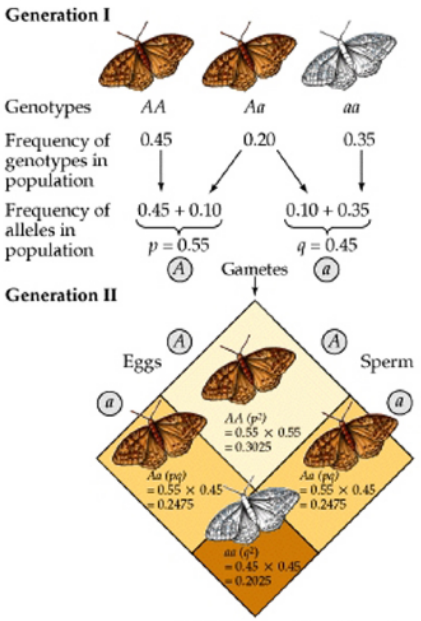
BIOL2107, Fall '23

Lecture 15



So, now we have analyzed four "variations" from the "predictable" Mendelian-type of inheritance-

(a) variations that arise as a consequence of "**extensions**" to Mendelian genetics, where the function of the genes in question may interact to give different F_2 phenotypes.

(b) variations that arise because of "**chromosomal linkage**" (thus defying Mendel's Second law) **X-linkage** & **autosomal linkage.**

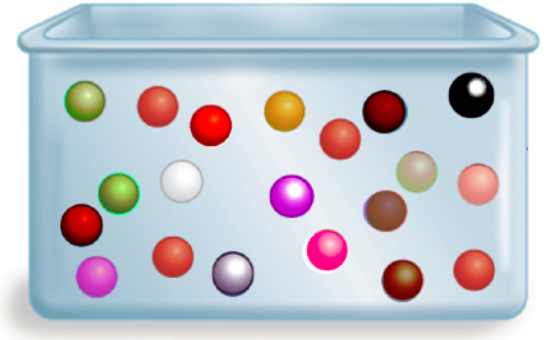
(c) Cytoplasmic / Maternal Inheritance

(d) Epigenetics

Genetic Variation within Populations

To recap (in light of the last few lectures):

For a population to evolve, its members must possess variation, which is the raw material on which "agents" or "forces" of evolution act (**genetic variation** within a **gene pool**).



We observe **phenotypes** in nature: i.e. the physical expressions of genes.

A **heritable trait,** however, is a genetic characteristic of an organism that is mainly influenced by the organism's genes (we cannot forget totally the influence of environment on this expression).

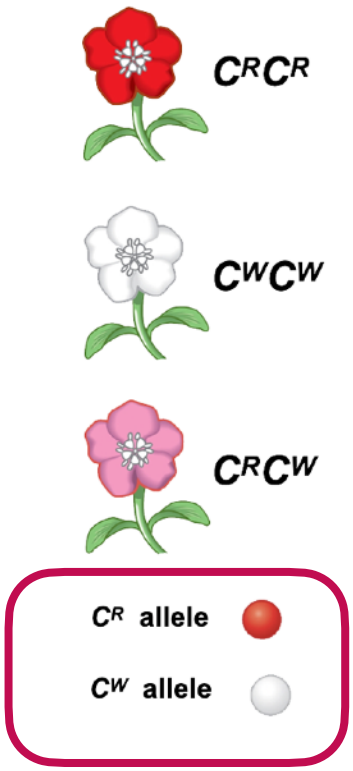
The genetic component that governs a given **phenotypic trait** is called it's **genotype**.

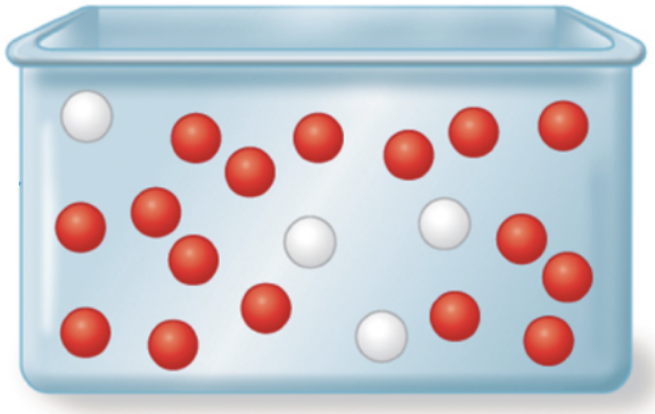
A population evolves when individuals with different **genotypes** survive or reproduce at different rates.

From Mendel, we know that **Genes** are “units”, which have different forms called **alleles**.

A single individual has only some of the **alleles** found in a population.

The sum of all the alleles in a population is its **gene pool**, which contains the variation (different **alleles**) that produce the differing phenotypes, upon which change can come about...evolution.

