

POPULATION GENETICS

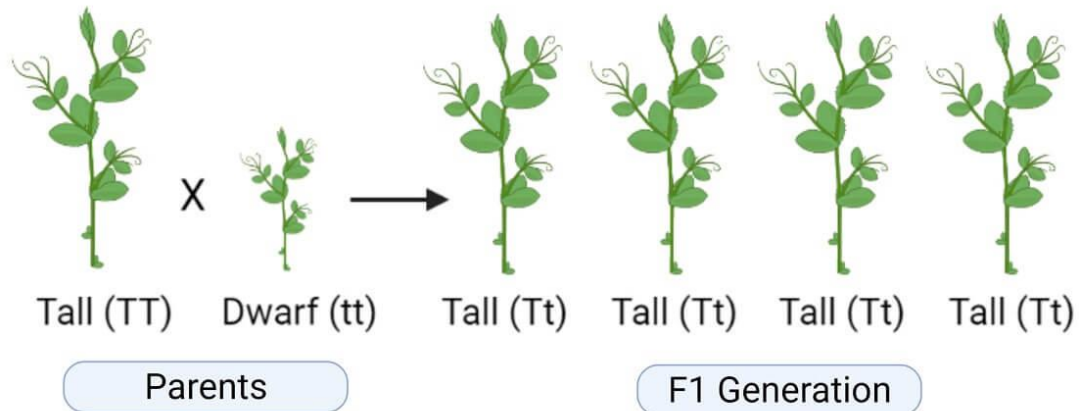
Anindita Bhadra

LS2201

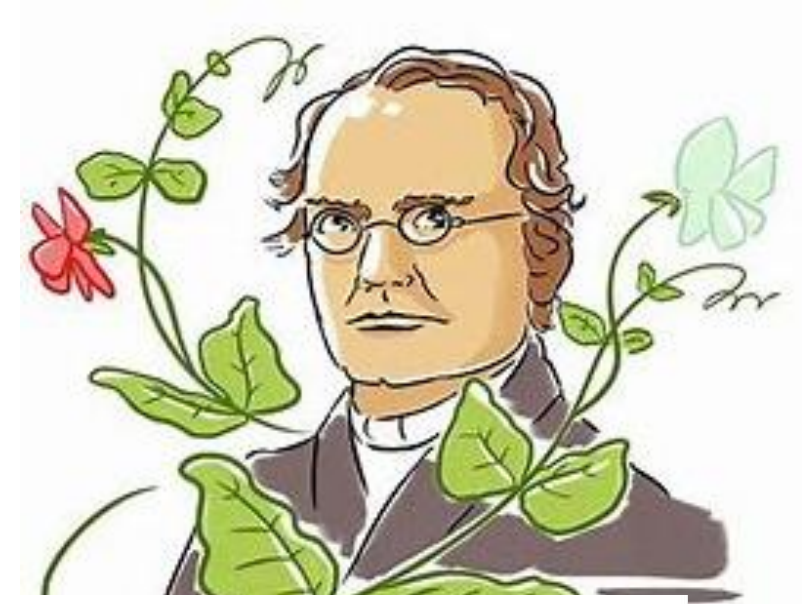


Mendelian Genetics

Mendel's Law of Dominance



Law of Dominance



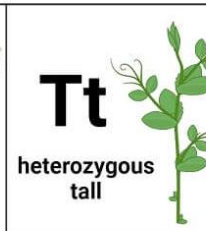
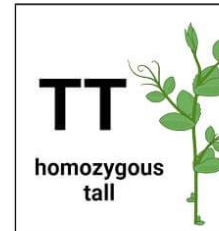
Heterozygous Tall (Tt)

T t

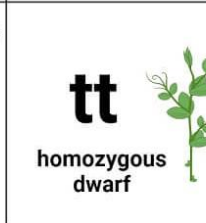
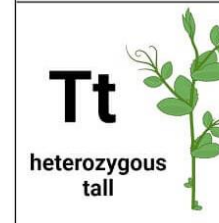


Heterozygous Tall (Tt)

