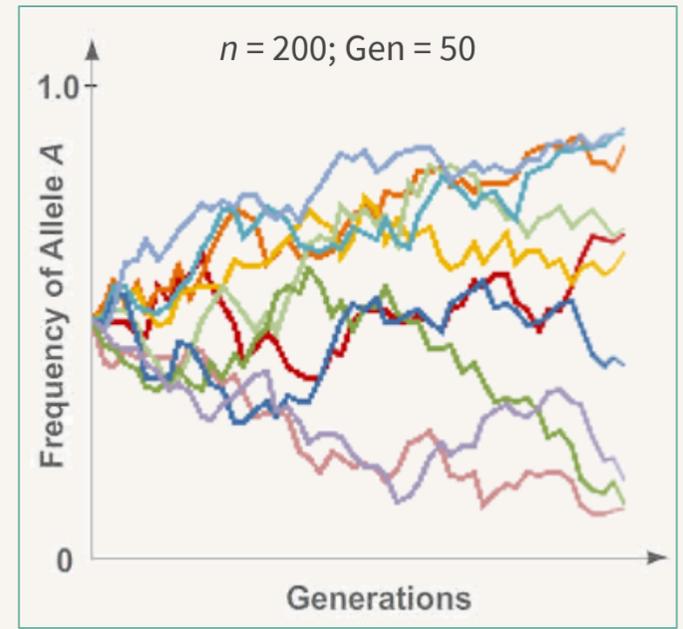
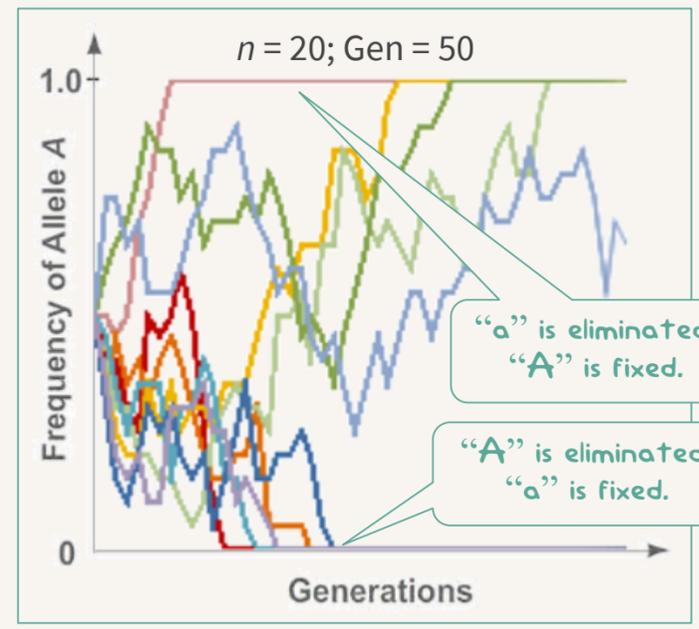
Professor marginalia, via Wikimedia Commons, CC BY-SA 3.0



Allele “A” frequencies fluctuates around the initial value (0.5)



Original population has two alleles, A and a, at a particular gene locus, with an equal frequency of 0.5 each. The allele A is simulated for a random change in the frequency distribution over 50 generations for different sized populations, with ten simulations each. Sources: top charts from Wikimedia Commons; bottom charts are results from the Genetic Drift Simulation program at <https://www.whfreeman.com>, with the same initial values.



# Fixation of alleles

Random fluctuation of allele frequencies may eventually lead an allele to reach a zero frequency and is permanently eliminated.

Consequently, the other allele is “fixed” in the population.



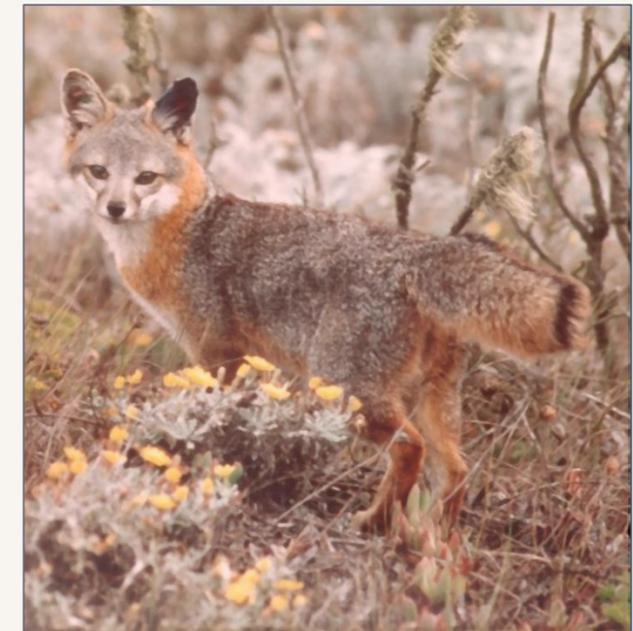
Individuals in the population will be homozygous for this allele in the future.

Rate of allele fixation is influenced by population size.



Fixation in a larger population takes a longer time.

