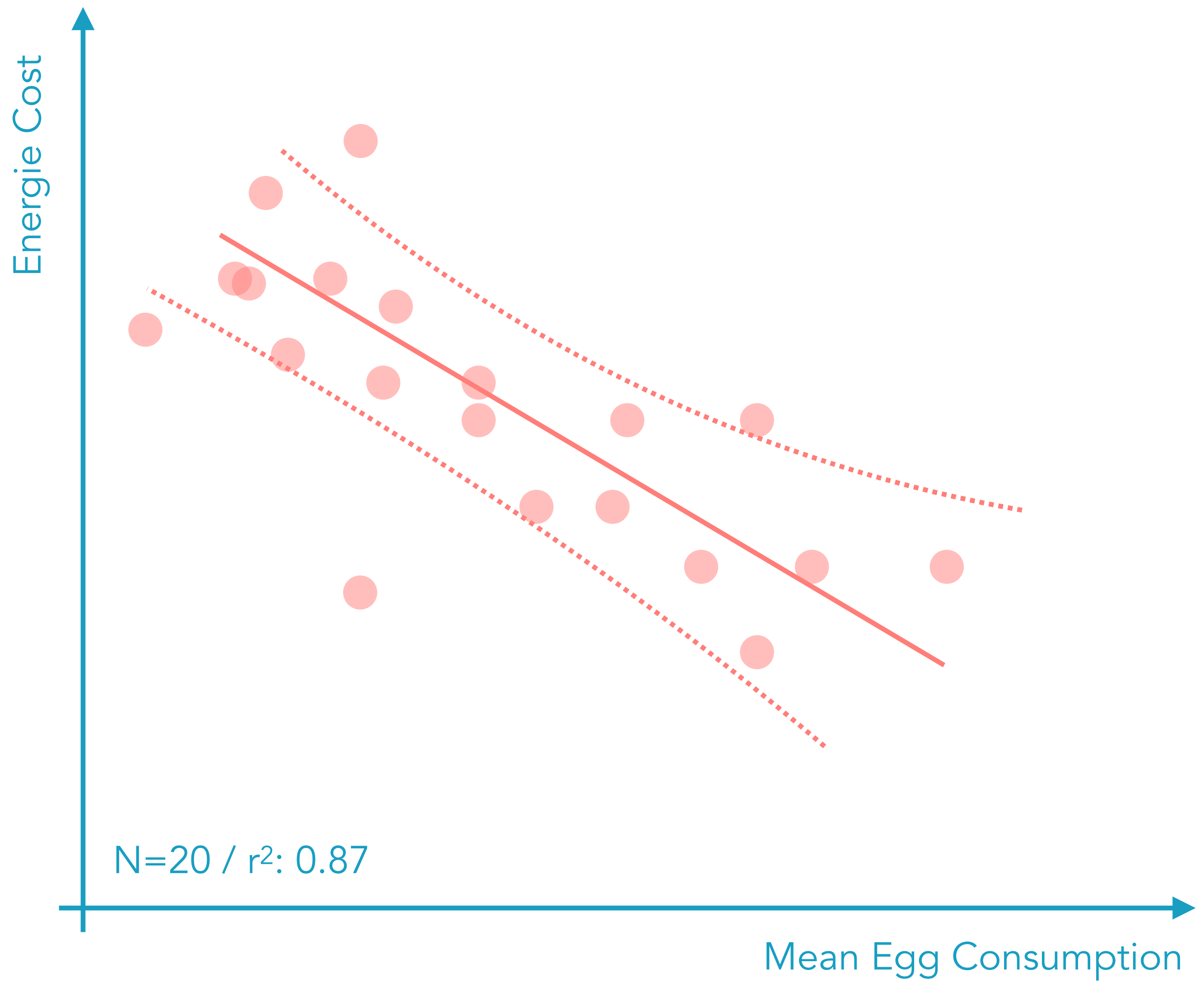
Evolutionary Genetics

LV 25600-01 | Lecture with exercises | 4KP

Why Population Genetics?

PopGen ▷ Bullet Points

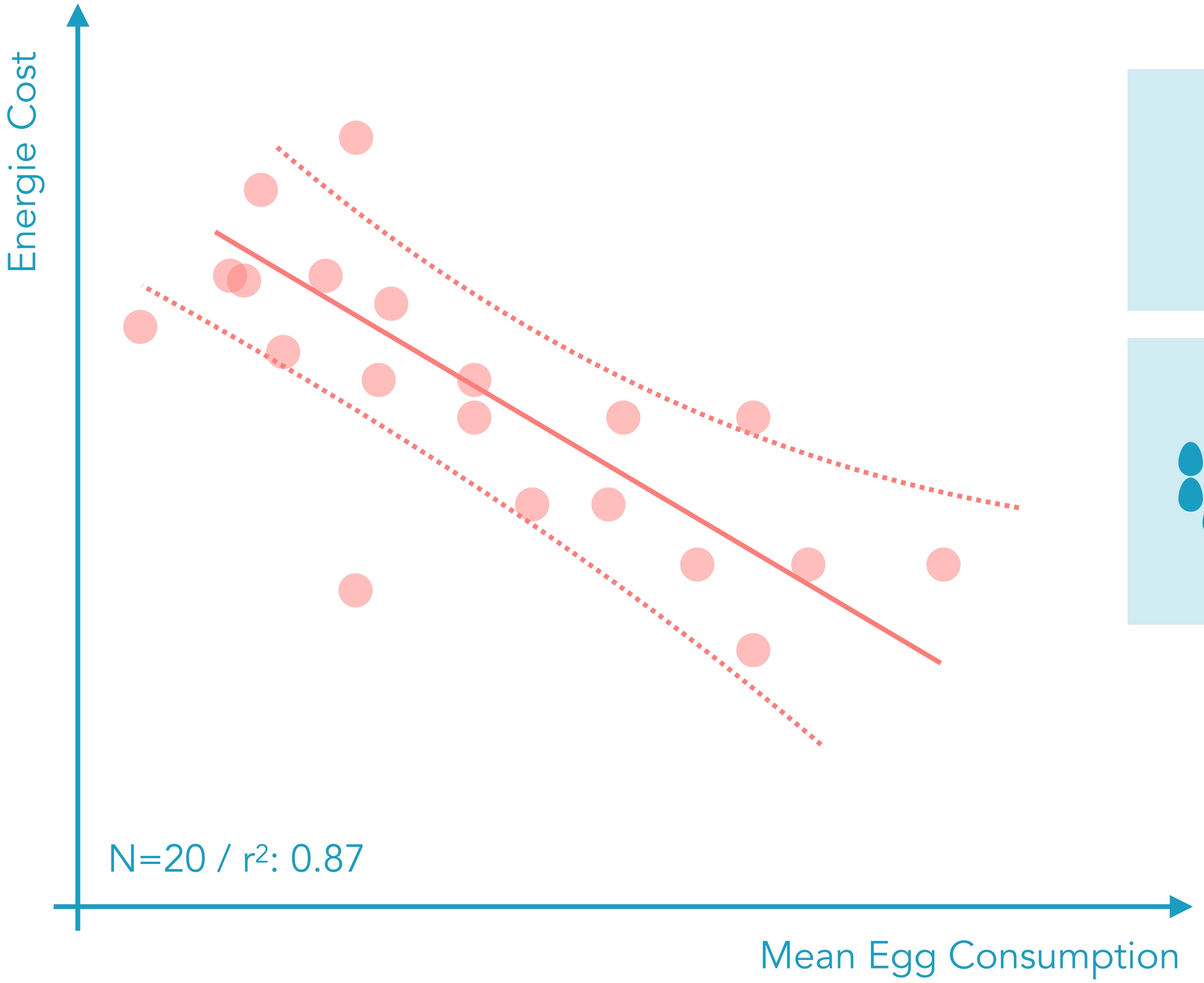


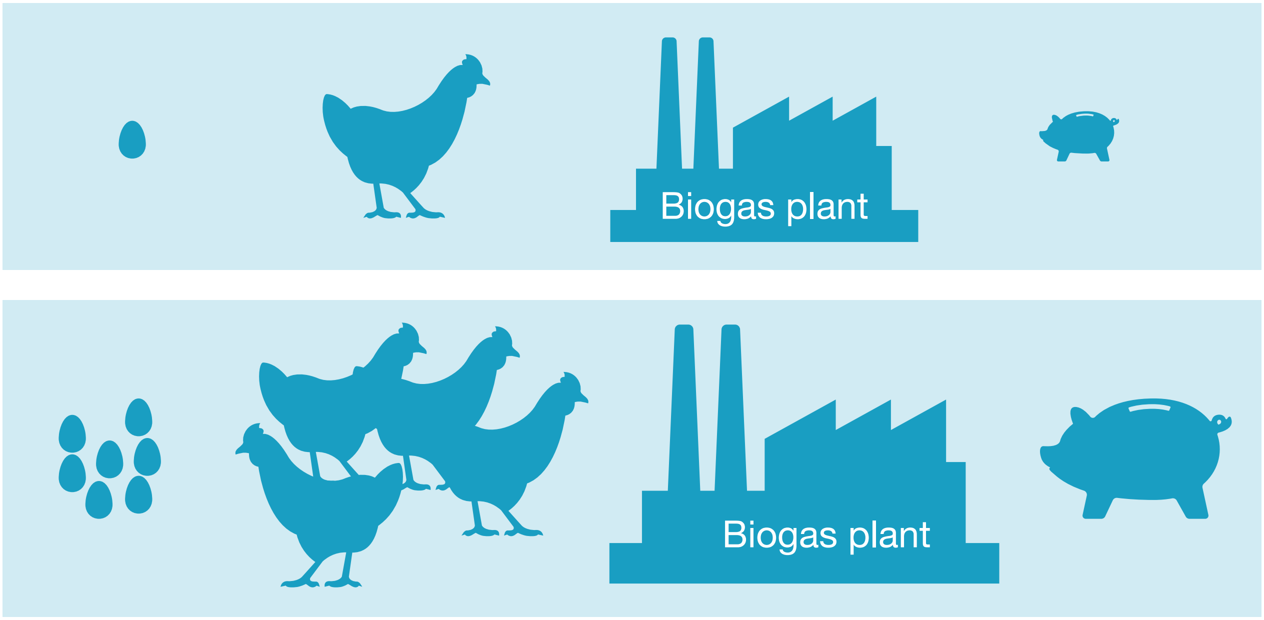
Egg consumption influences electricity prices

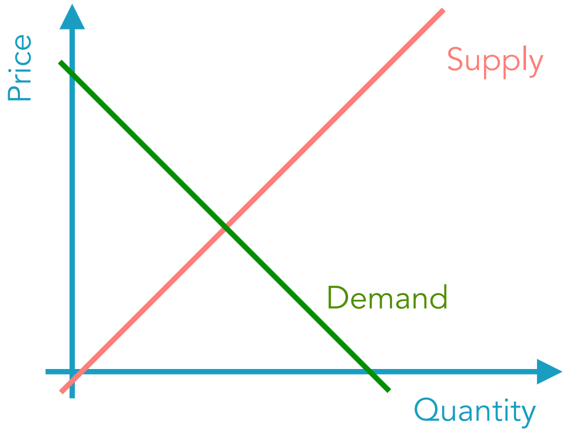
In this study, 60 households from 20 different regions were asked about their egg consumption. The average number of eggs consumed was compared with the local energy price. We found a significant negative correlation between egg consumption and energy price. The relationship between egg consumption and local electricity tariffs has not been described in this way before. Further research is needed to better understand this relationship.