T



t



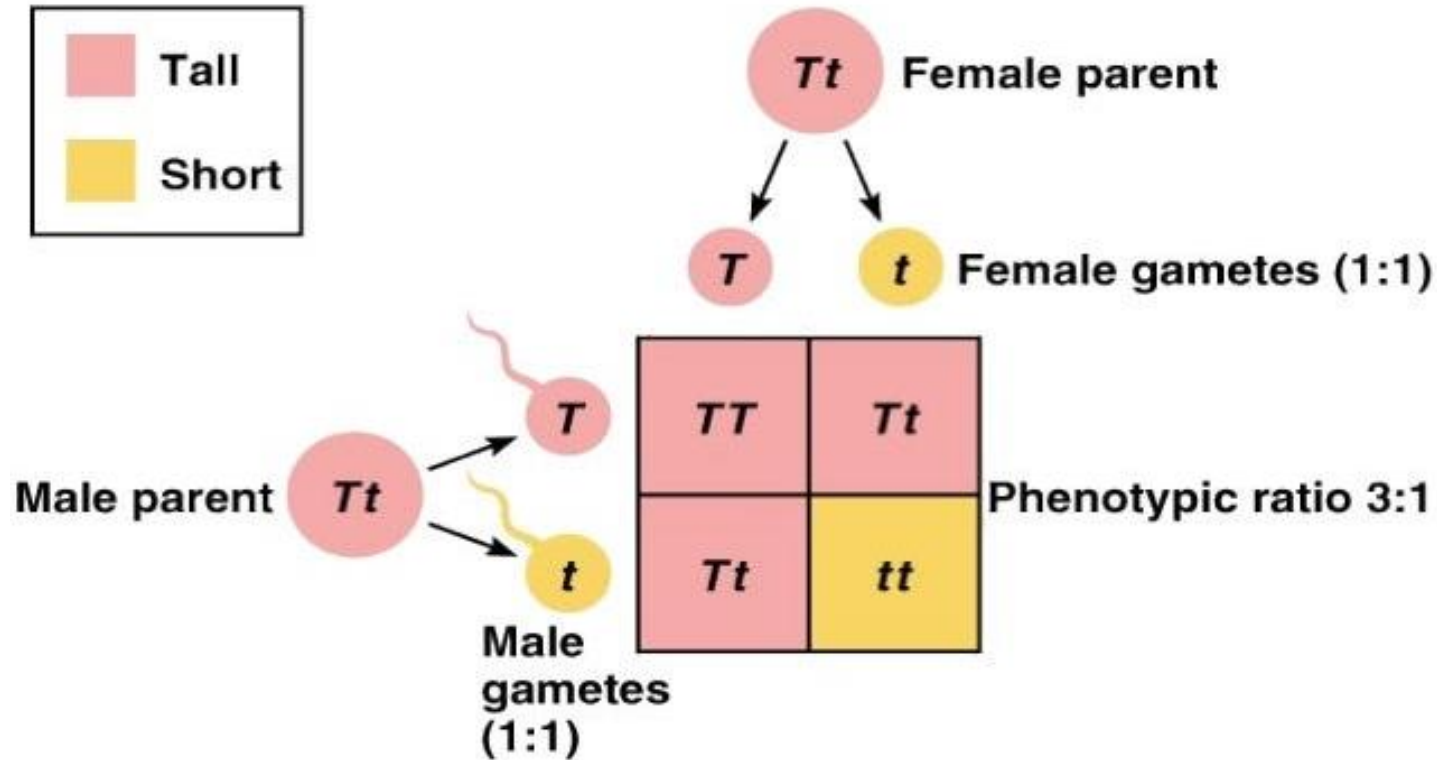
F2 Generation

phenotypic ratio = 3:1

genotypic ratio = 1:2:1

tallness = dominant character

Mendelian Genetics



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Law of Segregation

Mendelian Genetics

“Genes for different traits can segregate independently during the formation of gametes”

“The Law of **Independent Assortment**, said that separate genes for separate traits are passed independently of one another from parents to offspring”

Diagram illustrating a dihybrid cross between two heterozygous individuals ($AaBb \times AaBb$).