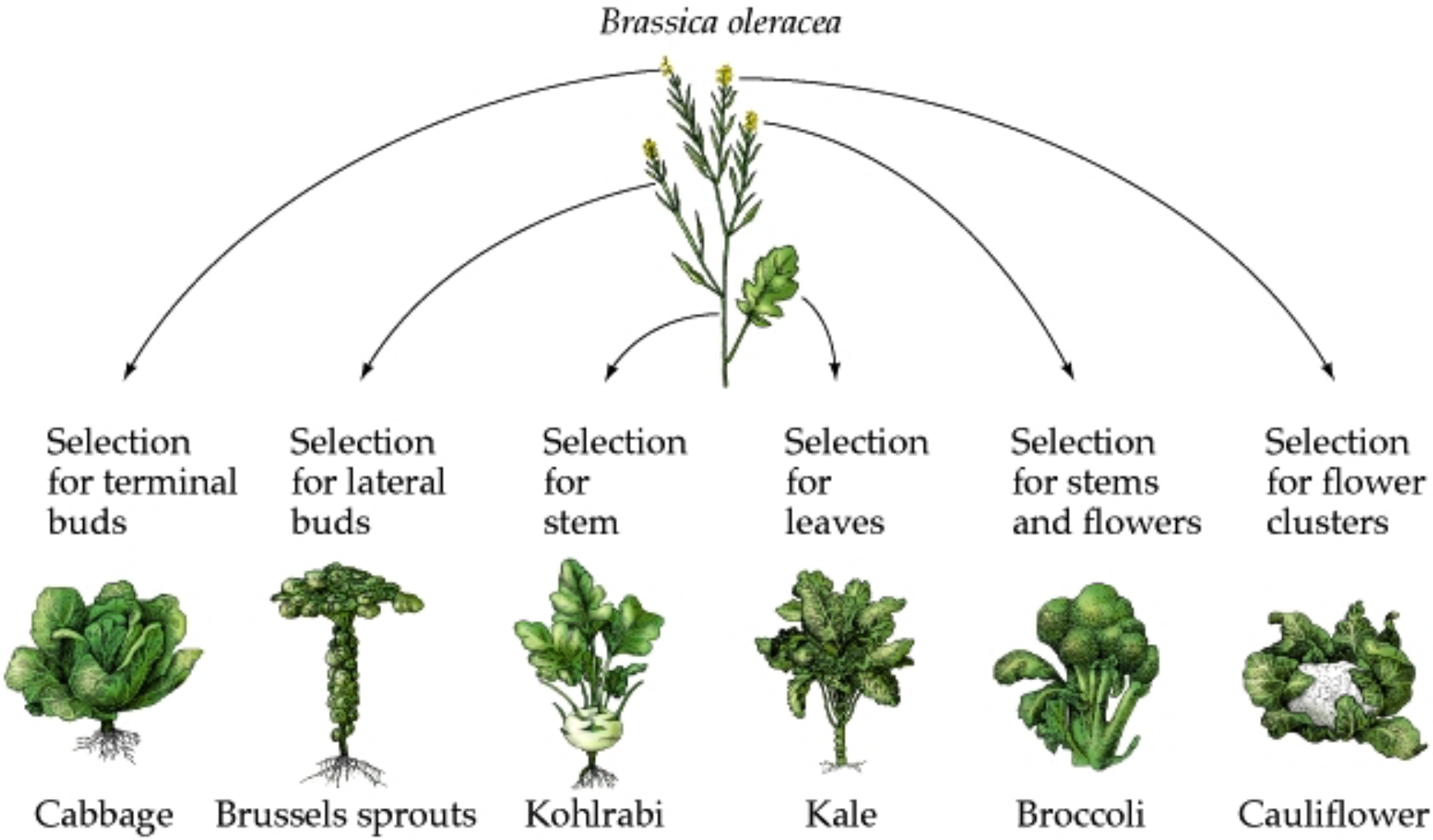


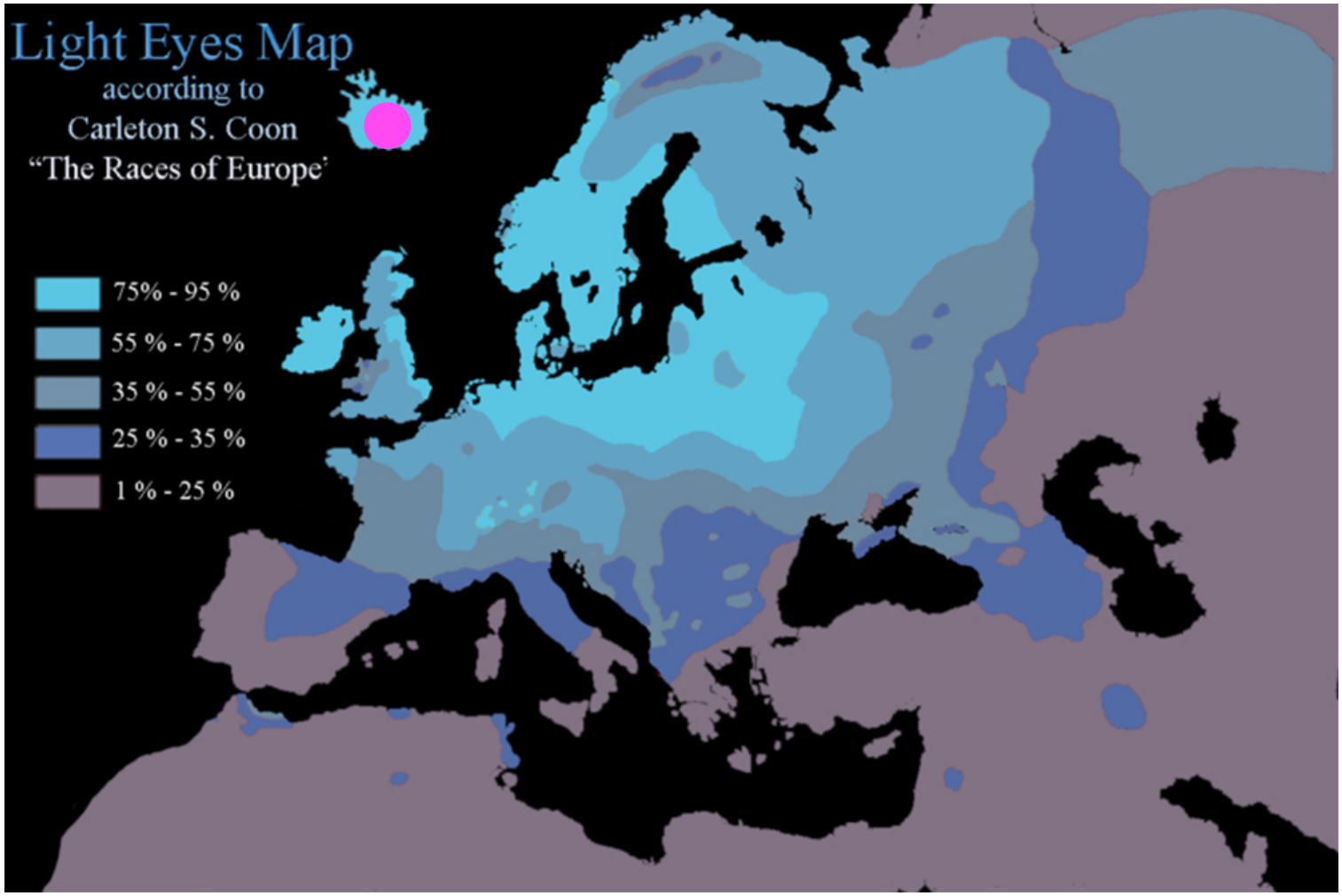


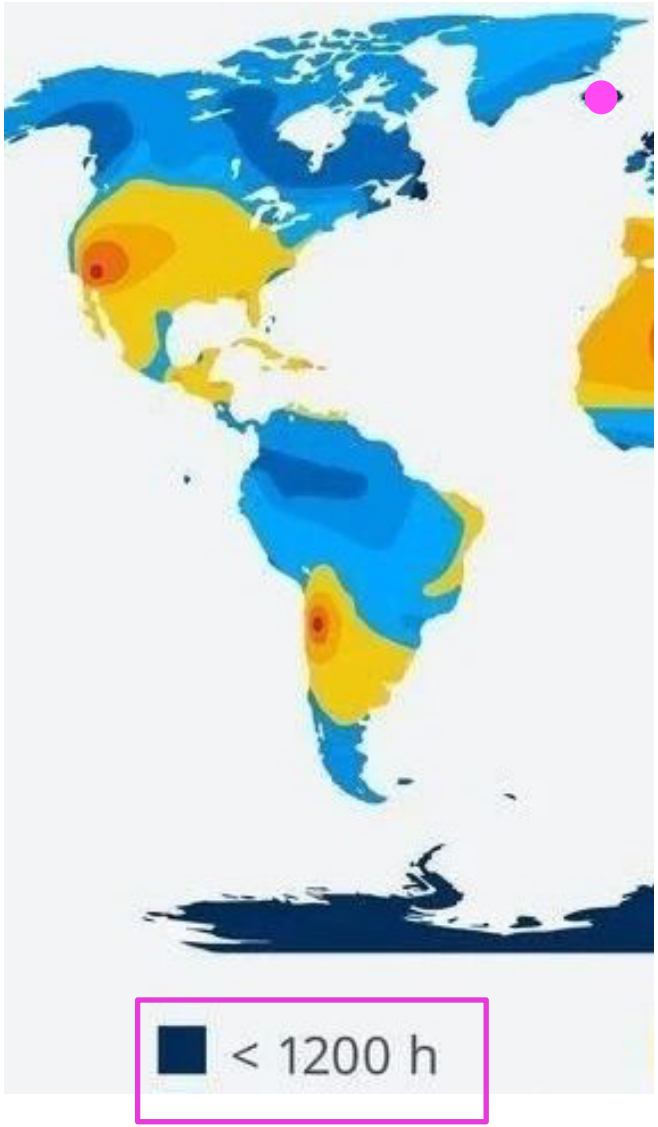
Also that the **allelic frequency** is the frequency of finding that particular allele within a given **gene pool**

Most populations are **genetically variable**.

Natural populations possess inherent genetic variation.