Source: JC Walser (2022) Frontier in Correlations

PopGen ▷ Bullet Points







PopGen ▷ Bullet Points

The *Arabidopsis thaliana* mobilome* and its impact at the species level

Leandro Quadrana[1], Amanda Bortolini Silveira[1], George F Mayhew[2], Chantal LeBlanc[3], Robert A Martienssen[4,5], Jeffrey A Jeddeloh[2], Vincent Colot[1]*

eLife

* The **mobilome** is the entire set of mobile genetic elements in a genome.

Abstract - Transposable elements (TEs) are powerful motors of genome evolution yet a comprehensive assessment of recent transposition activity at the species level is lacking for most organisms. Here, using genome sequencing data for 211 *Arabidopsis thaliana* accessions taken from across the globe, we identify thousands of recent transposition events involving half of the 326 TE families annotated in this plant species. **We further show that the composition and activity of the 'mobilome' vary extensively between accessions in relation to climate and genetic factors.** Moreover, TEs insert equally throughout the genome and are rapidly purged by natural selection from gene-rich regions because they frequently affect genes, in multiple ways. Remarkably, loci controlling adaptive responses to the environment are the most frequent transposition targets observed. These findings demonstrate the pervasive, species-wide impact that a rich mobilome can have and the importance of transposition as a recurrent generator of large-effect alleles.

Quadrana *et al.* eLife 2016;5:e15716. DOI: 10.7554/eLife.15716

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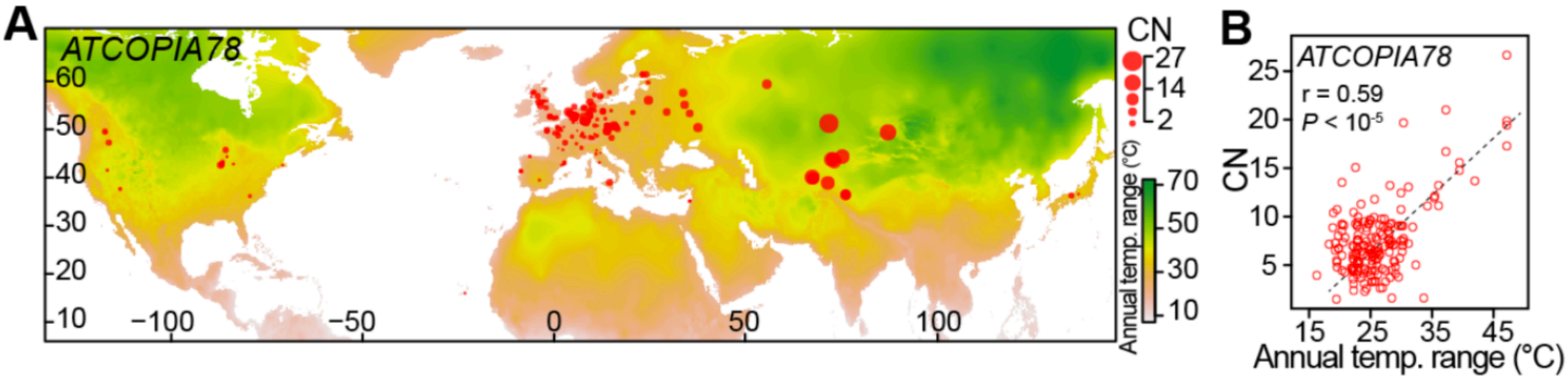

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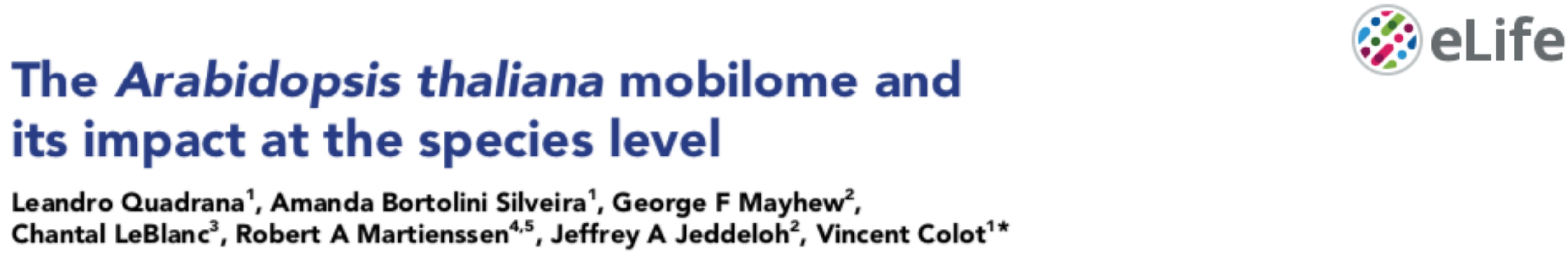
PopGen ▷ Bullet Points



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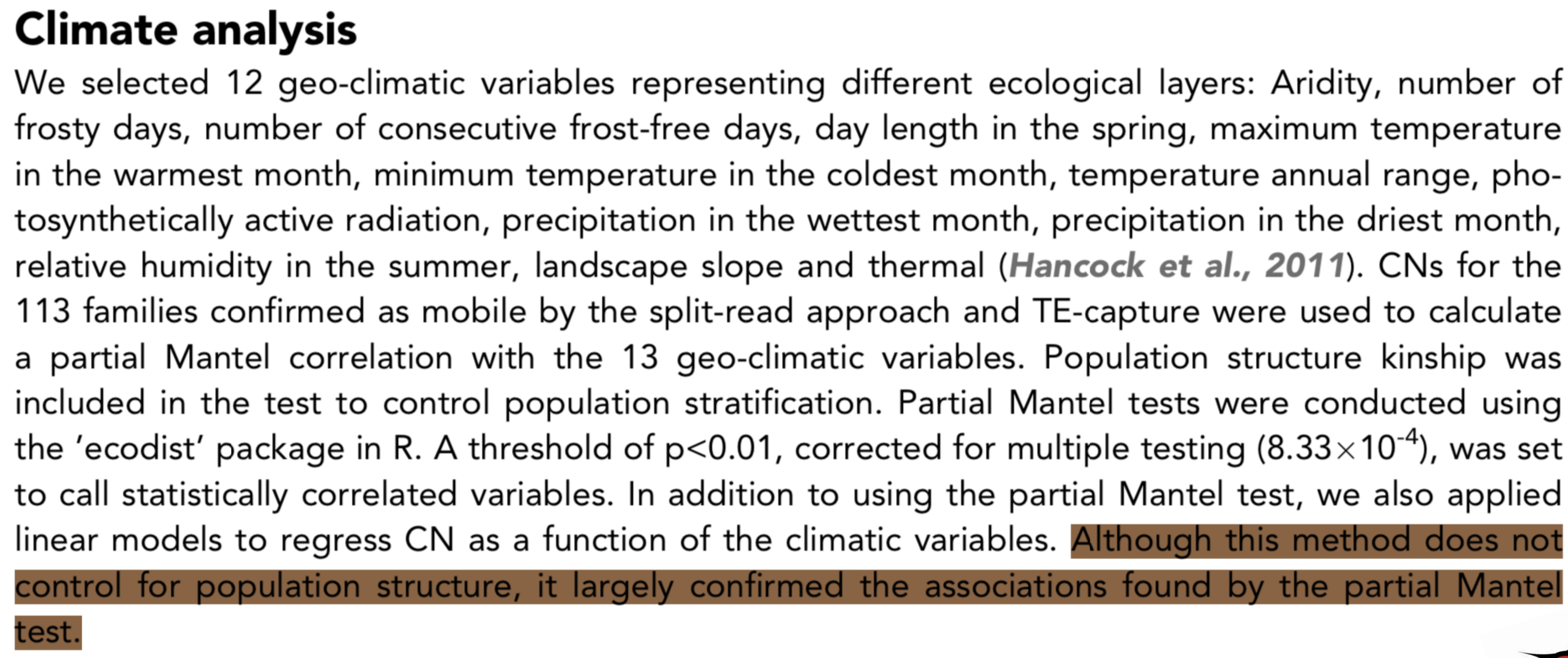

Climate analysis

We selected 12 geo-climatic variables representing different ecological layers: Aridity, number of frosty days, number of consecutive frost-free days, day length in the spring, maximum temperature in the warmest month, minimum temperature in the coldest month, temperature annual range, photosynthetically active radiation, precipitation in the wettest month, precipitation in the driest month, relative humidity in the summer, landscape slope and thermal (***Hancock et al., 2011***). CNs for the 113 families confirmed as mobile by the split-read approach and TE-capture were used to calculate a partial Mantel correlation with the 13 geo-climatic variables. Population structure kinship was included in the test to control population stratification. Partial Mantel tests were conducted using the 'ecodist' package in R. A threshold of $p<0.01$, corrected for multiple testing (8.33×10^{-4}), was set to call statistically correlated variables. In addition to using the partial Mantel test, we also applied linear models to regress CN as a function of the climatic variables. Although this method does not control for population structure, it largely confirmed the associations found by the partial Mantel test.