Parents: $AaBb$ (yellow) \times $AaBb$ (yellow)

Gametes: AB, Ab, aB, ab

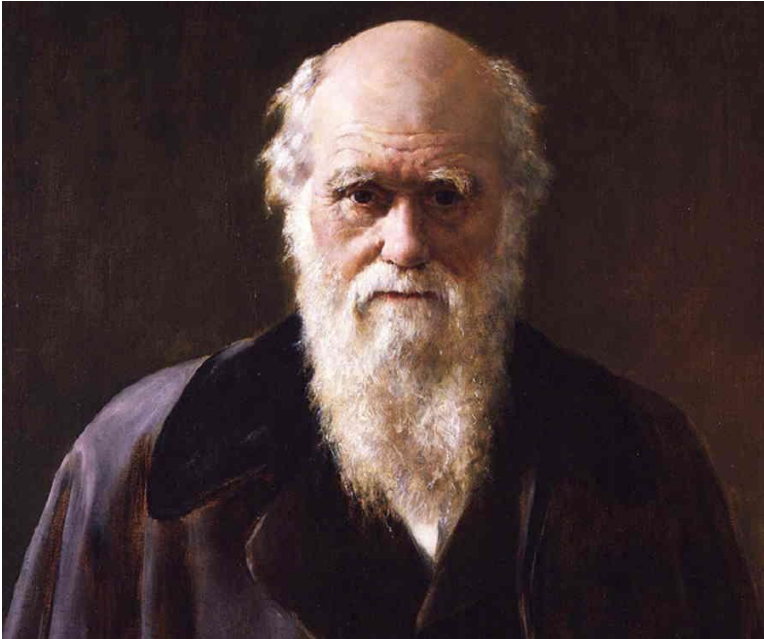
| | AB | Ab | aB | ab |
|----|--------|--------|--------|--------|
| AB | AA, BB | AA, Bb | Aa, BB | Aa, Bb |
| Ab | AA, Bb | AA, bb | Aa, Bb | Aa, bb |
| aB | Aa, BB | Aa, Bb | aa, BB | aa, Bb |
| ab | Aa, Bb | Aa, bb | aa, Bb | aa, bb |

| | AR | Ar | aR | ar |
|----|--------|--------|--------|--------|
| AR | Yellow | Yellow | Yellow | Yellow |
| Ar | Yellow | Yellow | Yellow | Yellow |
| aR | Yellow | Yellow | Green | Green |
| ar | Yellow | Yellow | Green | Green |



Law of Independent Assortment

Mendel and Darwin



**Variation
(1859)**

**Particulate
inheritance
(1866)**



Hugo de Vries (1848-1935), Carl Correns (1864-1935), and Erik Tschermak (1871-1962)

Wilhelm Johannsen (1909) coined the word Gene

Mendel and Darwin

In the end of the 19th century, two camps of thought emerged in England, which were further divided with the re-discovery of Mendel's work.

The **Biometricians** argued about the importance of continuous variation.

The **Mendelians** argued that most heritable characters were discontinuous.

This dispute led to a lot of excellent research, but the rift continued between the two schools of thought known as the **mutationists** and the **selectionists**.

Mendel and Darwin



William Bateson



R. A. Fisher



Francis Galton

The Modern Synthesis



R. A. Fisher



Sewall Wright



J B S Haldane

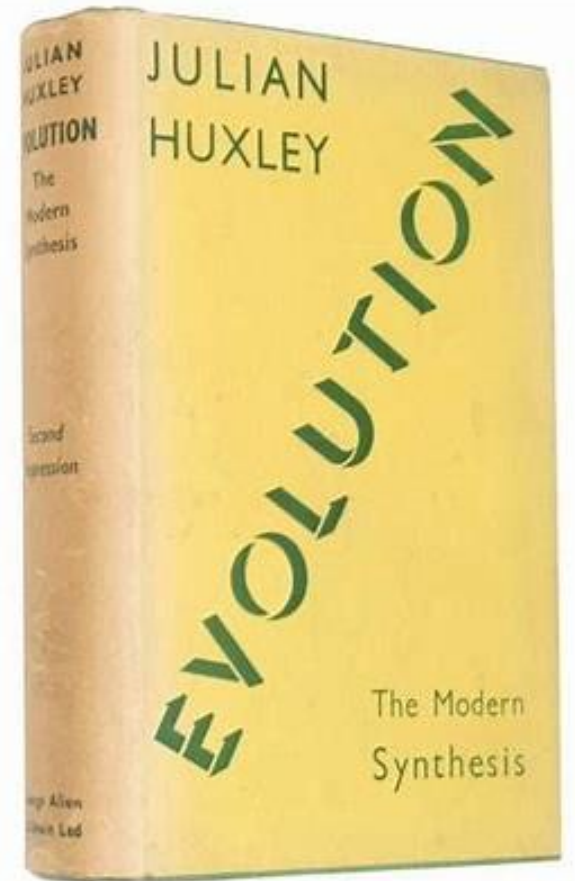
Populations rather than individuals should be the focus of evolution.