Light Eyes Map
according to
Carleton S. Coon
"The Races of Europe"
75% - 95 %
55 % - 75 %
35 % - 55 %
25 % - 35 %
1 % - 25 %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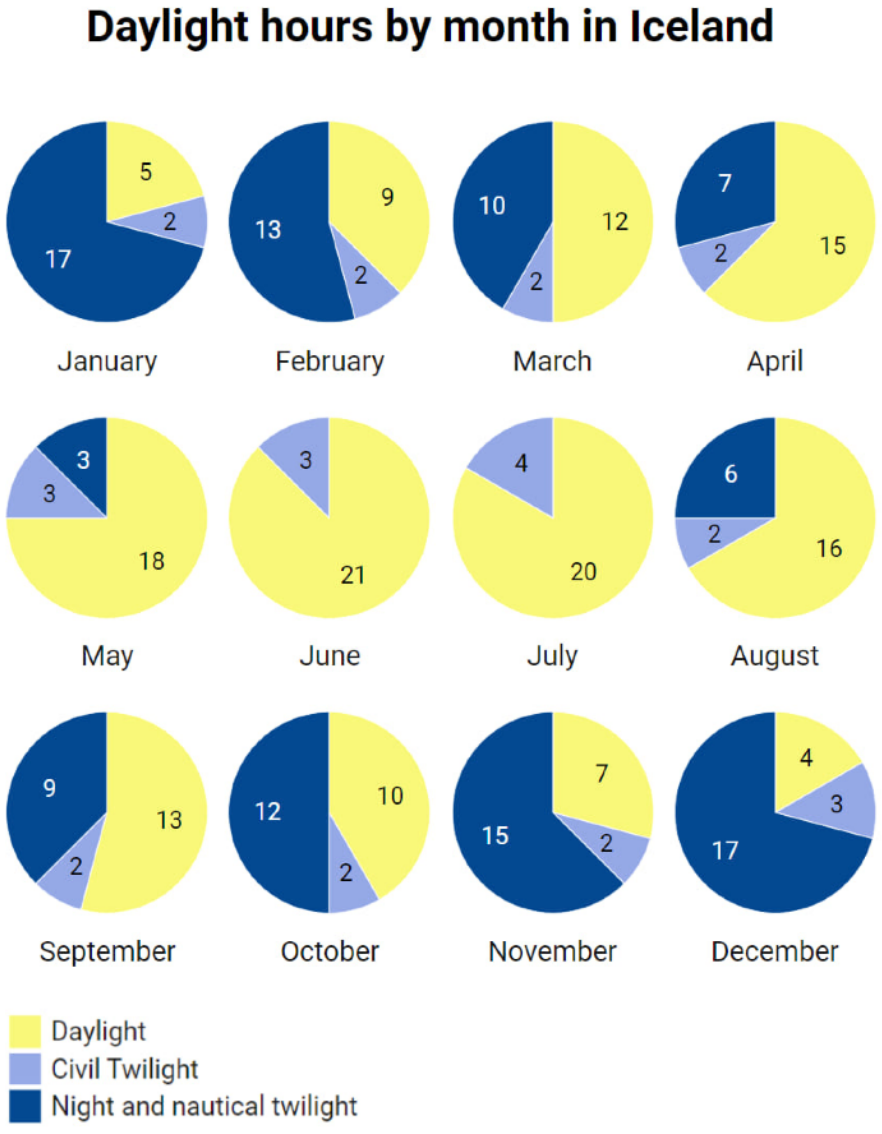

Daylight hours by month in Iceland
5
2
17
January
9
13
2
February
10
12
2
March
7
2
15
April
3
3
18
May
3
21
June
4
20
July
6
2
16
August
9
13
2
September
12
10
2
October
7
15
2
November
4
3
17
December
Daylight
Civil Twilight
Night and nautical twilight

Evolution: Natural Selection…

Genetic Toolkit movie (lecture 6)…

Over the course of the last 600 million years, "what is evolution really working on… it's the recipe, it's the genes"

Looking at evolution of populations through the eyes of a geneticist, you can think of **Natural Selection** in terms of **phenotypes** and **genotypes**.

Beneficial phenotypes -with some type of advantage will be selected over others… But, how are these genes "assessed"? -through the survivors passing on their particular form of genes… their "alleles" on to the next generation.

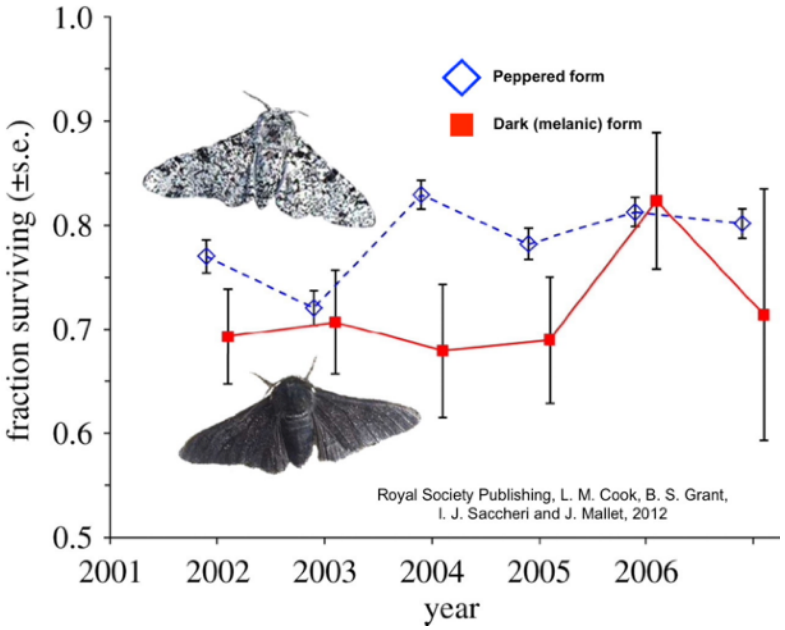
Over time, the gene pool of a given population will have more copies of those **alleles** that code for beneficial phenotypes, and less copies of alleles for harmful traits. The central thesis of this argument is that ***-through selection of phenotypes, natural selection actually changes the allele frequencies in a population's gene pool.***

What is **Natural Selection** working on? What is it selecting for?

For survival and reproduction.

In the same way that through “artificial selection”, crop breeders, farmers select the crops / animals with the most desirable traits…

In the experiment below; by eating the “easily viewed” moths the birds effectively change the phenotype of the moth population (the frequency of the two alleles) over time, i.e the allele frequencies will shift to match this selective regimen.



experimental data from studies by Michael Majerus (1954-2009).

Such analyses demonstrated ~9% drop in highly pigmented moths (on average).. over just a 6 year time course.

Hardy Weinberg Principle:

"the frequency of **alleles** and **genotypes** in a population will remain constant over time -in the absence of other evolutionary influences".