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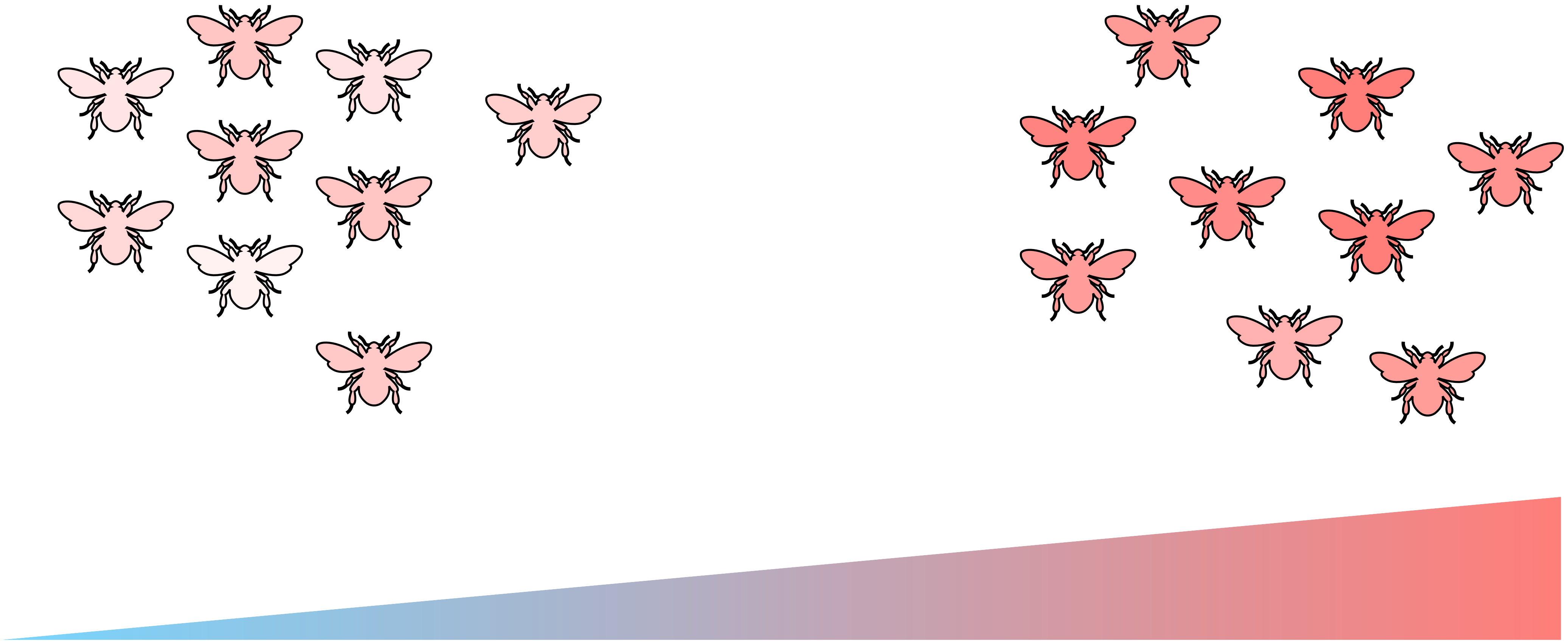
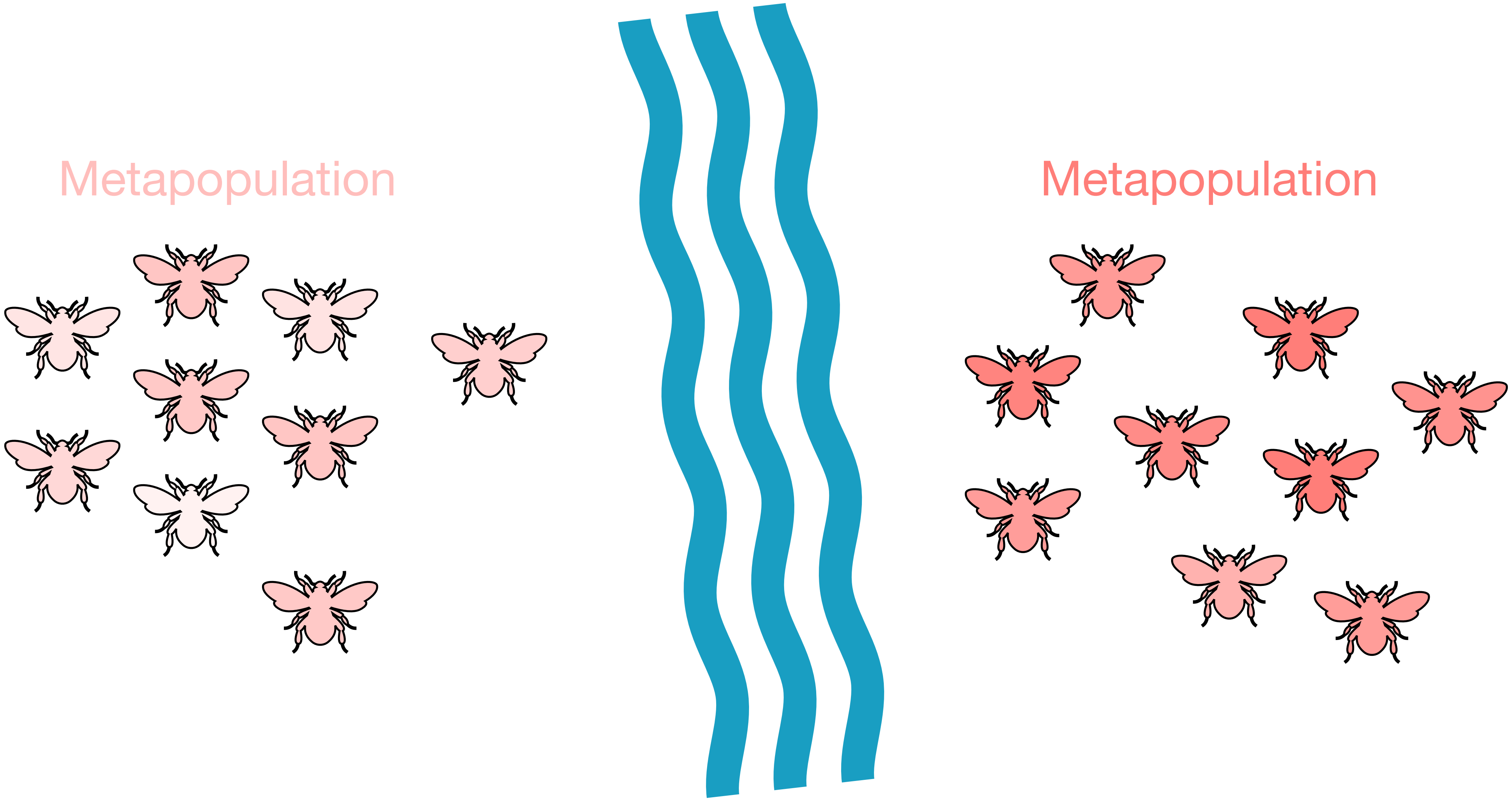
Population Differences

The composition vary extensively between populations in relation to climate factors.



Environmental Gradient

Population Structure



The better we understand a species, take into account the population structure and try to understand the factors that can influence it, the more relevant the interpretation of the results might be despite the fanciness of the method used.

PopGen ▷ Bullet Points

Sample	SNP1	SNP2	SNP3
S1	T	C	G
S2	T	C	G
S3	A	G	C
S4	A	G	C
S5	T	G	G
S6	T	G	G
S7	A	C	G

Genetic markers, such as single nucleotide polymorphisms (SNPs), are specific DNA sequences with known locations in the genome. They vary among individuals and are used to study genetic diversity, inheritance patterns, and evolutionary relationships. SNPs are particularly valuable due to their abundance, stability, and association with traits or diseases.

An alignment containing only SNPs is typically referred to as a SNP alignment or a variant alignment. It consists of nucleotide sequences for multiple samples or taxa, reduced to positions that show variation (polymorphisms) across the dataset. Such alignments are often used in population genetics, phylogenetics, and comparative genomics.