Gene frequencies are important, and not just the presence or absence of genes.

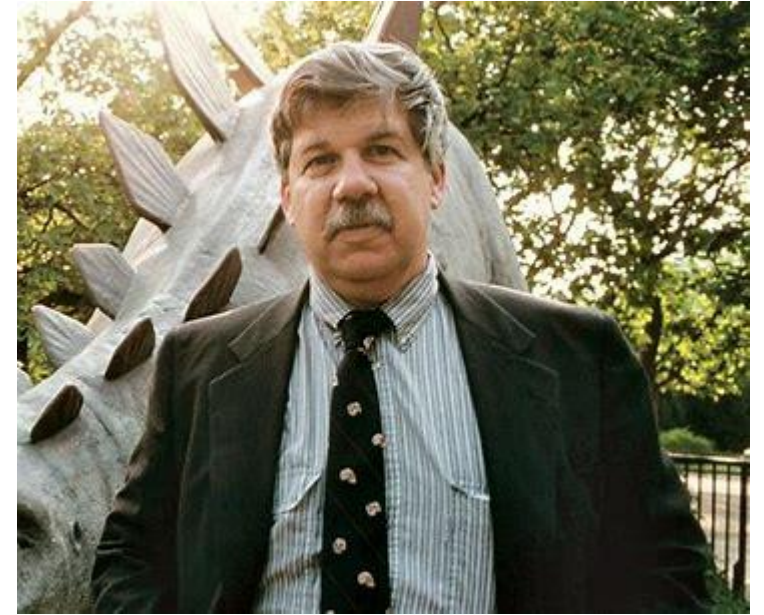
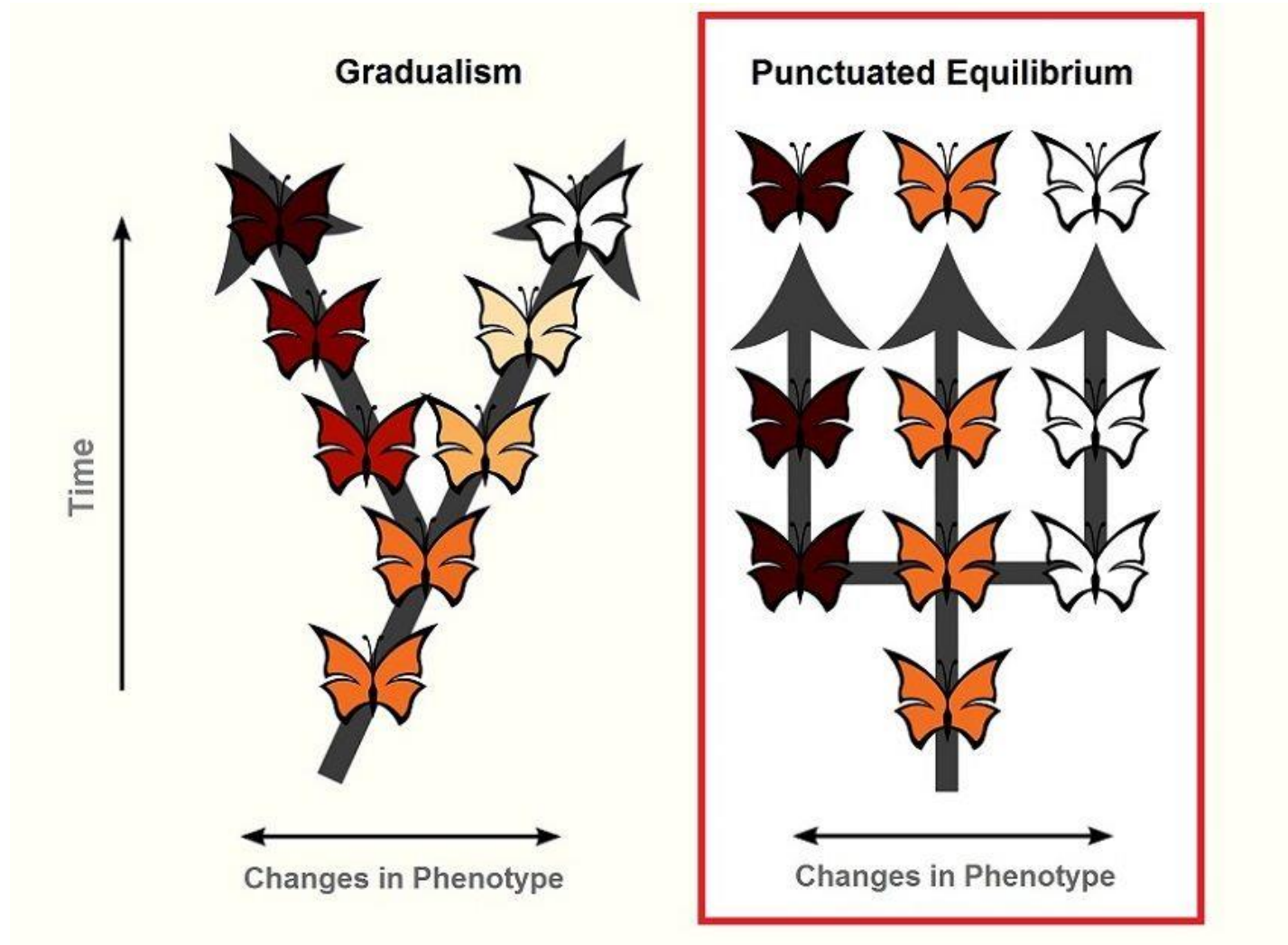
1930s to 40s