In essence, the Hardy–Weinberg equilibrium describes the-

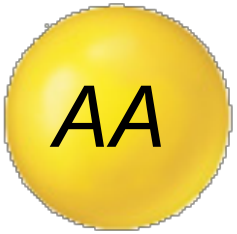
"**Perfect, Mendelian Population**", without ANY Evolutionary variation.

The resulting **HW Principle** relates "**genotypes**" to measurable "**allele frequencies**".

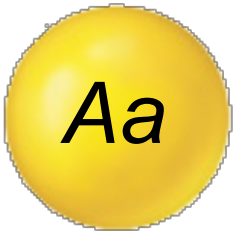
and gives us some appreciation as to how such "**Mendelian populations**" will / will not change over time.

…because in "true" Mendelian populations" according to the **HW Principle** the population is at **equilibrium**

Genotypes



Homozygous DOMINANT



Heterozygous

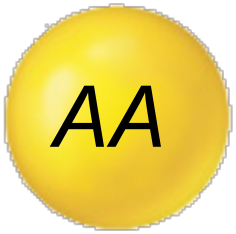
Homozygous recessive

Allele Frequency:

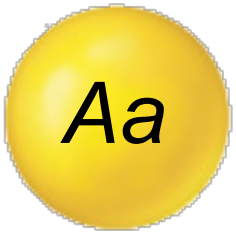
= frequency of **“A”** and the frequency of **“a”** in the above population

Genotype Frequency -in a population is the number of individuals with a given **Genotype**

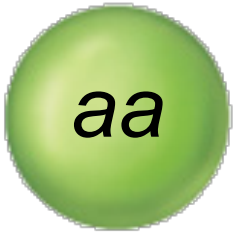
Let **Allele frequency** **of "A" = "p"** and **of "a" = "q"**



p^2



$2pq$

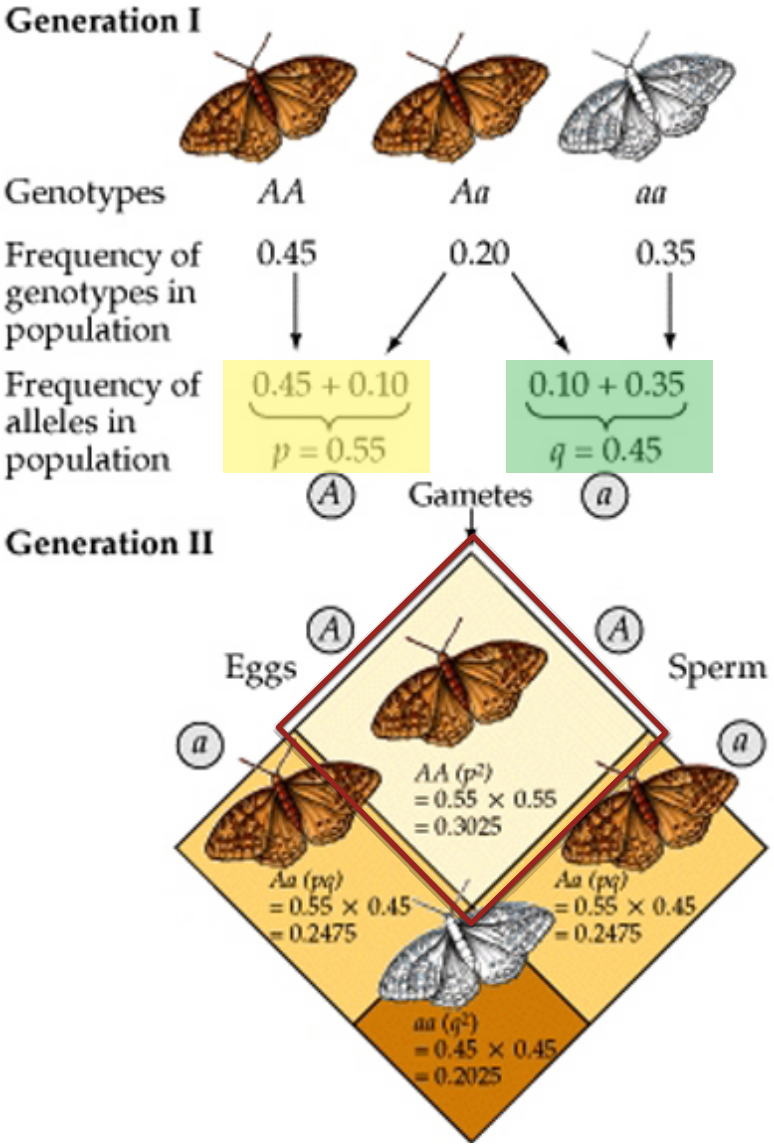


q^2

at equilibrium… **Genotype frequency = 1**

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

Hardy–Weinberg equation


Generation I
Genotypes
AA
Aa
aa
Frequency of genotypes in population
0.45
0.20
0.35
Frequency of alleles in population
0.45 + 0.10
p = 0.55
0.10 + 0.35
q = 0.45
A
Gametes
a
Generation II
A
A
Eggs
Sperm
a
a
AA (p²)
= 0.55 × 0.55
= 0.3025
Aa (pq)
= 0.55 × 0.45
= 0.2475
Aa (pq)
= 0.55 × 0.45
= 0.2475
aa (q²)
= 0.45 × 0.45
= 0.2025

Genotype Frequency -in a population is the number of individuals with a given Genotype