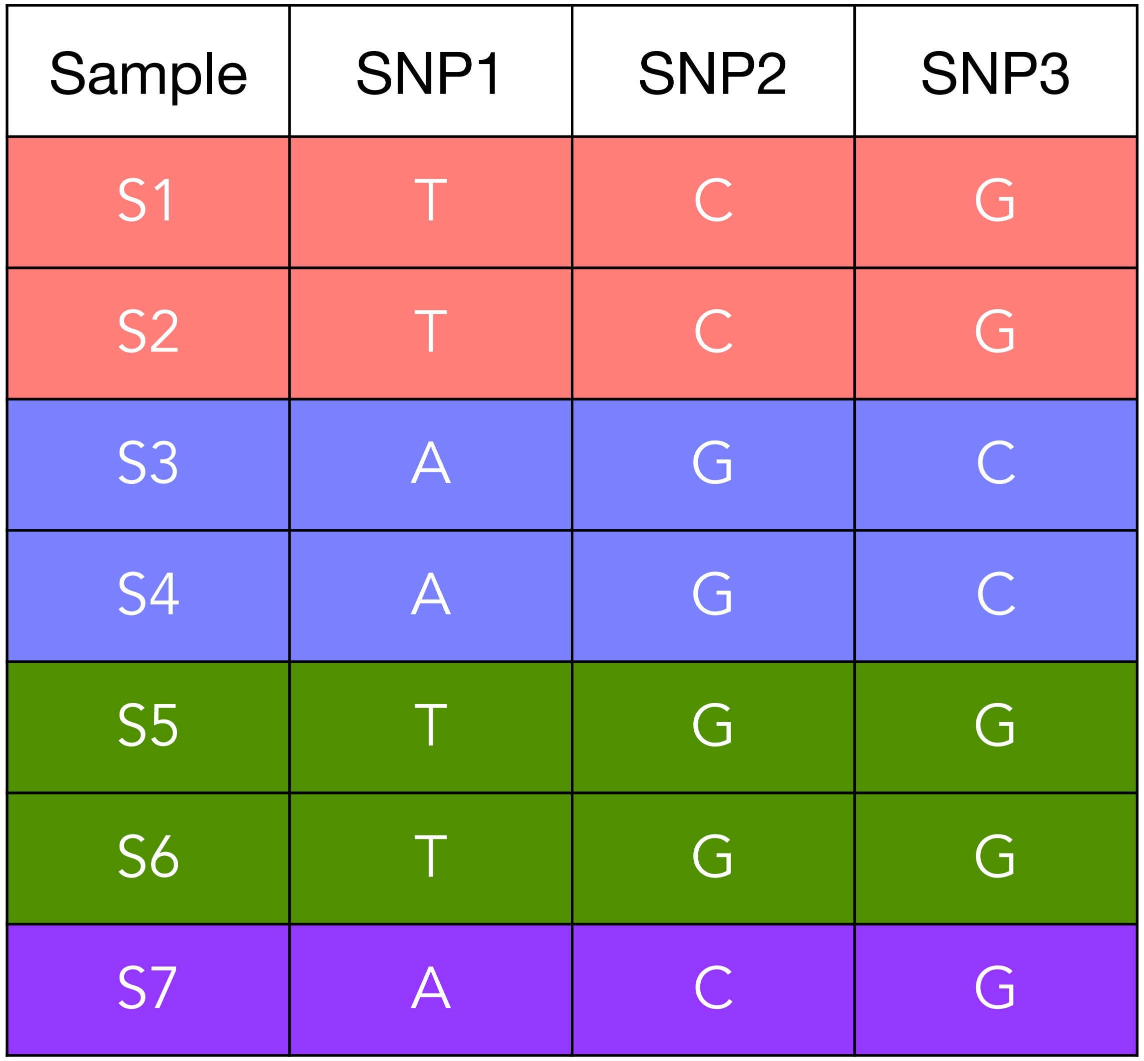
PopGen ▷ Bullet Points

Sample	SNP1	SNP2	SNP3
S1	T	C	G
S2	T	C	G
S3	A	G	C
S4	A	G	C
S5	T	G	G
S6	T	G	G
S7	A	C	G

Pairwise-Distance

	S1	S2	S3	S4	S5	S6	S7
S1	0						
S2	0	0					
S3	3	3	0				
S4	3	3	0	0			
S5	1	1	2	2	0		
S6	1	1	2	2	0	0	
S7	1	1	2	2	2	2	0

PopGen ▷ Bullet Points



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S4	A	G	C
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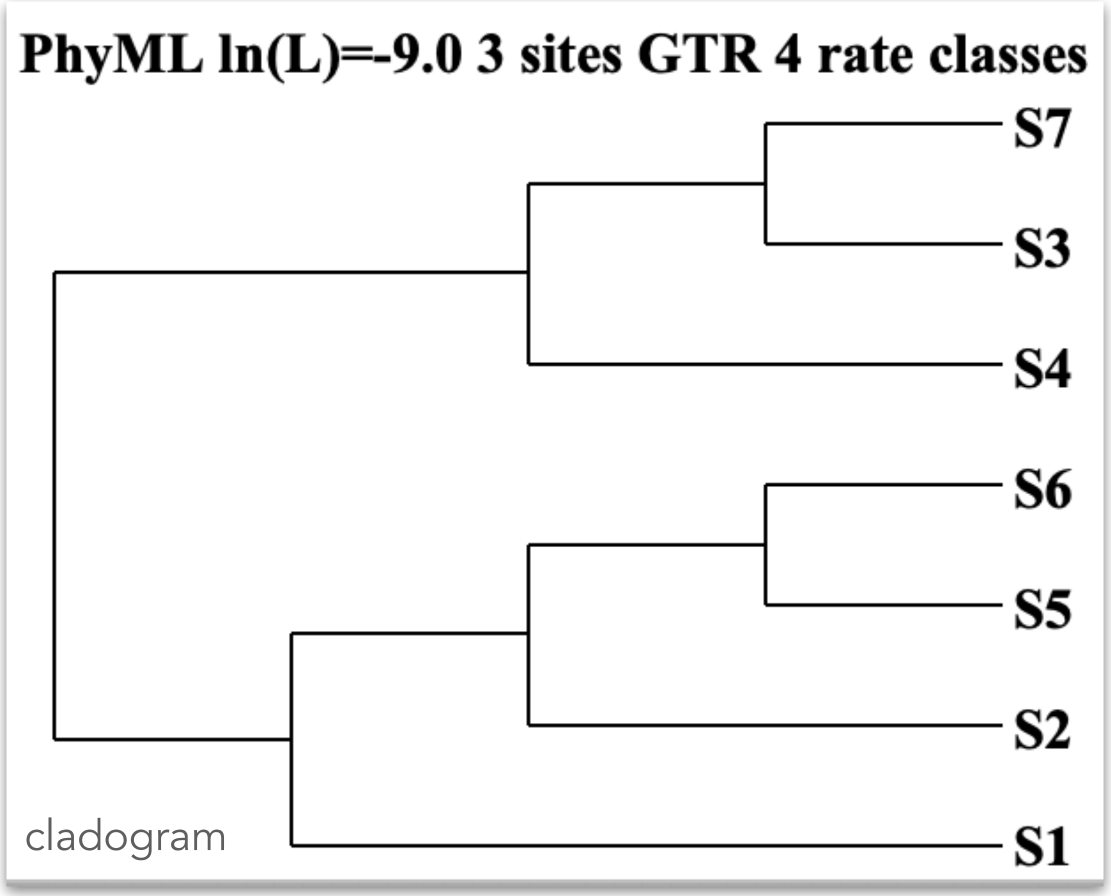
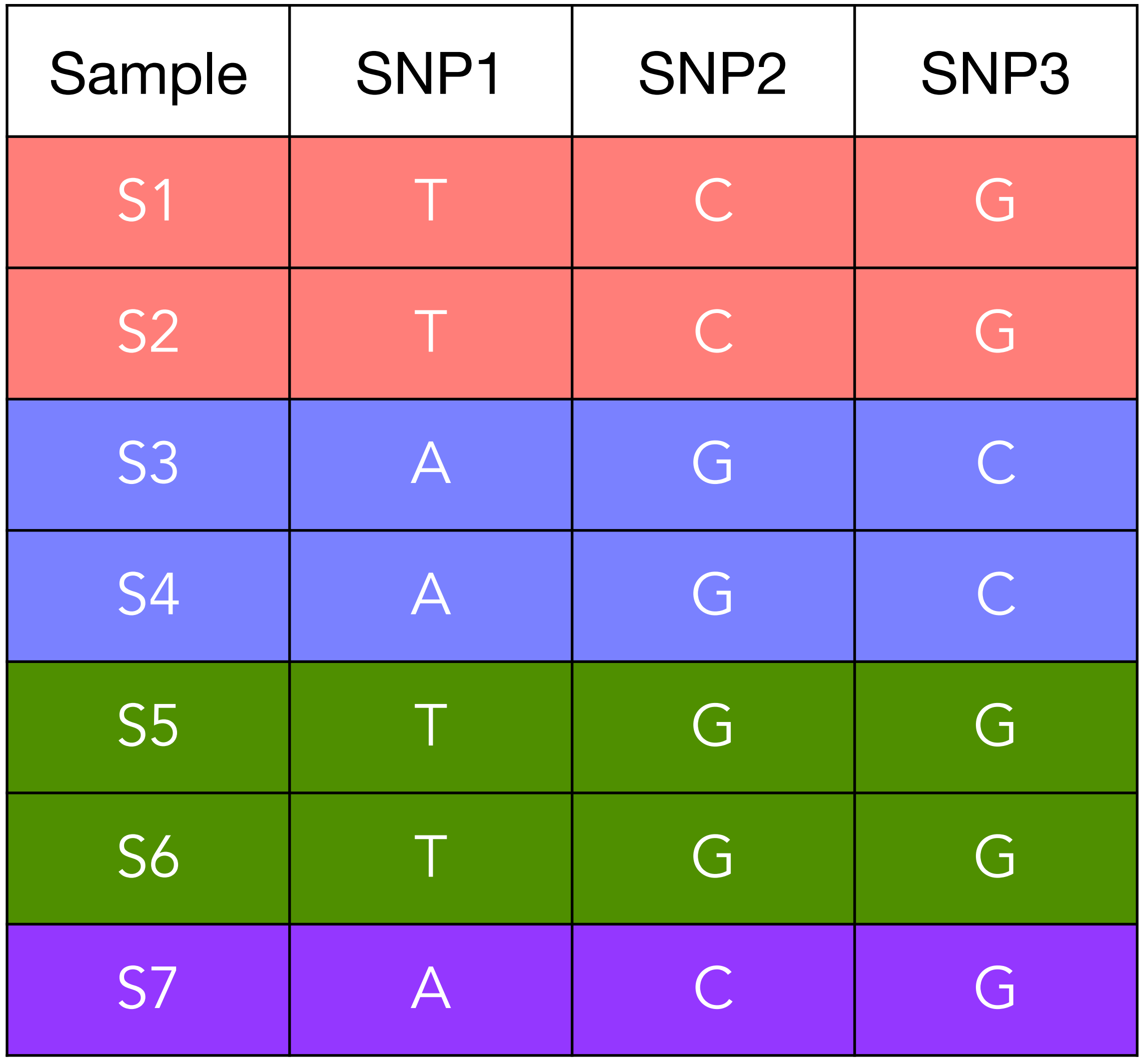
S6	T	G	G
S7	A	C	G

Phylogenetic-Analysis



PopGen ▷ Bullet Points



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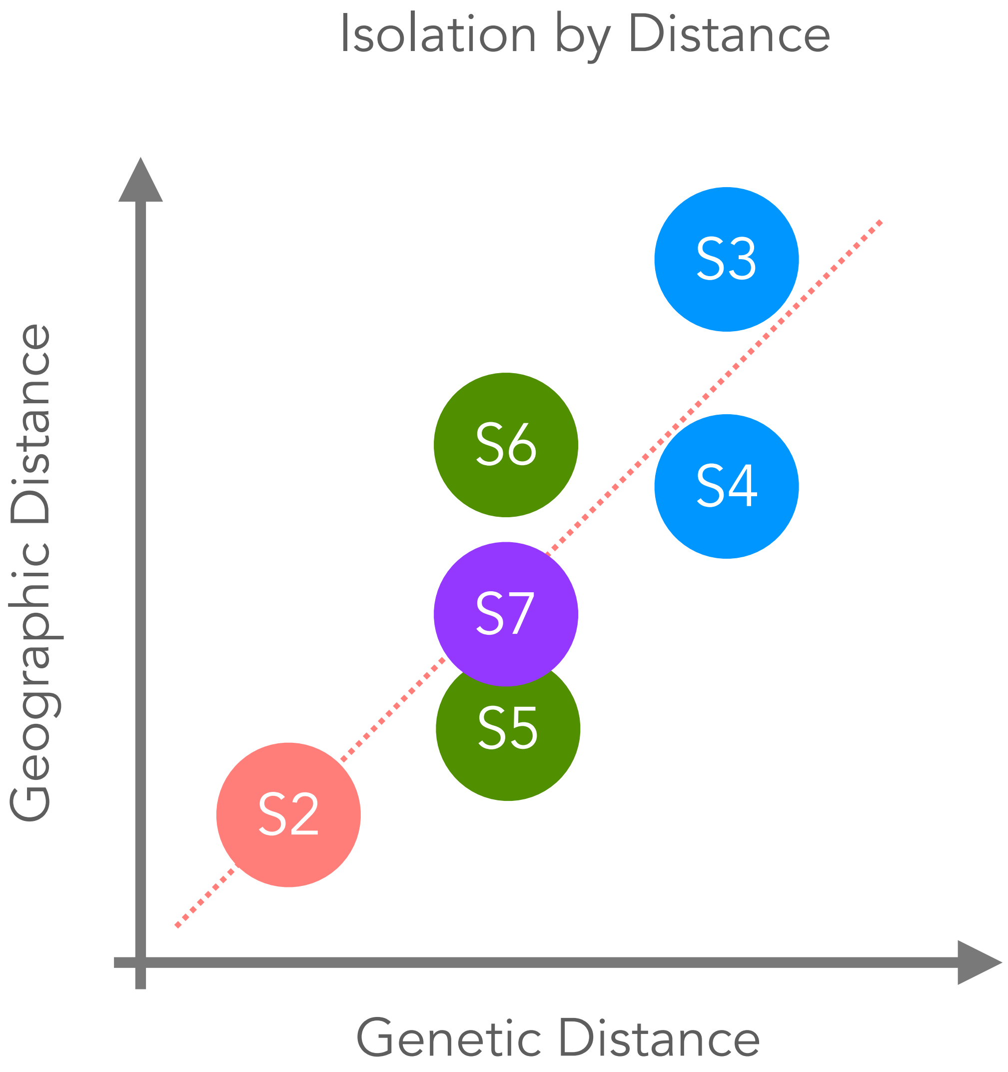


Isolation by Distance

PopGen ▷ Bullet Points

Theoretical Population Genetics

Coalescent

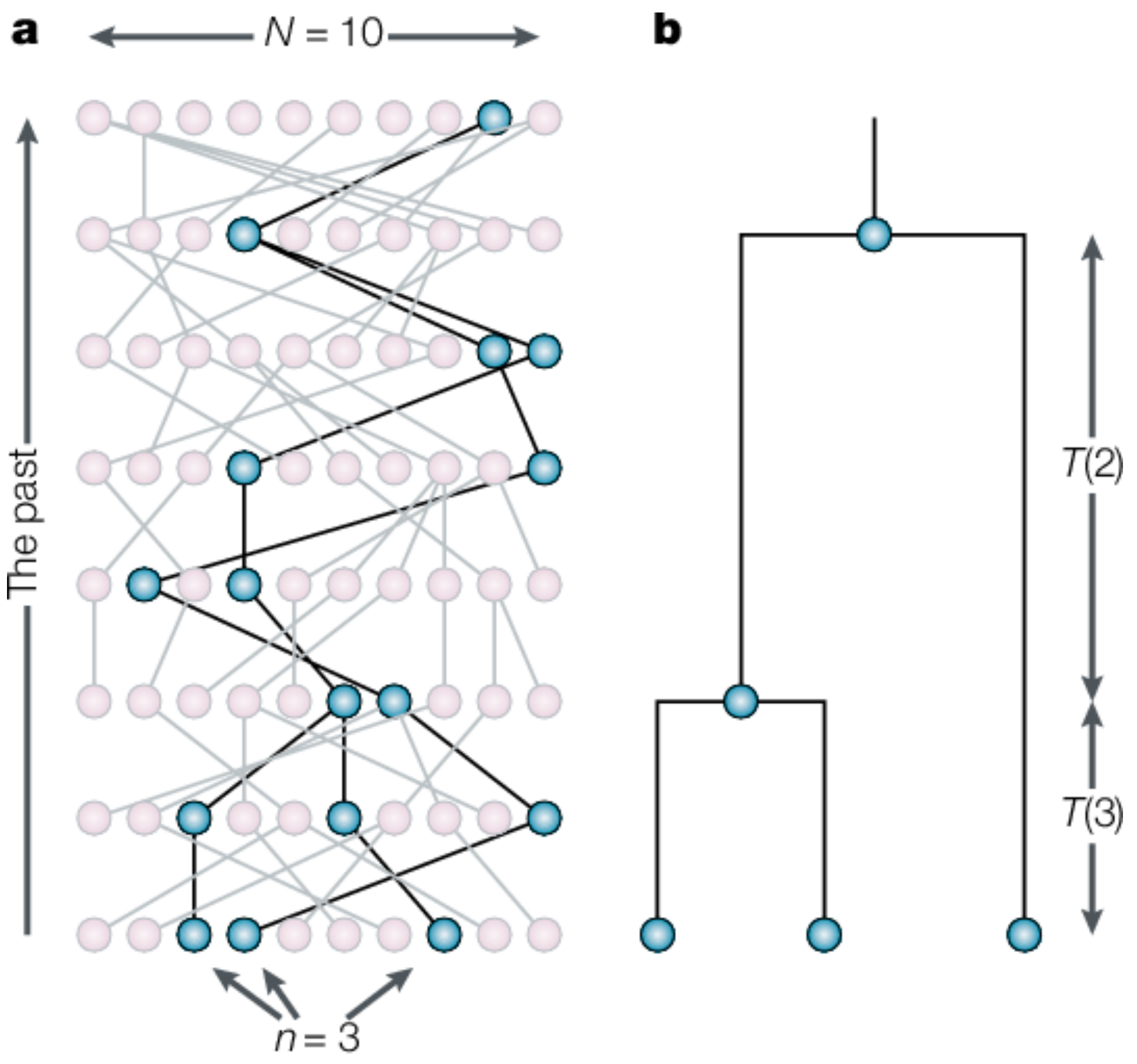
backward-in-time ← NOW → forward-in-time

Wright–Fisher

Coalescent theory is a model of how gene variants sampled from a population may have originated from a common ancestor.