The Modern Synthesis – Key features

- (1) genetic variation is the source of phenotypic variation;
- (2) this variation arises due to mutations that are random with respect to fitness;
- (3) accumulated mutations selected over time are the basis of evolution within a taxon (gradualism);
- (4) adaptation is solely the result of natural selection; and
- (5) evolution occurs at the population level (Futuyma 2017)



The Modern Synthesis – Critique



Stephen Jay Gould

The Population

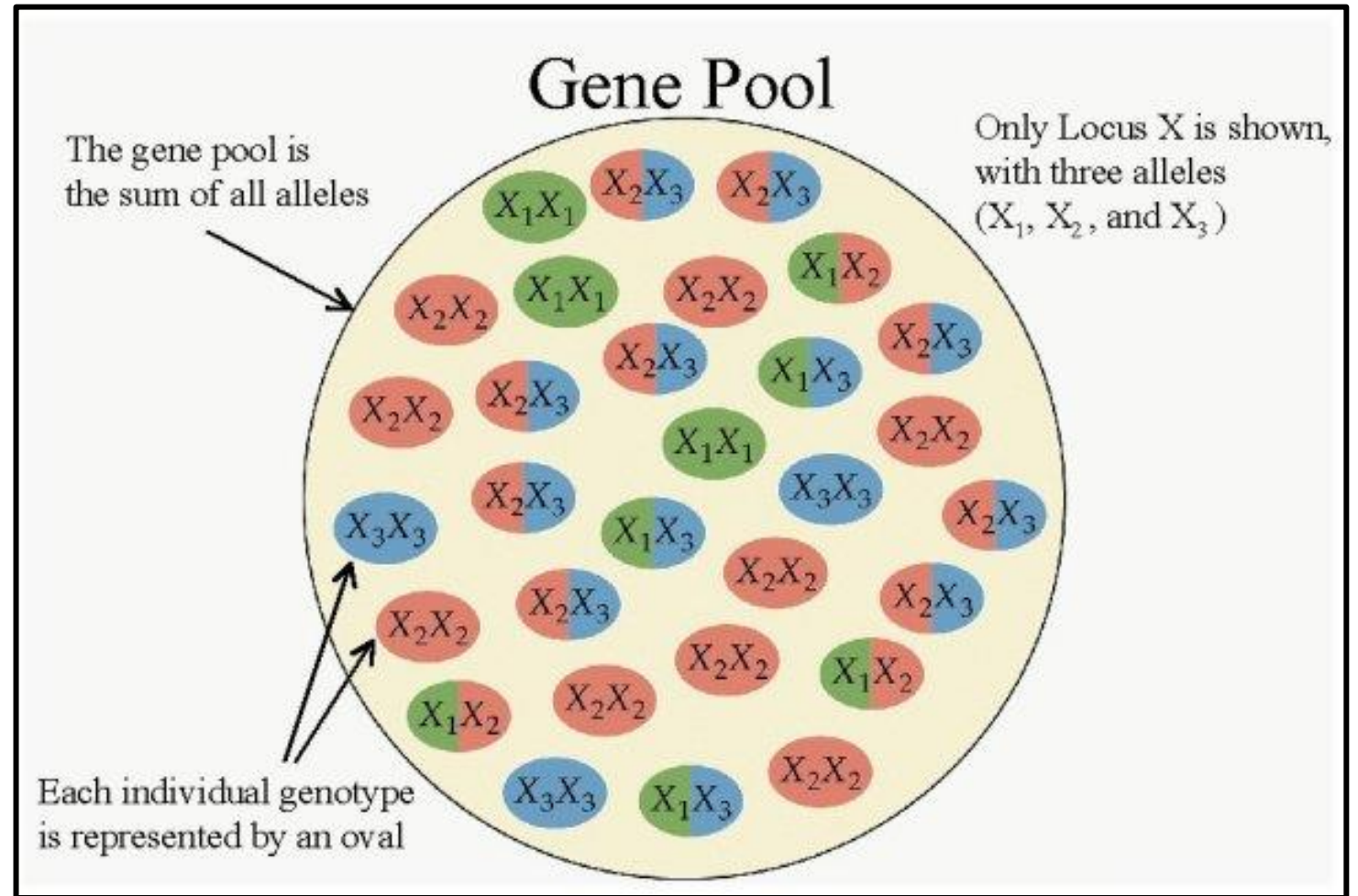
A group of sexually interbreeding or potentially interbreeding individuals. Since Mendelian laws apply to the transmission of genes among these individuals, Sewall Wright has called such a group a **Mendelian population**.

The size of a population may vary, but it is usually considered to be a local group (deme), each member of which has an equal chance of mating with any other member of the opposite sex. Populations have two important attributes – **gene frequencies** and a **gene pool**.

Gene (allelic) frequencies are simply the proportion of the different alleles of a gene in a population.

The Population

The gene pool is the sum total of genes at all loci in every member of an interbreeding population. It can be considered as a **gametic pool** from which samples are drawn at **random** to form the zygotes of the next generation.



True?

**In a Mendelian population, dominant alleles will
always dominate the population**



**There will be more individuals with the dominant
phenotype.**

The Hardy Weinberg Principle

In 1908, Hardy in England and Weinberg in Germany, disproved the argument that dominant alleles would dominate the population, following Mendelian inheritance.

They demonstrated that gene frequencies do not depend on dominance or recessiveness, but remain essentially unchanged from one generation to the next under certain conditions.

The theory describing the genotypic equilibrium in the population, based on stable gene frequencies and random mating is known as the Hardy-Weinberg principle.