Let **Allele frequency** **of "A" = "p"** and **of "a" = "q"**

AA

p^2

Aa

$2pq$

aa

q^2

at equilibrium… **Genotype frequency = 1**

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

Hardy–Weinberg equation

C^RC^R

C^WC^W

C^RC^W

C^R allele

C^W allele

C^RC^R

C^WC^W

C^RC^W

C^R allele

C^W allele

Gene pool of the initial (parental) generation:

80% C^R (p = 0.8) 20% C^W (q = 0.2)

Sperm

C^R p = 0.8 C^W q = 0.2

Eggs

C^R p = 0.8

C^W q = 0.2

	0.64 (p^2) C^RC^R	0.16 (pq) C^RC^W
0.16 (gp) C^RC^W		0.04 (q^2) C^WC^W

Genotypes in this offspring generation:

64% C^RC^R, 32% C^RC^W, and 4% C^WC^W

Alleles in this offspring generation:

64% C^R (from C^RC^R plants) + 16% C^R (from C^RC^W plants) = 80% C^R = 0.8 = p

4% C^W (from C^WC^W plants) + 16% C^W (from C^RC^W plants) = 20% C^W = 0.2 = q

With random mating, these gametes will result in the same mix of genotypes in the next generation:

64% C^RC^R, 32% C^RC^W, and 4% C^WC^W plants

C^RC^R

C^WC^W

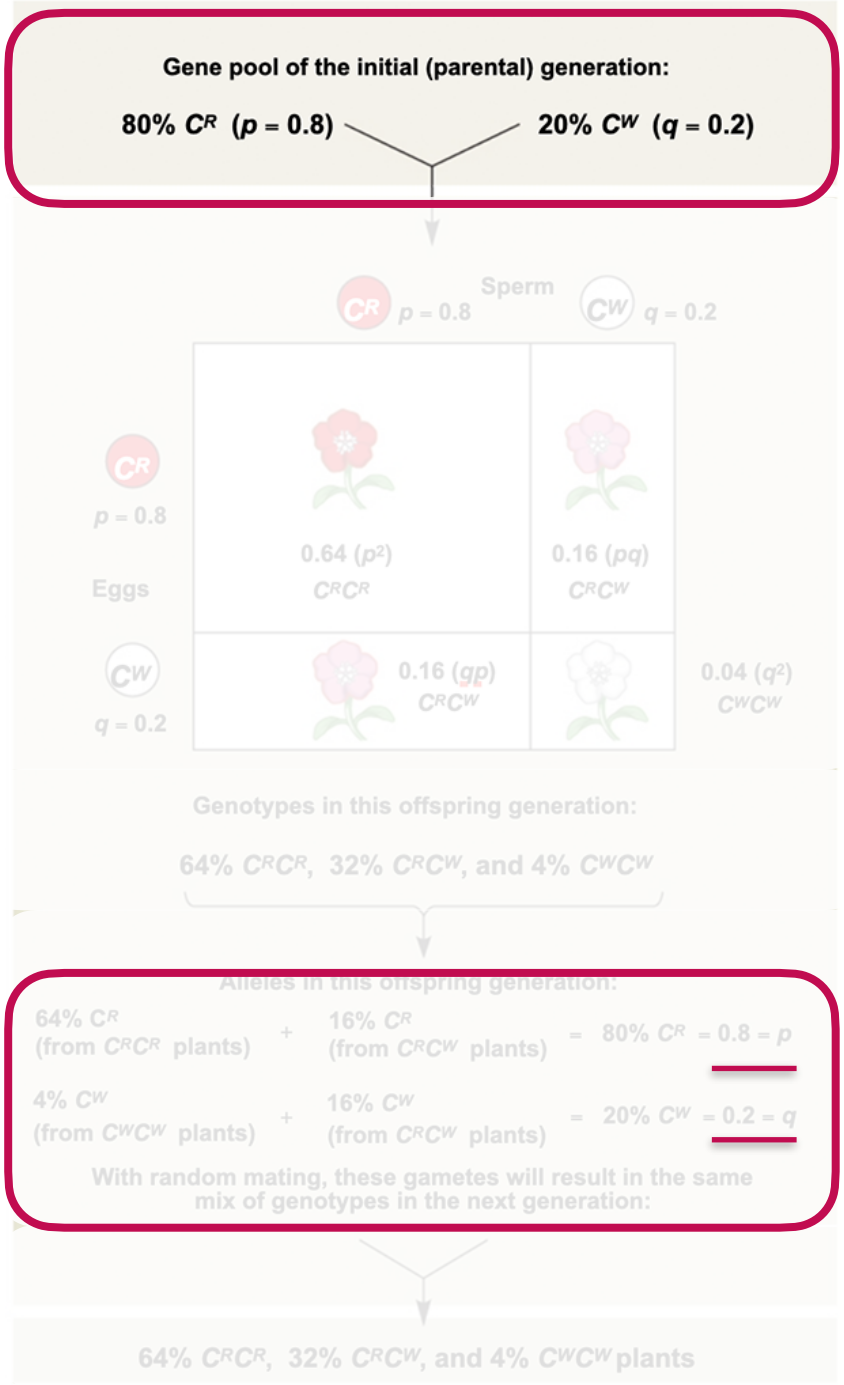
C^RC^W

C^R allele

C^W allele

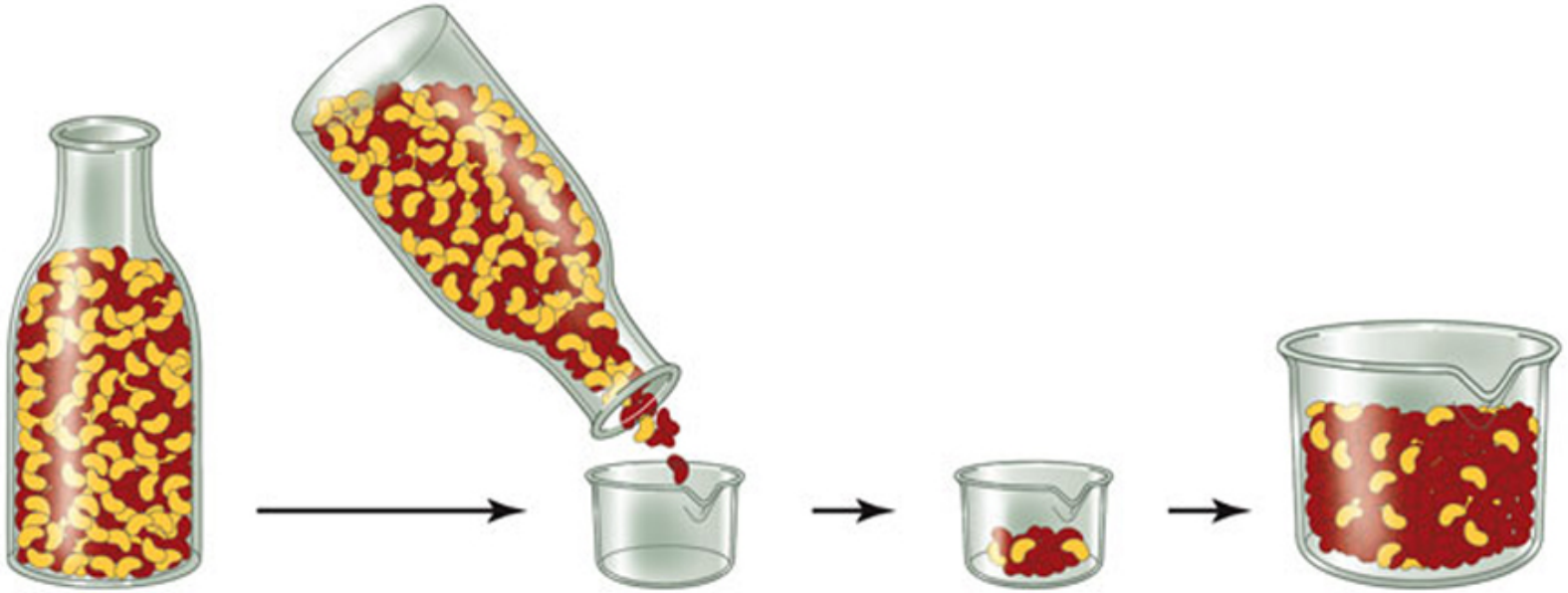
$$p^2 + 2pq + q^2 = 1$$

Hardy–Weinberg equilibrium

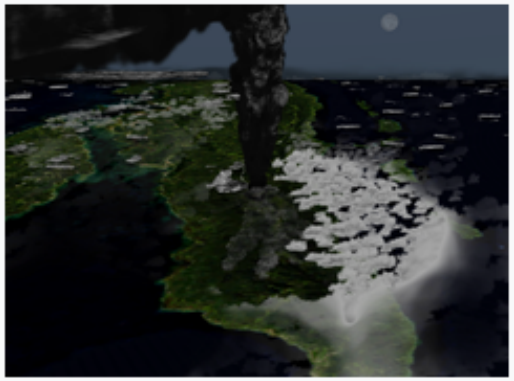


Major Changes in the **HW equilibrium** often signal dramatic changes in population stability…

It can also indicate recovery of a population from dramatic events… such as a **bottle neck effect**.



Youngest Toba eruption



Artist's impression of the eruption from about 42 kilometres (26 mi) above Northern Sumatra

Volcano	Toba Caldera Complex
Date	75,000 ± 900 years BP
Type	Ultra-Plinian
Location	Sumatra, Indonesia 2.6845°N 98.8756°E
VEI	8
Impact	Second-most recent supervolcanic eruption; impact disputed

Lake Toba is the resulting crater lake