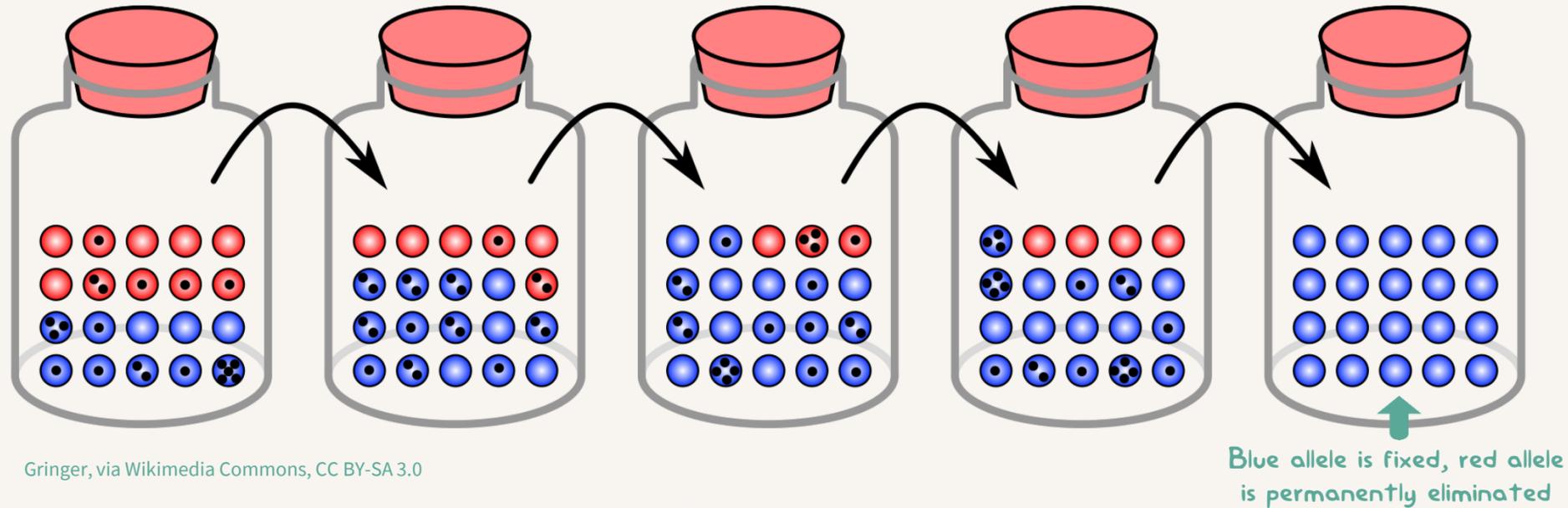
NPS-USDI, via Wikimedia Commons, Public Domain



Kristian Peters, via Wikimedia Commons, CC BY-SA 3.0

Island Fox *Urocyon littoralis* in the Channel Islands, California, USA (top) and the Apollo butterflies *Parnassius apollo* (bottom), examples of monomorphism for the same fixed allele (high genetic similarity).

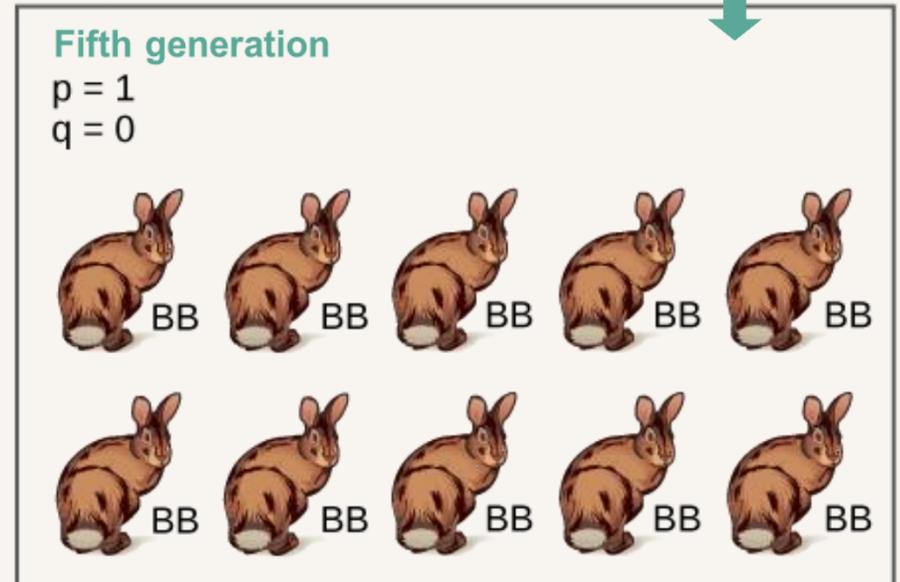
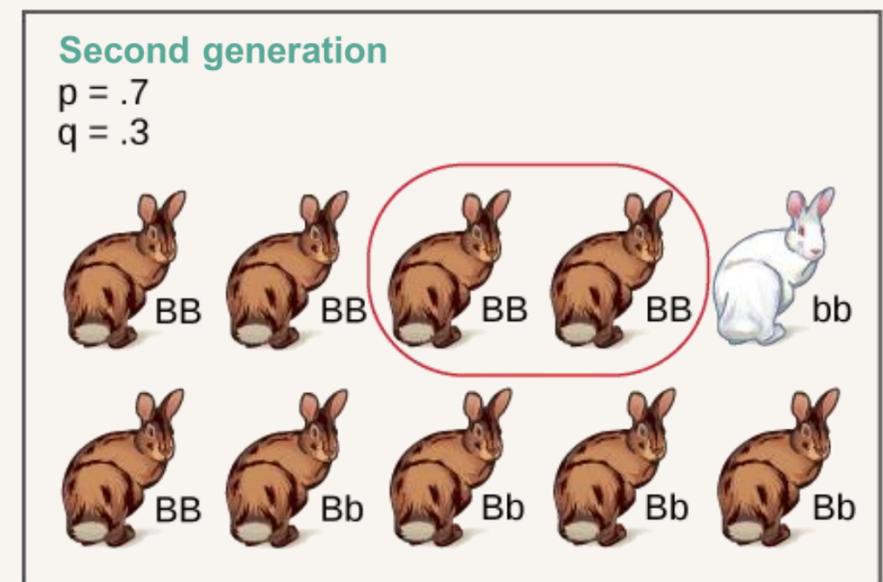
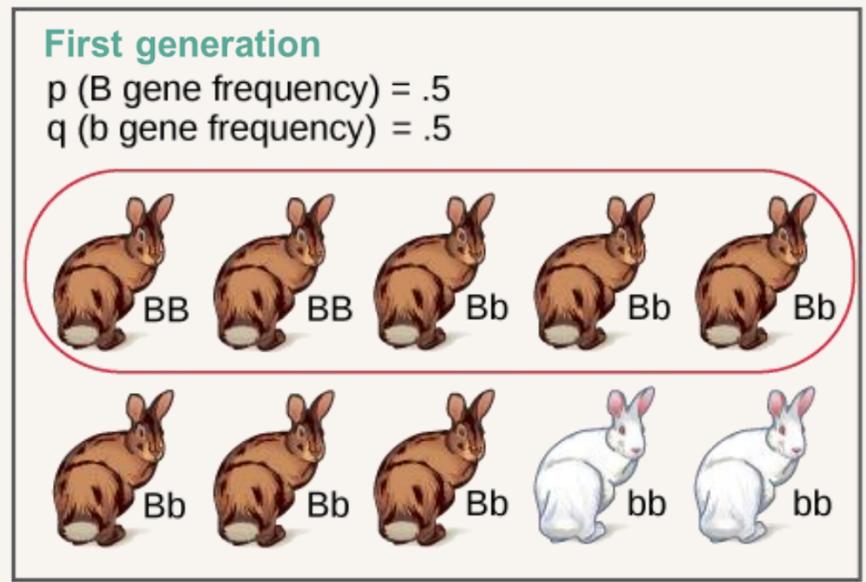
# Allele fixation