The Wright–Fisher model, described in forward time, is a stochastic model of how gene frequencies in a population change across generations due to random sampling, mutation, selection, and genetic drift. It predicts the probability of allele frequencies evolving in a finite population over successive generations.

Basic principle behind coalescent



Coalescent theory models the genetic relationships among individuals by tracing their shared ancestry back through time to a common ancestor. It assumes a population framework where lineages "coalesce" at each generation, with the rate of coalescence influenced by population size and structure. It is widely used to study genetic variation, demographic history, and evolutionary processes.

PopGen ▷ Bullet Points

Extinction

PopGen ▷ Bullet Points

Evolutionary suicide (Ferrière, 2000), also known as Darwinian extinction (Webb, 2003) or evolution to extinction (Dieckmann et al., 1995) is an evolutionary phenomenon in which the process of adaptation causes the population to become extinct. For example, individuals might be selected to switch from eating mature plants to seedlings, and thereby deplete their food plant's population.

According to estimations about 99.9% of all species are extinct.

PopGen ▷ Bullet Points

Panda depend on bamboo but have troubles digesting it.

Even though bamboo is their main food source, giant pandas are horrible at digesting it. A look at the giant-panda gut microbiome (bacteria living in the stomach and intestines) showed that the animals have relatively few bacteria that help digest fibrous plants such as bamboo. The animals also do not have the genes for plant-digesting enzymes in their own genome. This combined scenario may have increased their risk for extinction.

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Mendelian Inheritance

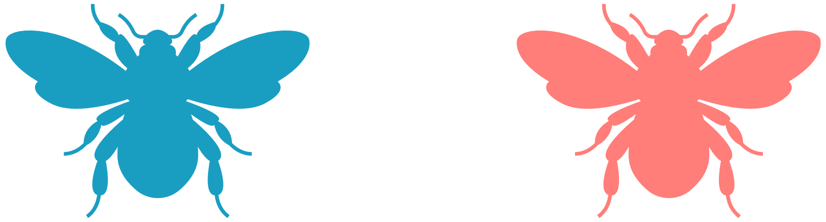
Mendelian inheritance is a type of biological inheritance following the principles originally proposed by Gregor Mendel in 1865 and 1866, re-discovered in 1900 by Hugo de Vries and Carl Correns, and later popularized by William Bateson.