The **most recent** Toba eruption was a supervolcanic eruption that occurred around 75,000 years ago at the site of present-day Lake Toba in Sumatra, Indonesia. It is one of the Earth's largest known explosive eruptions.

The **Toba catastrophe theory** holds that this event caused a global volcanic winter of six to ten years and possibly a 1,000-year-long cooling episode.

In 1993, science journalist Ann Gibbons posited that a population bottleneck occurred in human evolution about 70,000 years ago, and she suggested that this was caused by the eruption.

Geologist Michael R. Rampino of New York University and volcanologist Stephen Self of the University of Hawai‘i at Mānoa support her suggestion. In 1998, the bottleneck theory was further developed by anthropologist Stanley H. Ambrose of the University of Illinois at Urbana–Champaign. Both the link and global winter theories are controversial.[1]

The **Hardy–Weinberg equation** can also be used as the “ultimate” evolutionary “null hypothesis”…

Hardy–Weinberg equilibrium

When a population is at "equilibrium" **there can be no differences in the survival and reproductive success of individuals.** i.e there is NO selective elimination of *a* alleles (**NO SELECTION**), meaning that the frequency of *a* will gradually decline (and the frequency of *A* correspondingly increase) over the generations. As we discuss below, we call this differential success of alleles.

Populations must not be added to or subtracted from by migration. (**NO GENE FLOW).** Consider a second population adjacent to the one we used in the preceding example in which all the alleles are *A* and all individuals have the genotype *AA.* Then there is a sudden influx of individuals from the first population into the second. The frequency of *A* in the second population changes in proportion to the number of immigrants.