Gringer, via Wikimedia Commons, CC BY-SA 3.0

## Fixation event

- The blue allele (left) and allele **B** in the rabbit population (below) are “fixed” after 5 generations.
- All individuals in the population become permanently homozygous for that particular trait.
- The genetic diversity decreases (allele **b** is eliminated).



*B* allele is fixed, *b* allele is permanently eliminated

Figure 19.4 (with modifications), Rye et al., via OpenStax, CC BY 4.0

## 2. Bottleneck effect on a population

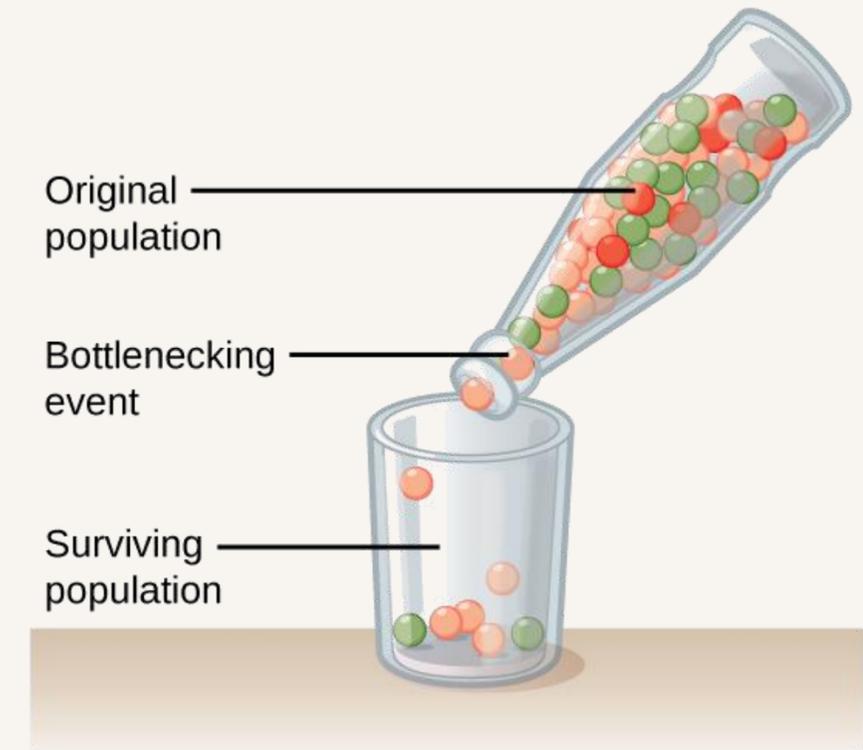
A bottleneck event increases the effect of genetic drift through a drastic reduction of a population size.

Occurs when an entire population is wiped out by some catastrophic event, except for a small group of survivors.

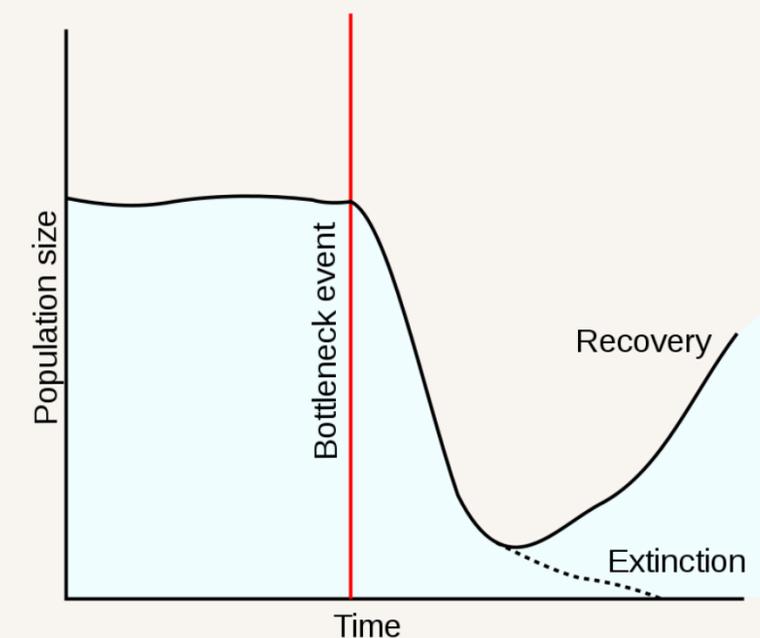
➤ Environmental events, e.g. droughts, fires, floods, diseases.

➤ Human activities, e.g. culling, poaching, chemical application.

Allele frequencies in the subsequent population is determined by the those in the surviving individuals.