Source: Wikipedia

PopGen ▷ Bullet Points

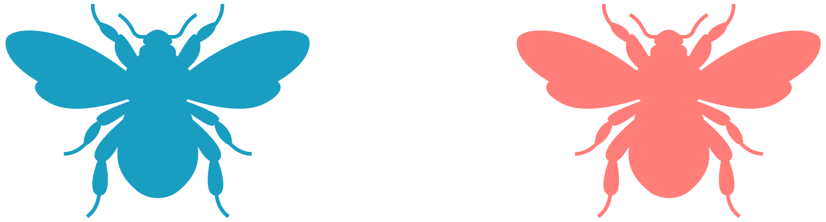
Mendelian Inheritance

Phenotype



Mendelian Inheritance

Phenotype



Genotype

AA
Aa

aa

A: dominante allele
a: recessive allele

homozygote: AA aa
heterozygote: Aa

AA Aa aa | AA AB BB | A_1A_1 A_1A_2 A_2A_2

A: Wild-Type
a: Mutant

A: Wild-Type
B: Mutant

A_1: Wild-Type
A_2: Mutant

PopGen ▷ Bullet Points

x	A	A
A	AA	AA
A	AA	AA

x	a	a
a	aa	aa
a	aa	aa

punnett square

x	A	a
A	AA	Aa
a	Aa	aa

25% - **50%** - 25%

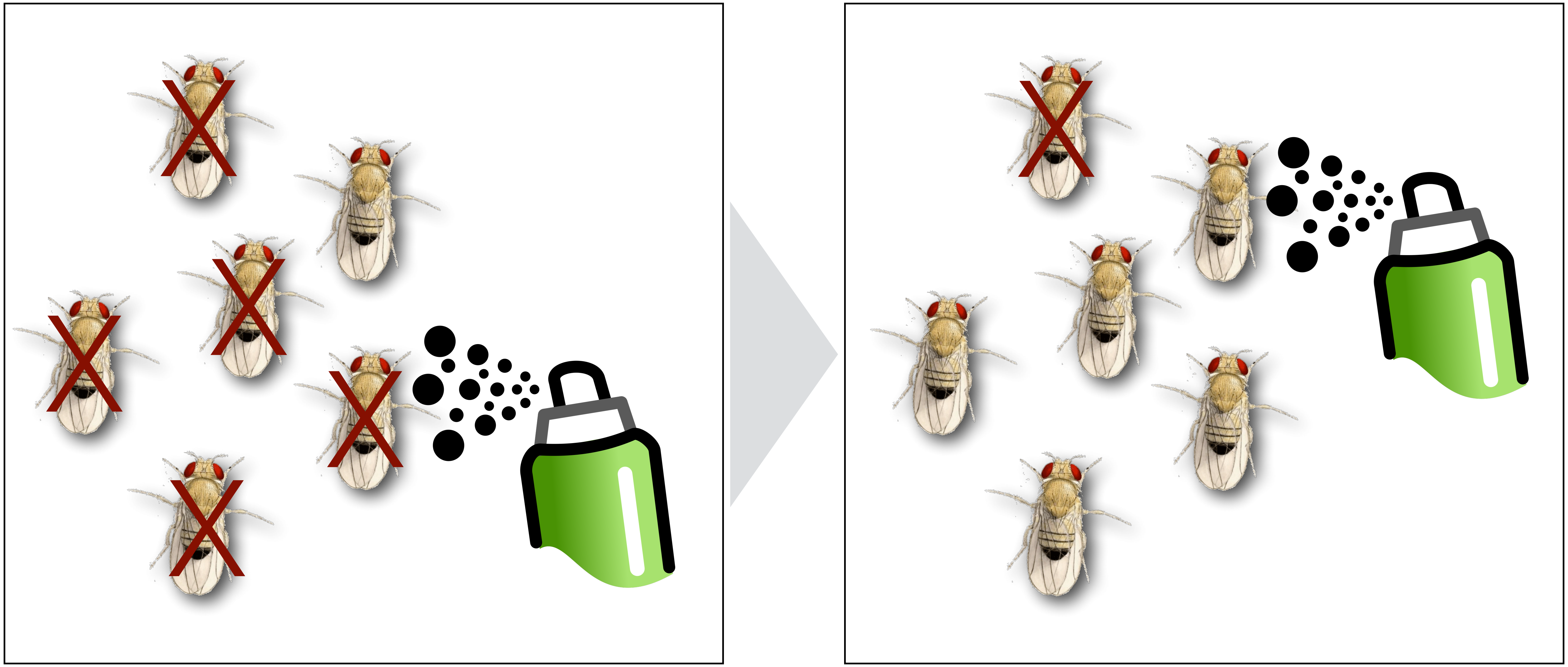
x	A	A
a	Aa	Aa
a	Aa	Aa

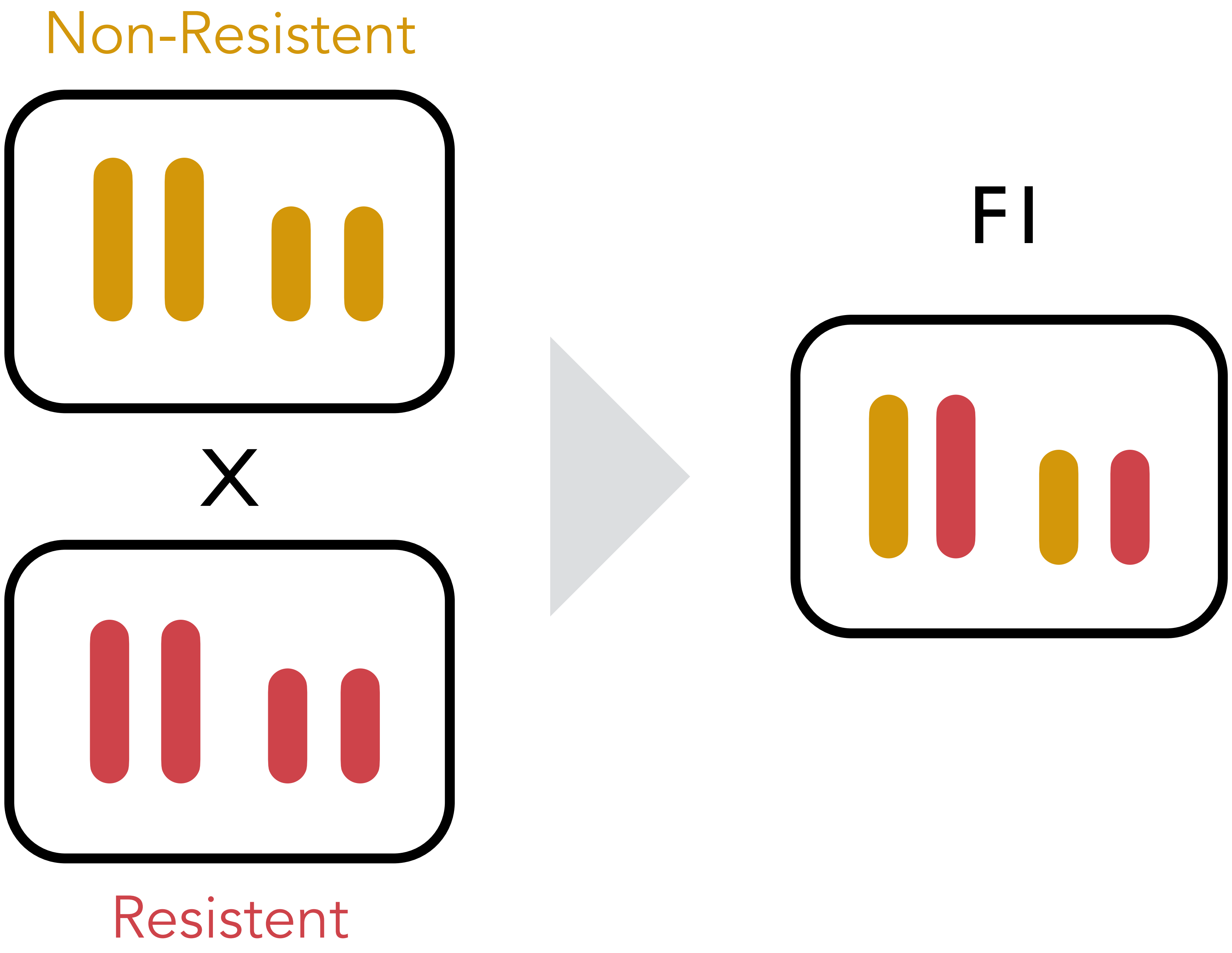
0% - **100%** - 0%