The Hardy Weinberg Principle

Total no. of individuals = 36

Generation 1:

No. of OO individuals = 12

Frequency of OO = $12/36 = 0.333$

Similarly,

Frequency of OG = 0.333

Frequency of GG = 0.333

| Generation | OO | OG | GG |
|------------|----|----|----|
| 1 | 12 | 12 | 12 |
| 2 | 7 | 22 | 7 |
| 3 | 8 | 20 | 8 |

Frequency of allele O is p and allele G is q,

$$p = (2 \times \text{OO} + \text{OG}) / 2 \times 36$$

$$q = (2 \times \text{GG} + \text{OG}) / 2 \times 36$$

The Hardy Weinberg Principle

Hence,

$$p = [(2 \times 12) + 12] / 2 \times 36 = 0.5$$

$$q = [(2 \times 12) + 12] / 2 \times 36 = 0.5$$

Generation 2:

$$p = [(2 \times 7) + 22] / 2 \times 36 = 0.5$$

$$q = [(2 \times 7) + 22] / 2 \times 36 = 0.5$$

Generation 3:

$$p = [(2 \times 8) + 20] / 2 \times 36 = 0.5$$

$$q = [(2 \times 8) + 20] / 2 \times 36 = 0.5$$

The Hardy Weinberg Principle

Note,

p and q, i.e., the gene or **allelic frequencies remain unchanged** across generations, though the genotypic frequencies may change.

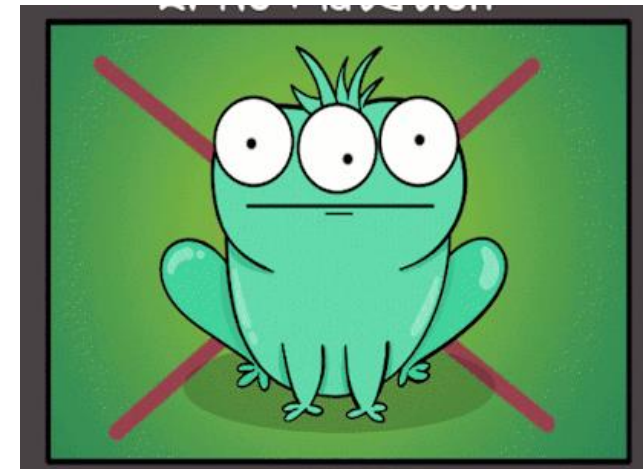
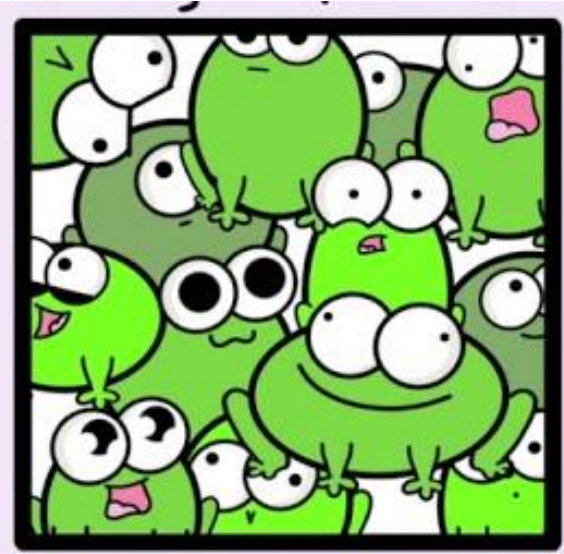
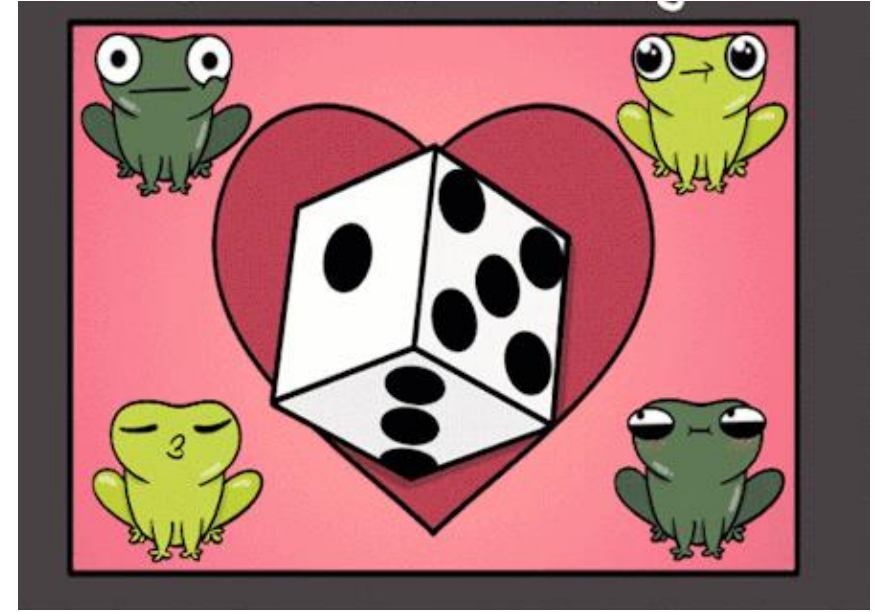
$$p + q = 1 \text{ and obviously, } (p + q)^2 = 1$$