OpenStax/Rice University, via Wikimedia Commons, CC BY 4.0

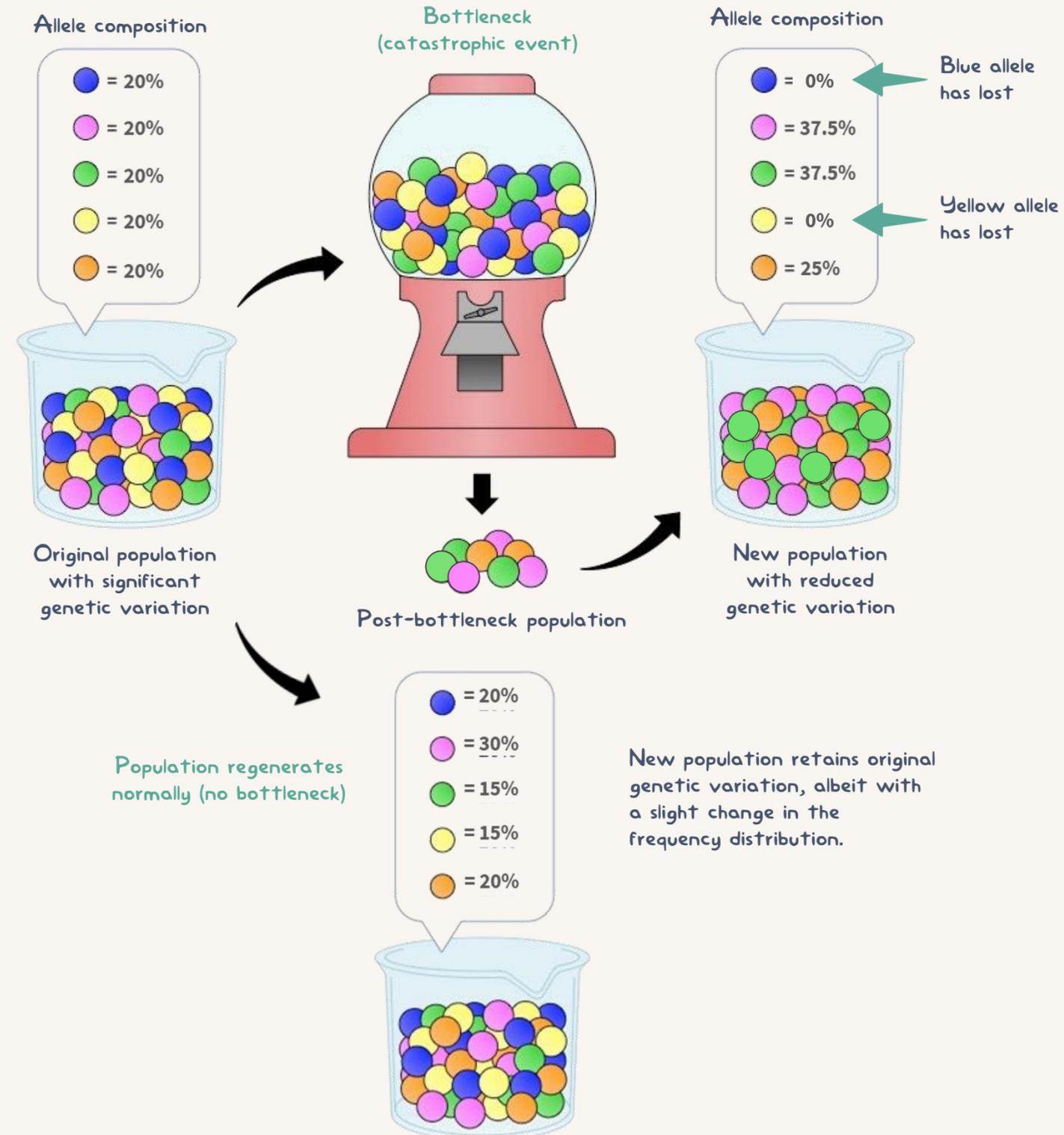


Mysid at English Wikipedia, via Wikimedia Commons, CC BY-SA 3.0

# Simulation of a population bottleneck

A population contains a gene pool consisting of equal numbers of five alleles.

A bottleneck event, such as a natural catastrophe, instantly wipes a large part of the population, significantly reducing its size.



The size reduction leaves the population with a gene pool consisting of only three alleles, as the blue and yellow alleles disappear from the population together with the perishing individuals.

The post-bottleneck population returns to its original size, but the blue and yellow alleles have permanently disappeared from the population, reducing its genetic variation.

# Bottleneck effect in nature

## Human population bottleneck



NASA Landsat7 image, via Wikimedia Commons, Public Domain

The Youngest Toba eruption,  $\pm 75,000$  BP, has been claimed to have triggered a major environmental change, causing a bottleneck of the human population, reducing it to  $< 10,000$  individuals.

## Northern elephant seal



"Mike" Michael L. Baird, via Wikimedia Commons, CC BY 2.0

*Mirounga angustirostris* was hunted almost to extinction in the 19<sup>th</sup> century. Its population bounced back after protection, but with a significant loss of genetic diversity in the surviving population, resulting in susceptibility to disease and environmental changes.

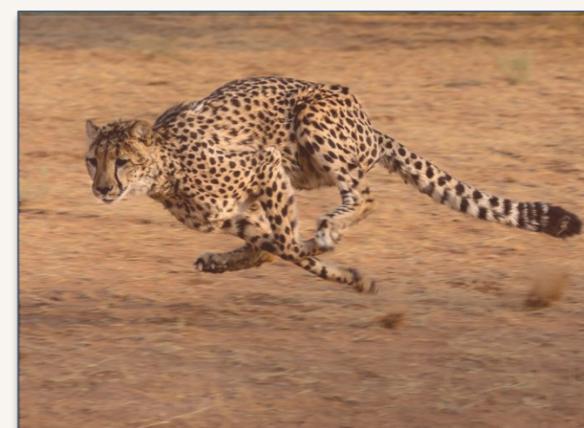
## Greater prairie chicken



Forbin42, via Wikimedia Commons, CC BY-SA 4.0

*Tympanuchus cupido's* population drastically declined to around 50 as the result of hunting and habitat loss, causing a severe loss in its genetic diversity.

## Cheetah



DrZoltan, via Pixabay, Pixabay license

The cheetah *Acinonyx jubatus* has survived two bottleneck events ( $\sim 100,000$  BP and  $\sim 12,000$  BP) that greatly decreased its genetic variability, resulting in a high juvenile mortality and susceptibility to diseases and infections.

# Bottleneck effect in nature

## Mauna Kea silversword



Crinthecity, via Wikimedia Commons, CC0

*Argyroxiphium sandwicense* ssp. *sandwicense*, a highly endangered flowering plant endemic to Hawaii, experienced a population bottleneck in the 1970s through its conservation efforts. Outplanted individuals were offspring of just two maternal founders with a low amount of polymorphic loci, resulting in an atypical morphology and reproduction.