The population must be sufficiently large to prevent sampling errors. Population size affects the Hardy–Weinberg equilibrium such that it technically holds true only for "infinitely" large populations. A change in the frequency of an allele due to the random effects of limited population size is called So, effectively **NO GENETIC DRIFT**.

There can be no mutation. If *A* alleles mutate into *a* alleles (or other alleles, if the gene has multiple alleles), and vice versa, then again we see changes in the allele frequencies over the generations. In general, because mutation is so rare, it has a very small effect on changing allele frequencies on the timescales studied by population geneticists.

Individuals must mate at random. For the Hardy–Weinberg equilibrium to hold, mate choice must be made without regard to genotype, *AA, Aa,* or *aa* individuals should choose and be chosen at random. **non-random mating**

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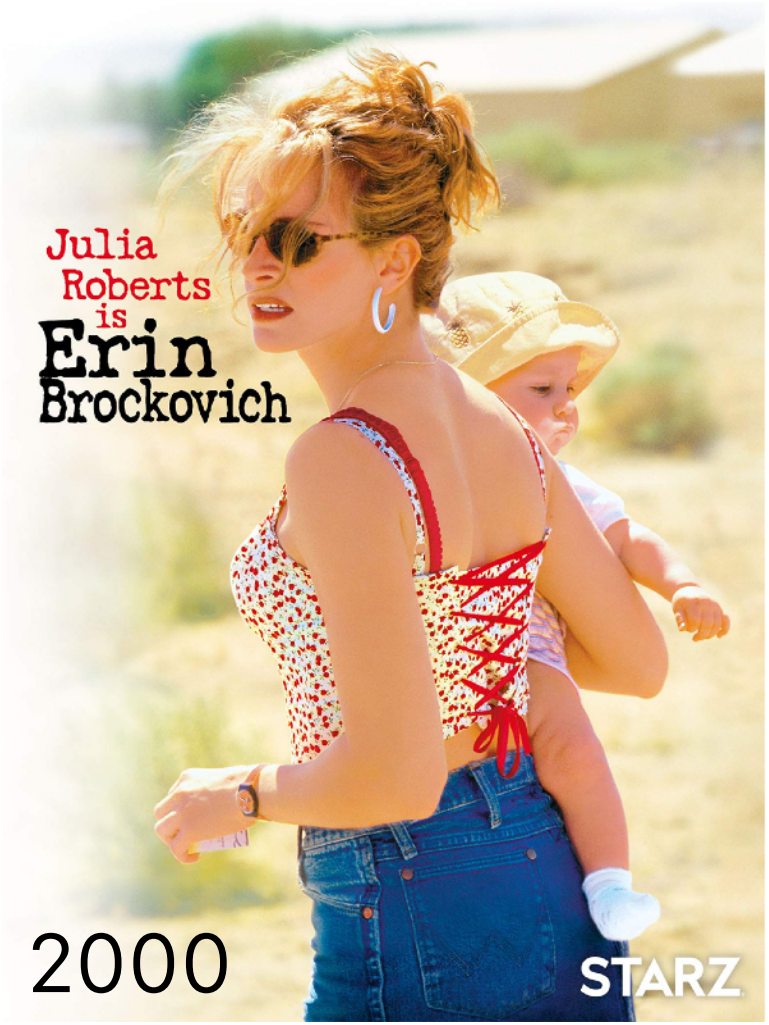
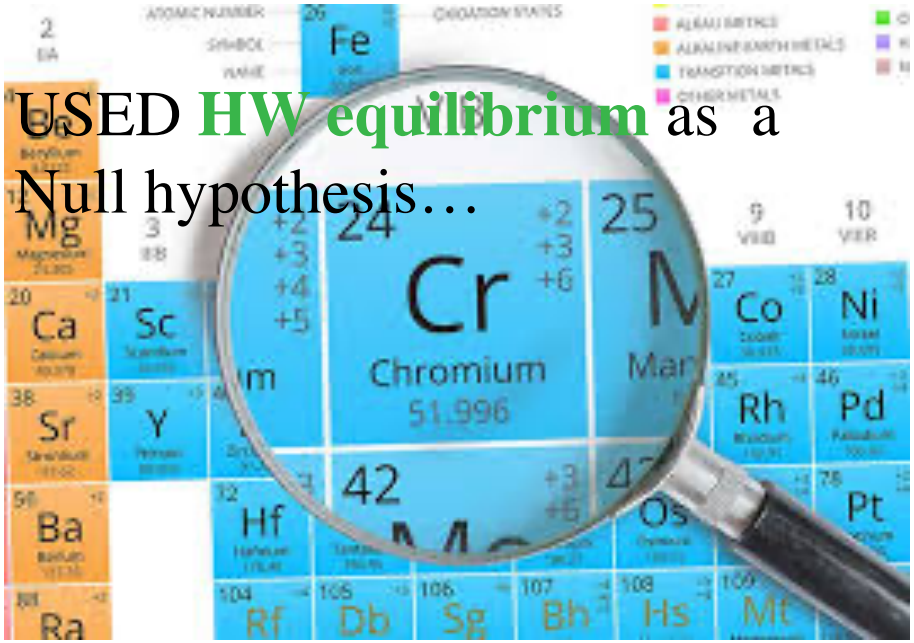


USED **HW equilibrium** as a Null hypothesis…



Erin Brokovich

Julia Roberts

Hardy–Weinberg equilibrium

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increased Cancer in Hinkley -caused by INCREASE in mutation rate ?