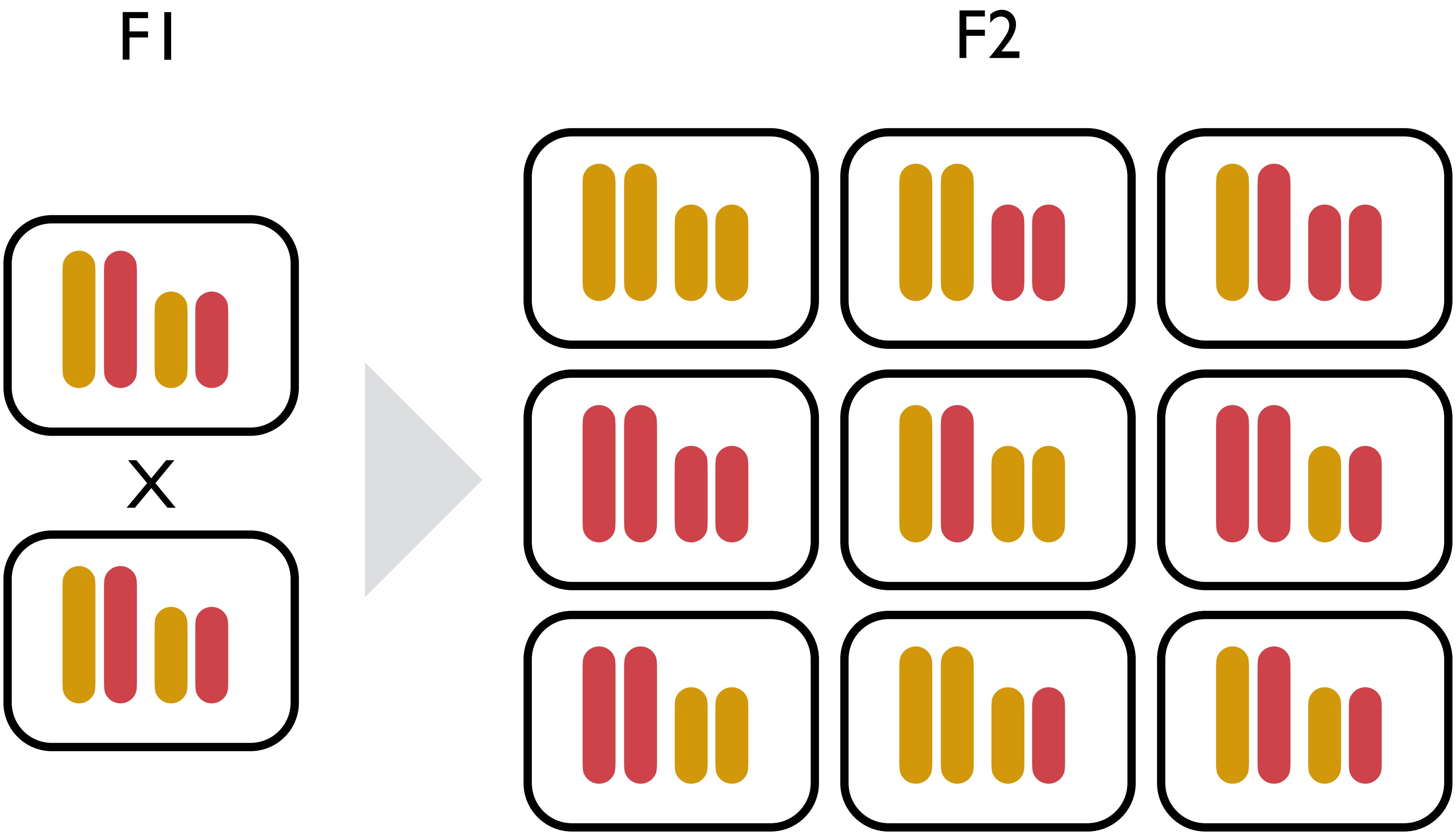
x	A	A
A	AA	AA
a	Aa	Aa

50% - **50%** - 0%

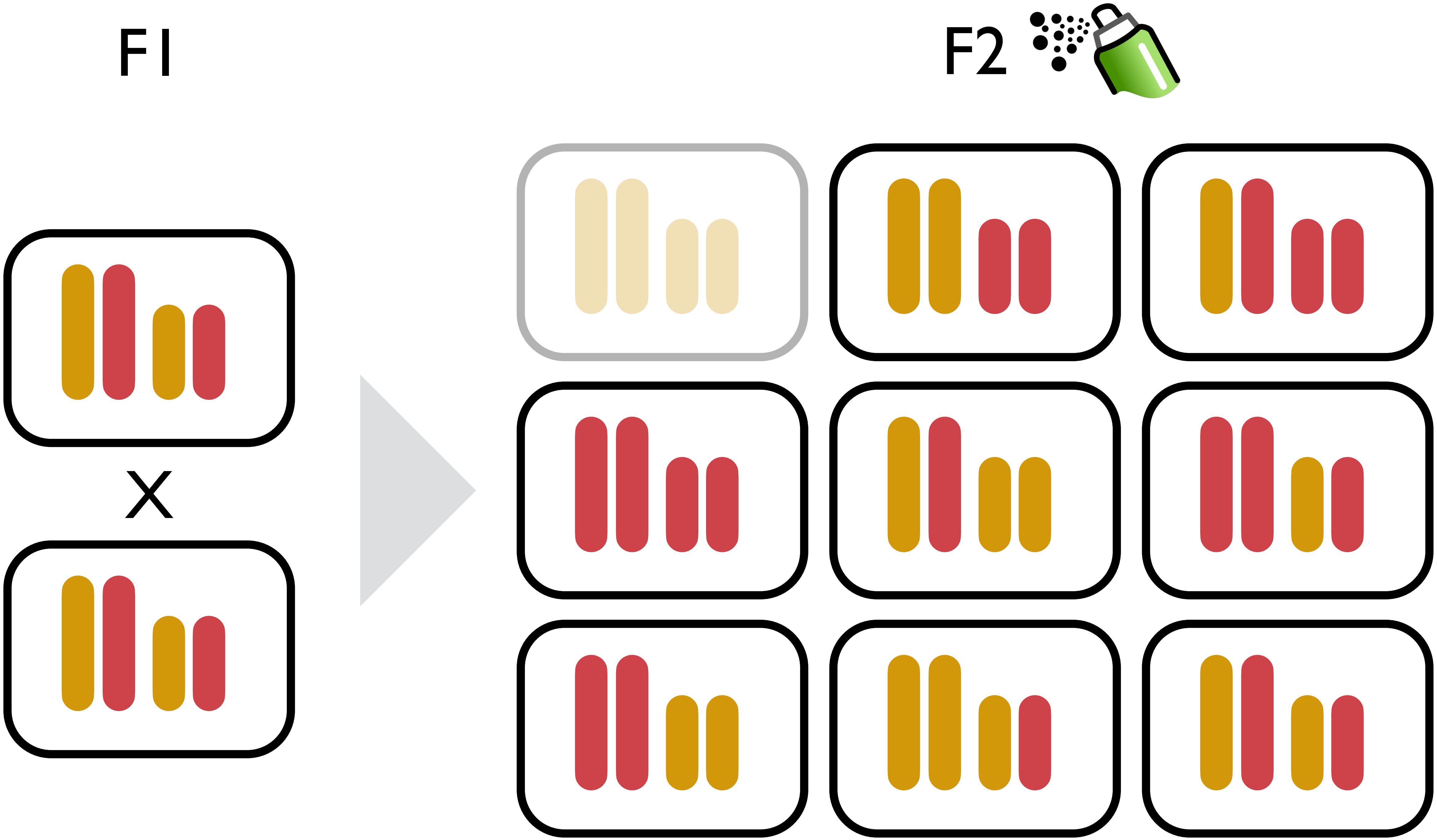
Resistance



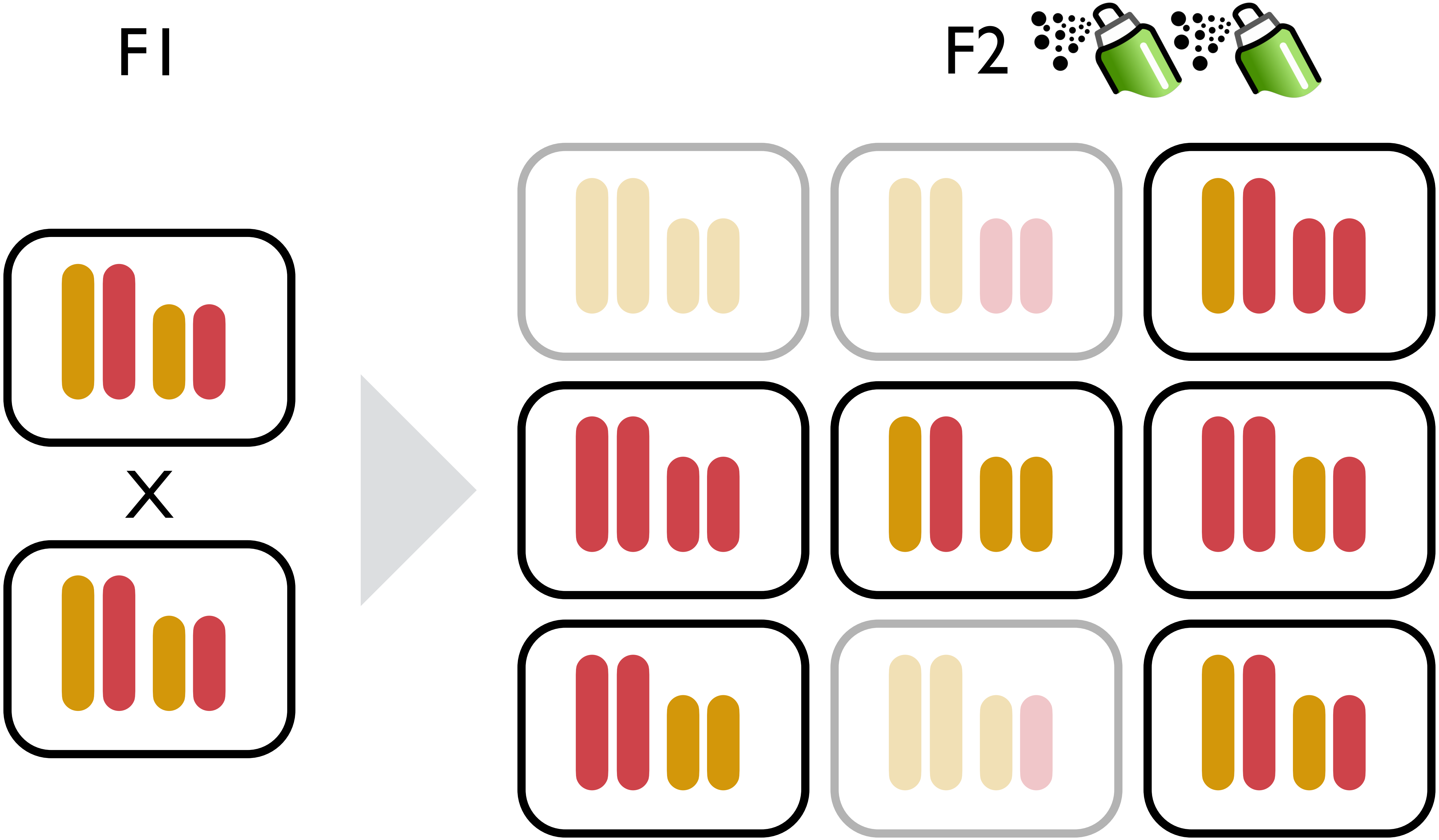

Non-Resistent
X
Resistent
FI


F1
X
F2

PopGen ▷ Bullet Points



PopGen ▷ Bullet Points



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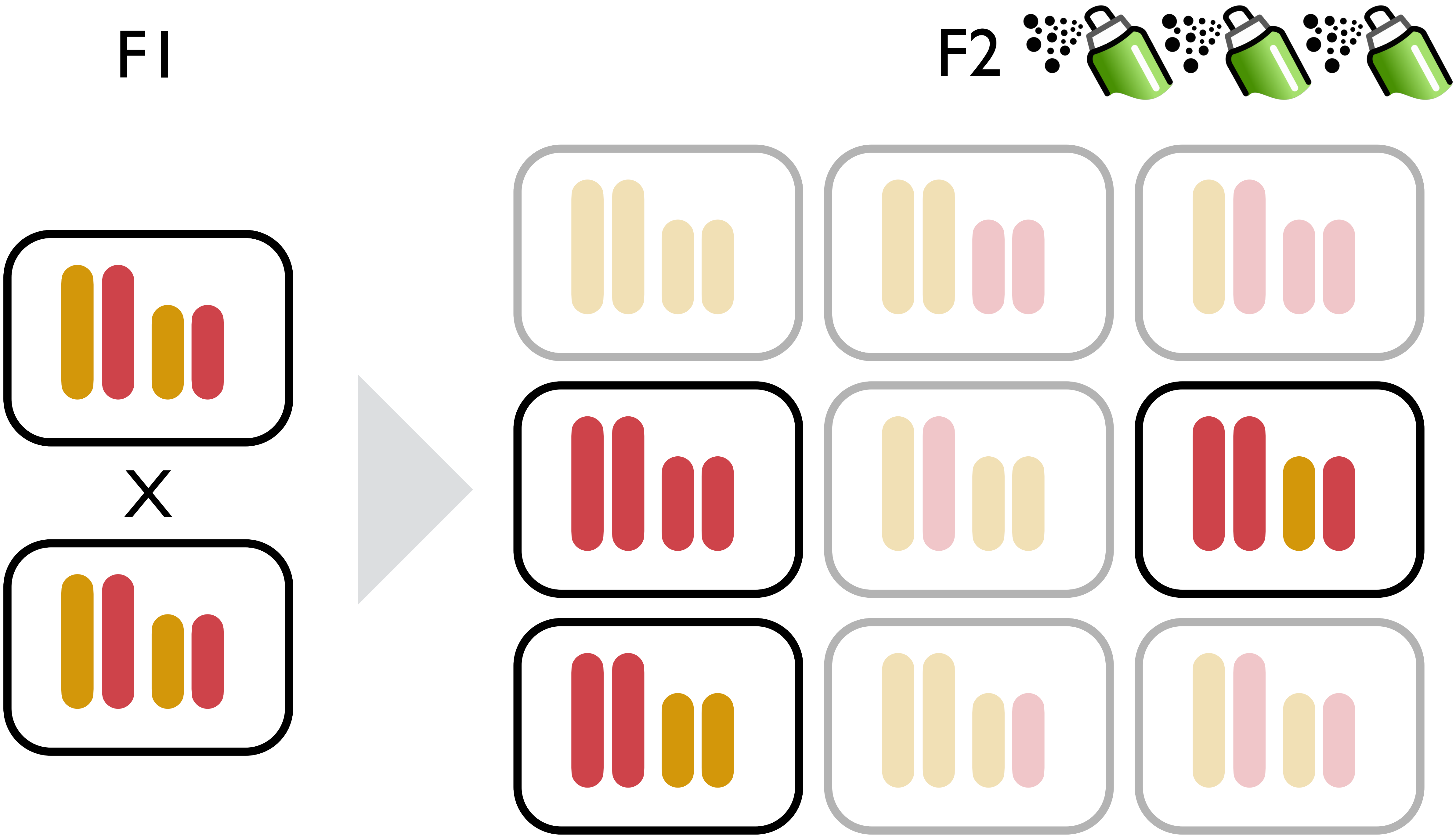

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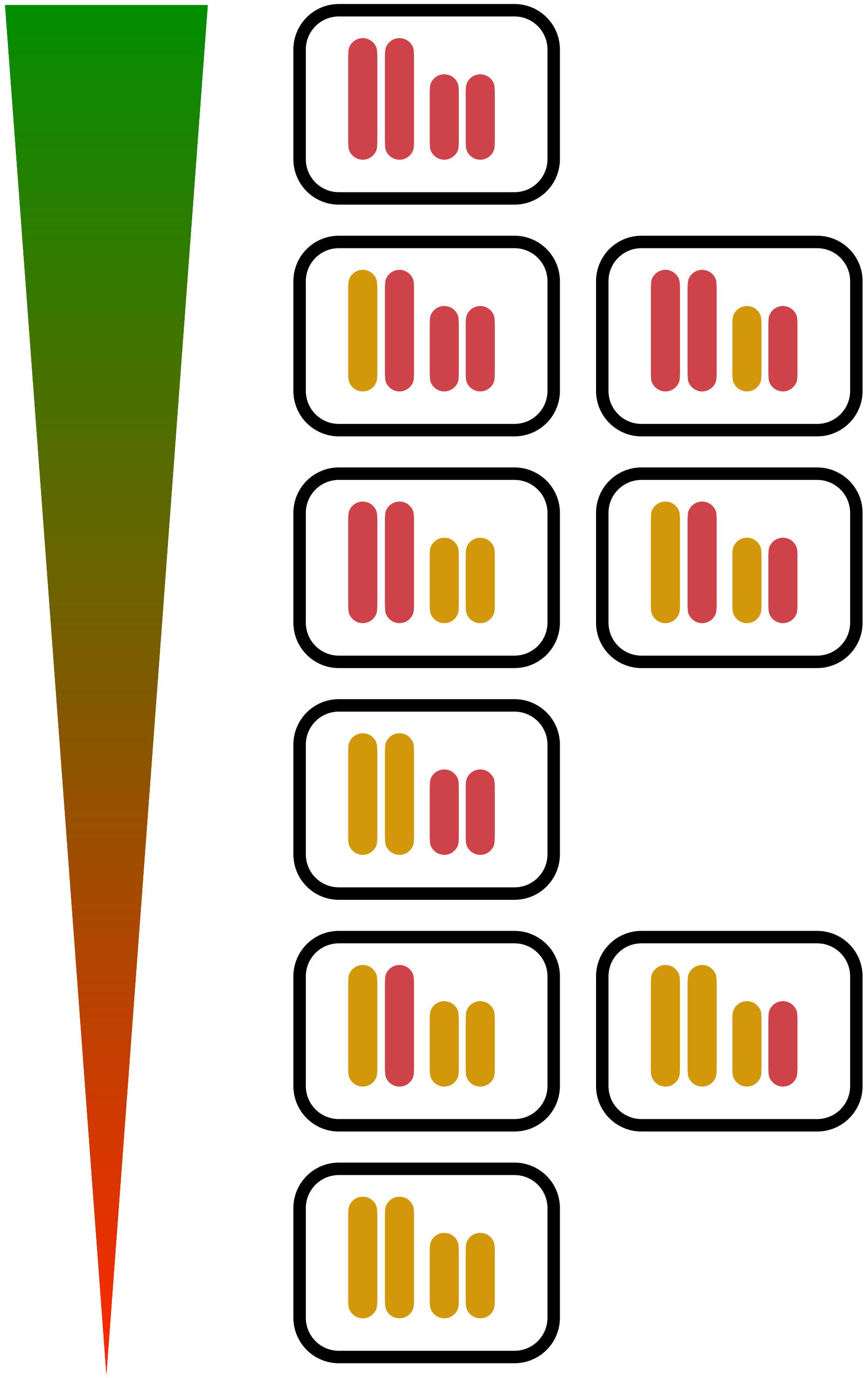
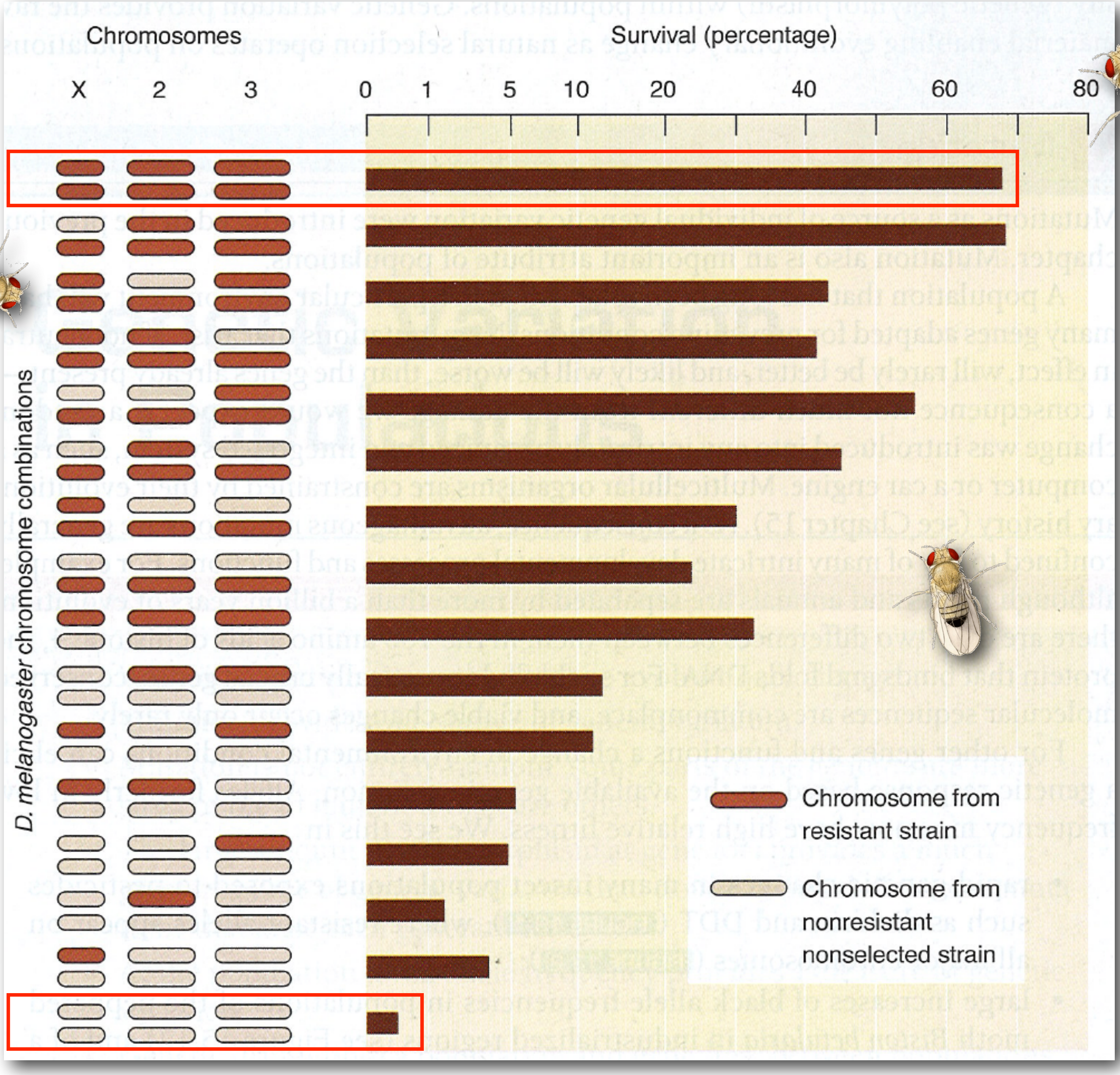
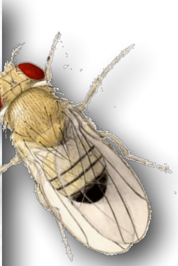
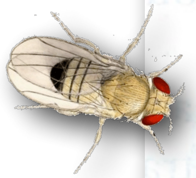
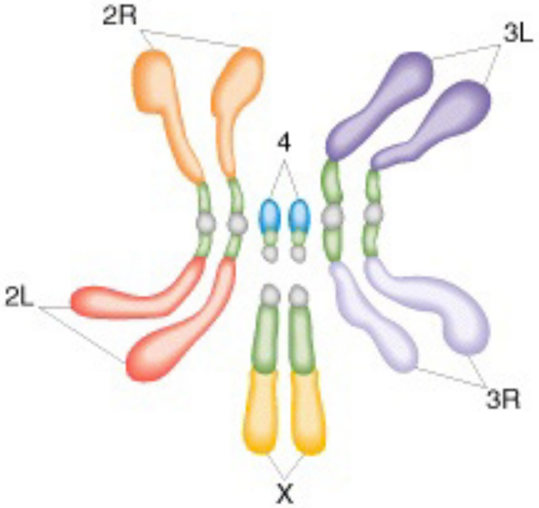