## Wollemi pine



Meneerke bloem, via Wikimedia Commons, CC BY-SA 3.0

Discovered in 1994, *Wollemia nobilis* has an incredibly low genetic diversity. Fewer than a hundred trees are known to be growing in the wild, and all are genetically indistinguishable, suggesting that the species has been through a severe genetic bottleneck, that all genetic variability was lost.

## The Ngorongoro Crater lions

A population bottleneck in 1962 caused by *Stomoxys* biting flies crashed the population of lions *Panthera leo* in the Ngorongoro Crater, Tanzania, leaving 6–15 survivors. The crater itself provides isolation, giving the lions little opportunity to mate with unrelated partners.

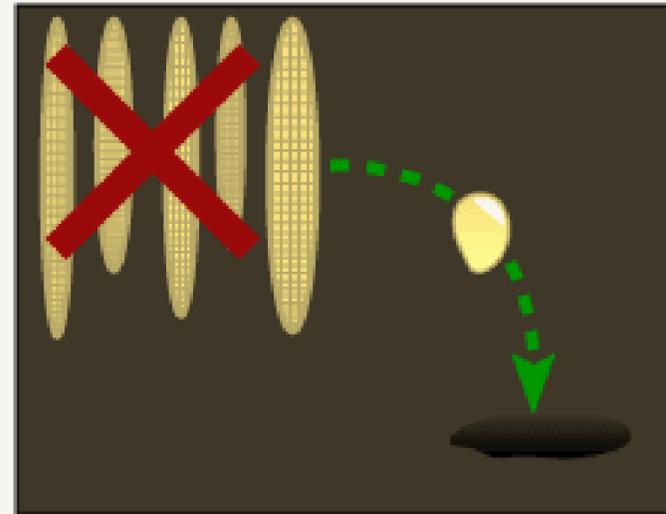


Michelle Maria, via Wikimedia Commons, CC BY 3.0

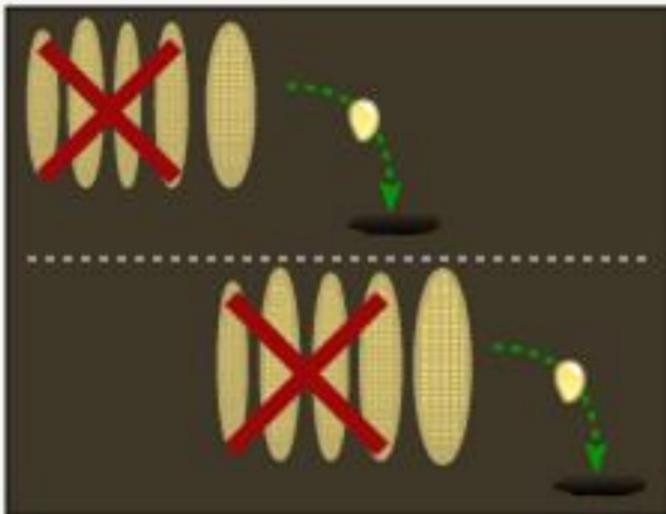
# Bottleneck effect in selective breeding



1. Natural variation occurs in the wild population.



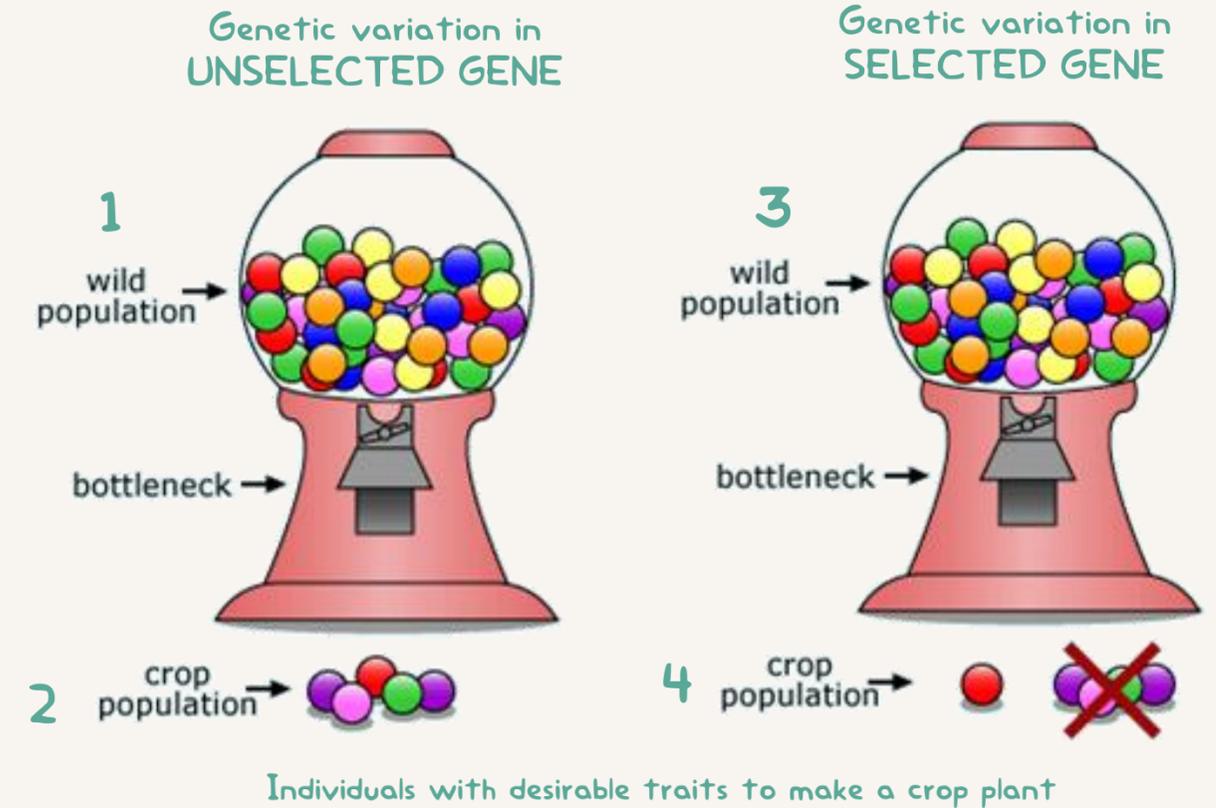
2. Seeds for the next generation are chosen only from individuals with the most desirable traits.



3. Repeat this process for several generations.



4. Over time, the quality of the crop increases.



Depiction of plant domestication through artificial selection.

The modern corn evolved from teosinte, a wild grass, through artificial selection performed by humans.



John Doebley, via Wikimedia Commons, CC BY 3.0

# Bottleneck effect in selective breeding

Igor Lukin, via Pixabay, Pixabay license



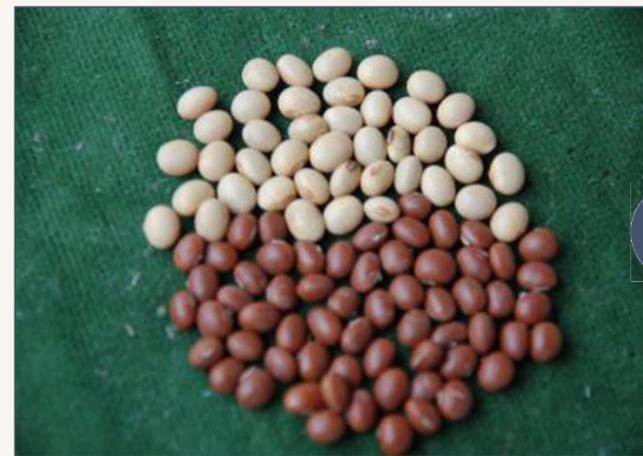
J. Warner, WikiCommons, (CC BY-SA 2.0)



Ellen Levy Finch, via Wikimedia Commons, CC BY-SA 3.0

**The Belgian Blue cow** is highly inbred due to the use of males from the same lineage to breed (Hanset et al. 2002) or the “popular sire effect”, and the fixation of a defective variant in the myostatin gene, which creates an exceptional muscular development, is sustained through linebreeding (Druet et al. 2014).

Yvonne Huijbens, via Pixabay, Pixabay license



Int Inst Trop Agric, via Flickr, CC BY-NC-SA 2.0

**Soybean** has experienced several genetic bottlenecks: domestication in Asia (highest impact), introduction of relatively few landraces to North America, and the past 75 years of selective breeding (Hyten et al. 2006)

**Dogs and cats** selective breeding causes constricting breed-specific bottlenecks, resulting in the emergence of breed-specific genetic disorders (e.g. eye diseases, joint problems, heart disease, hip dysplasia, respiratory problems, muscular atrophy).

# 3. Founder effect

The loss of genetic variation in a new, separate population established by only a small subset of a large population in a founder event.

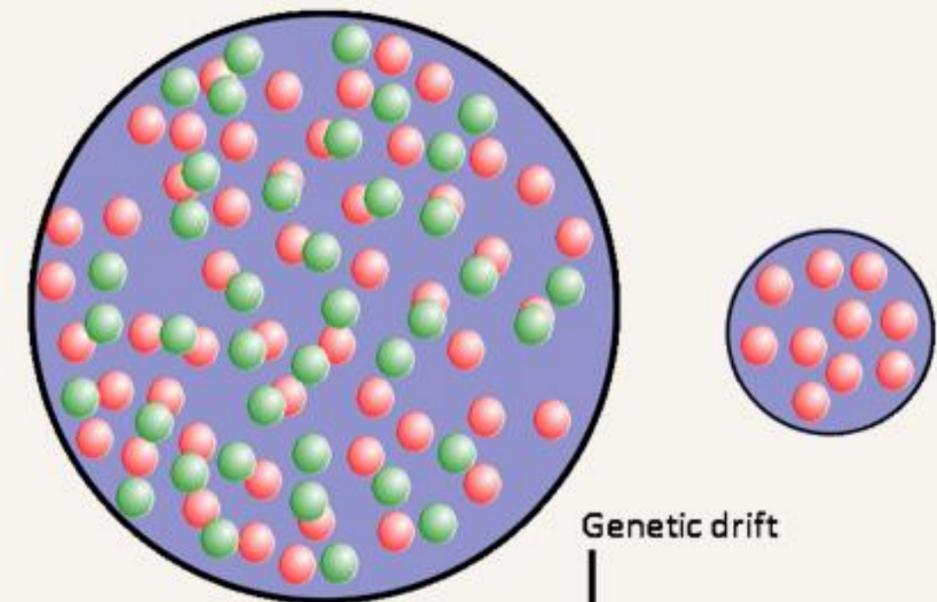
Occurs when a small group of individuals migrate to settle in a different location isolated from the origin.

Allele frequencies (genotypes & phenotypes) of the new population are determined by those in the founders.

Frequencies differ from those in the parent population (e.g. a rare allele may suddenly become prominent in the new population, and vice versa).

Originating population

Founder population



Genetic drift

Ancestral population frequencies		
	Red	Green
Original	0.50	0.50
After split	0.53	0.47
Generation 1	0.58	0.42
Generation 2	0.54	0.46
Generation 3	0.57	0.43
Generation 4	0.55	0.45
Generation 5	0.57	0.43

Founder population frequencies		
	Red	Green
After split	0.20	0.80
Generation 1	0.40	0.60
Generation 2	0.50	0.50
Generation 3	0.60	0.40
Generation 4	0.90	0.10
Generation 5	1.00	0.00

# Founder effect examples

## Ellis-van Creveld syndrome in the Amish population

The Amish population stemmed from their ~200-500 German founders migrating to Pennsylvania in the mid 18<sup>th</sup> century.

Amish people are moderately inbred, causing the prevalence of some recessive conditions, e.g. Ellis-van Creveld syndrome, a genetic disorder whose allele was traced back to a man and wife in the initial population.

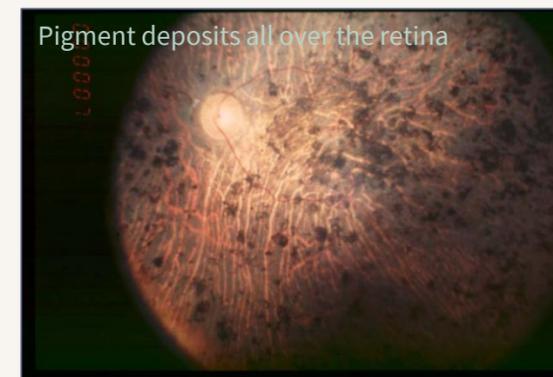
The founder effect and subsequent inbreeding resulted in 1 in 200 people homozygous for the allele; 1 in 8 is heterozygous.