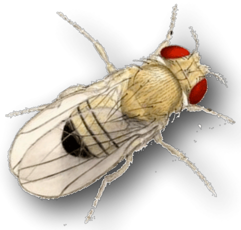
DDT-resistance in Dmel

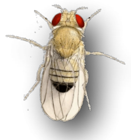


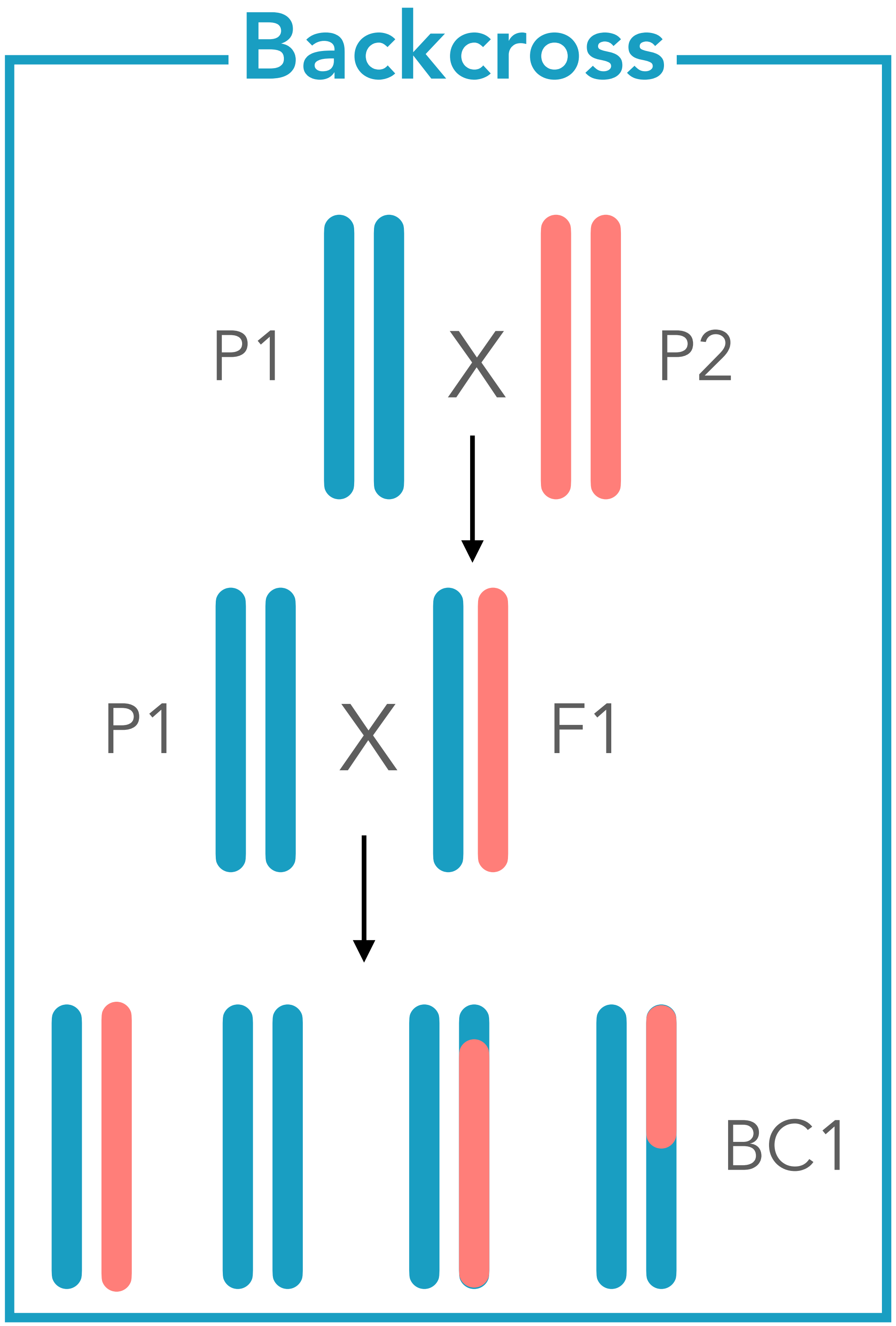




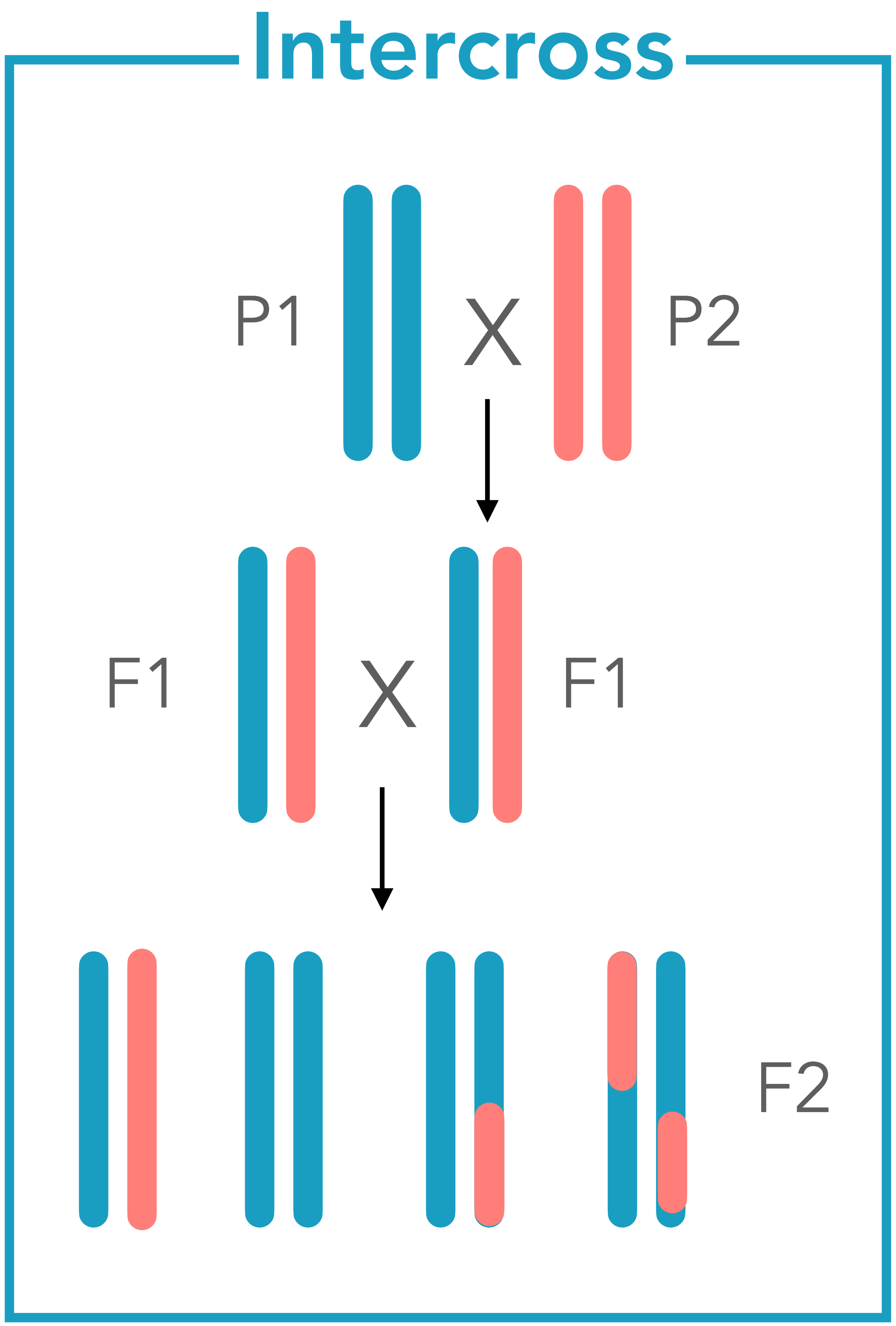








Backcross



Intercross

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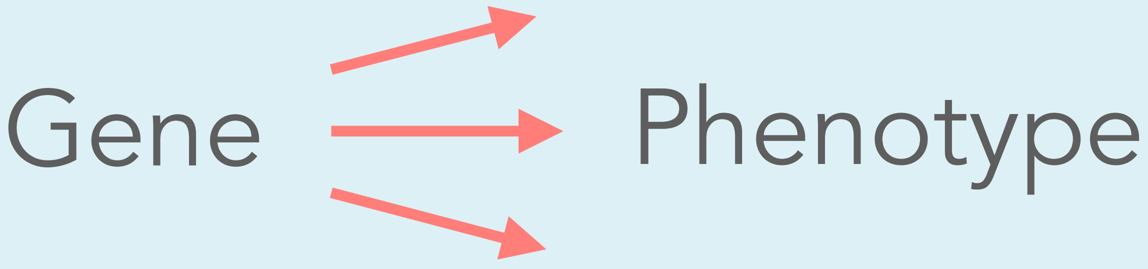
pleiotrophy → not one gene for one phenotype



heterogeneity → not one phenotype for one gene