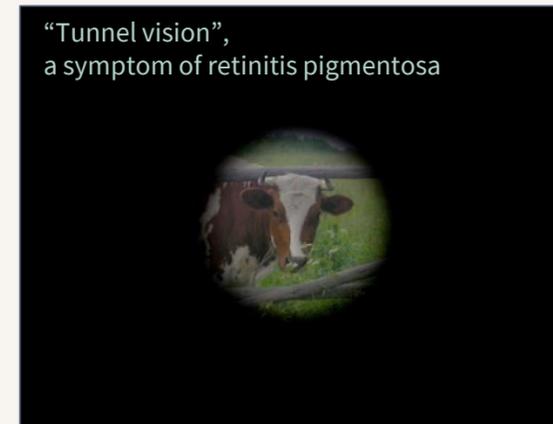
Ellis-van Creveld syndrome is characterized by dwarfism, heart defects, and polydactyly.

[Fig. 16-7], Audesirk et al. 2017. "Biology Life on Earth", used under a Fair Use rationale

## Retinitis pigmentosa in the Tristan da Cunha islands immigrants



Christian Hamel, via Wikimedia Commons, CC BY 2.0



Скампецкий, via Wikimedia Commons, CC BY 3.0



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Location of Tristan da Cunha islands

A number of individuals migrated to the Tristan da Cunha islands in the 1800s; at least one of them carried a recessive allele for retinitis pigmentosa, a rare eye disorder.

The estimated prevalence in the 1960s: of 240 residents, 4 had the disorder (homozygous), and at least 9 were carriers (heterozygous).

Retinitis pigmentosa causes loss of vision, with symptoms including trouble seeing at night, decreased peripheral vision, followed by "tunnel vision".

# 4. Selection

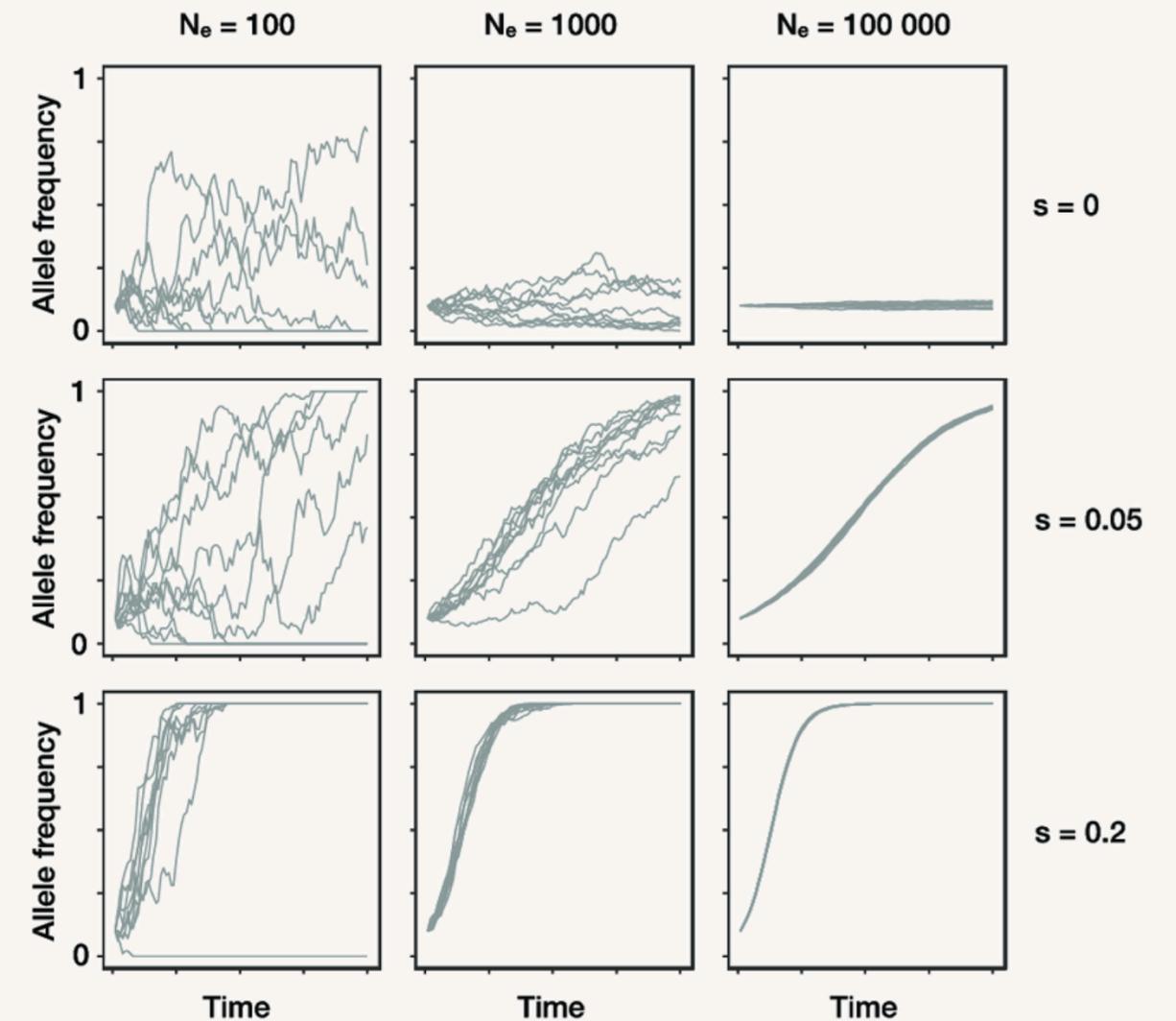
Selection occurs when certain traits are more or less likely to increase fitness and survival.

➔ **Selection in favor** = trait helps individuals survive and have offspring.

➔ **Selection against** = trait interferes with survival and reproduction.

The differences in fitness are expressed as changes in allele frequency over time.

Genotype difference contributes to organisms' difference in fitness.



Simulation of the combined effects of drift and selection on genetic change over time of a focal allele for different combinations of effective population size  $N_e$  and selection coefficient  $s$ .  $s$ -value  $> 0$  indicates a selective advantage of the focal allele;  $s = 0$  both alleles are neutral (changes are only due to genetic drift). Ten independent replicates, initial frequency for the focal allele = 0.1.

# Nonrandom mating

Mating patterns are important in the maintenance of genetic equilibrium in a population.

➔ **Random:** all individuals have an equal chance of being selected as a mate; no discrimination for or against any trait.

➔ **Nonrandom:** individuals do not have an equal chance of being selected; there is discrimination for or against certain trait.

Nonrandom mating upsets the Hardy-Weinberg genetic equilibrium of a population.

## Forms of nonrandom mating

Based on relatedness

Inbreeding

Outbreeding

Based on traits preference

Positive assortative mating

Negative assortative mating

Contributes to the natural selection process in evolution.

# Inbreeding

Occurs when individuals with similar genotypes (i.e. close relatives) are more likely (higher probability) to mate with each other.

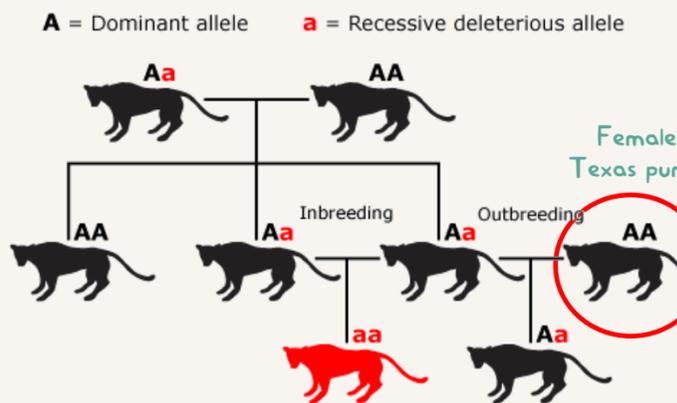
**Fact:** organisms seldom mate strictly randomly (e.g. limited mobility, inclination to stay near birthplace).

Probability of occurrence is much higher in a small population and following a bottleneck/founder event.

**Inbreeding depression:** a decline of reproductive fitness in a population as a result of inbreeding.

**Results:** a decrease of heterozygosity and fecundity, increase of mortality (see *Bottleneck & Founder effect* sections).

Connie Bransilver, via Flickr, CC BY 2.0



UC Museum of Paleontology, evolution.berkeley.edu, Fair Use

## Inbreeding depression in Florida panthers *Puma concolor c cougar*

Nearly extinct in the early 20<sup>th</sup> century (as little as six individuals left), the genetic bottleneck and subsequent inbreeding is thought to be responsible for the lower overall fitness in the current population. Genetic defects include undescended testicles, poor sperm quality, heart problems, kinked tails, cowlicks.

Florida panther genetic status improvement through outcrossing with different puma subspecies.



Dave Onorato/US-FWS, via www.fws.gov, used under a Fair Use rationale

# Oubreeding

Occurs when the probability of mating between distant relatives is higher than with close relatives.



Matings between individuals from different populations, subspecies, or species; less common.

**Results:** increased genetic variation (increased reproductive fitness) or outbreeding depression.



Many organisms possess mechanisms to promote outbreeding (e.g. dioecy, self-incompatibility).



**Outbreeding depression:** a decline of reproductive fitness in a population as a result of outbreeding (e.g. with chromosomal incompatibilities).

Mechanisms causing outbreeding depression

Genetic mechanism

Ecological mechanism

Incompatibility in coadapted gene complexes.

Different adaptation to different environmental conditions.

Outbreeding depression appears to be more common in plants (organisms with extremely limited dispersal).

Outbreeding can be useful in the restoration of genetic variation (see Florida panther).

# Assortative mating

Occurs when organisms discriminately choose their mate based on phenotypic traits (including sexual selection).

## Positive assortative mating (Assortative mating)

Choosing phenotypic traits  
SIMILAR to themselves.

Parents' genotypes	Expected offspring genotypes		
	<i>RR</i>	<i>Rr</i>	<i>rr</i>
<i>RR</i> X <i>RR</i>	4		
<i>Rr</i> X <i>Rr</i>	1	2	1
<i>rr</i> X <i>rr</i>			4
Total	5	2	5
<b>Proportion</b>	<b>0.42</b>	<b>0.17</b>	<b>0.42</b>

**Result:** a significant increase in homozygous genotypes (*AA*, *aa*).

## Random mating

Parents' genotypes	Expected offspring genotypes		
	<i>RR</i>	<i>Rr</i>	<i>rr</i>
<i>RR</i> X <i>RR</i>	4		
<i>RR</i> X <i>Rr</i>	2	2	
<i>RR</i> X <i>rr</i>		4	
<i>Rr</i> X <i>RR</i>	2	2	
<i>Rr</i> X <i>Rr</i>	1	2	1
<i>Rr</i> X <i>rr</i>		2	2
<i>rr</i> X <i>RR</i>		4	
<i>rr</i> X <i>Rr</i>		2	2
<i>rr</i> X <i>rr</i>			4
Total	9	18	9
<b>Proportion</b>	<b>0.25</b>	<b>0.50</b>	<b>0.25</b>

**Result:** homozygous and heterozygous genotypes = 50:50.

## Negative assortative mating (Disassortative mating)

Choosing phenotypic traits  
DIFFERENT from themselves.

Parents' genotypes	Expected offspring genotypes		
	<i>RR</i>	<i>Rr</i>	<i>rr</i>
<i>RR</i> X <i>Rr</i>	2	2	
<i>RR</i> X <i>rr</i>		4	
<i>Rr</i> X <i>RR</i>	2	2	
<i>Rr</i> X <i>rr</i>		2	2
<i>rr</i> X <i>RR</i>		4	
<i>rr</i> X <i>Rr</i>		2	2
Total	4	16	4
<b>Proportion</b>	<b>0.17</b>	<b>0.67</b>	<b>0.17</b>

**Result:** a significant increase in heterozygous genotypes (*Aa*).

## Design credits

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# INTRODUCTION TO POPULATION GENETICS

Bagian dari kuliah Ekologi Populasi di Fakultas Biologi Universitas Gadjah Mada



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