Note: Genotypic frequencies for any gene with two alleles can be represented as p^2 , $2pq$ and q^2 .

The HWE - Conditions

- Random mating
- The population should be large (infinite)
- All genotypes are equally viable
- No gene flow (migration; mutation; genetic drift)
- No selection

Is this possible in real populations?



The HWE - Conditions



Does HWE work for such cases?

The Hardy Weinberg Principle – two loci

Equilibrium is attained in one generation when a single locus is considered, independent of genes at other loci.

When we consider two or more loci, then their interactions give rise to more possible genotypes.

For example, consider two gene pairs Aa and Bb. The number of possible genotypes are $3^2 \rightarrow$ AABB, AABb, AaBB, AaBb, aaBB, aaBb.

The Hardy Weinberg Principle – two loci

Let the frequency of the alleles be p, q, r, s for A, a, B and b respectively.

The equilibrium ratios of their genotypes are: $(pr + ps + qr + qs)^2$



AB and ab gametes are non-recombinant or **coupling**.

Ab and aB gametes are recombinant or **repulsion**.

The Hardy Weinberg Principle - two loci



| | |
|-----------|------|
| p^2r^2 | AABB |
| $2p^2rs$ | AABb |
| $2p^2s^2$ | Aabb |
| $2pqr^2$ | AaBB |
| $4pqrs$ | AaBb |
| $2pqs^2$ | Aabb |
| Q^2r^2 | aaBB |
| $2q^2rs$ | aaBb |
| q^2s^2 | aabb |

The problem of attainment of equilibrium resolves itself to the time that it takes for the gametic frequencies to reach these values.

If we begin with only the heterozygotes (AaBb X AaBb) in which the frequencies of all genes are the same ($p=q=r=s=0.50$), all four types of gametes are immediately produced at equilibrium frequencies (0.25). Genetic equilibrium is reached within one generation.

The Hardy Weinberg Principle - two loci



However, this is a special case.

Frq of A = 0.6 (p)

Frq of B = 0.6 (q)

Frq of a = 0.4 (r)

Frq of b = 0.4 (s)

AABB = 0.30

Aabb = 0.30

aabb = 0.30

aaBB = 0.10

| Equilibrium frequency of gametes | | | | |
|----------------------------------|---------|-------------|-----------------|------------------|
| | Gametes | | | |
| Initial popln | Type | Initial Frq | Equilibrium Frq | |
| 30% AABB | AB | 0.30 | 0.30 – d | 0.6 X 0.4 = 0.24 |
| 30% Aabb | Ab | 0.30 | 0.30 + d | 0.6 X 0.6 = 0.36 |
| 30% aaBB | aB | 0.10 | 0.10 + d | 0.4 X 0.4 = 0.16 |
| 10% aabb | ab | 0.30 | 0.30 - d | 0.4 X 0.6 = 0.24 |

The Hardy Weinberg Principle - two loci



At equilibrium: $Ab \times ab = Ab \times aB$

If coupling and repulsion products in the initial population differ, this difference represents the change in gametic frequencies that must occur for equilibrium. This is called **disequilibrium (d)**.

$$\mathbf{d = (AB) (ab) - (Ab) (aB) = (0.3 \times 0.3) - (0.3 \times 0.1) = 0.06}$$

Inbreeding

Inbreeding occurs when individuals selectively mate with their relatives.

In such cases, gene copies in uniting gametes are more likely to be identical by descent than if they joined at random.

Due to inbreeding, the frequency of each homozygous genotype increases and the frequency of heterozygotes decreases by the same amount.

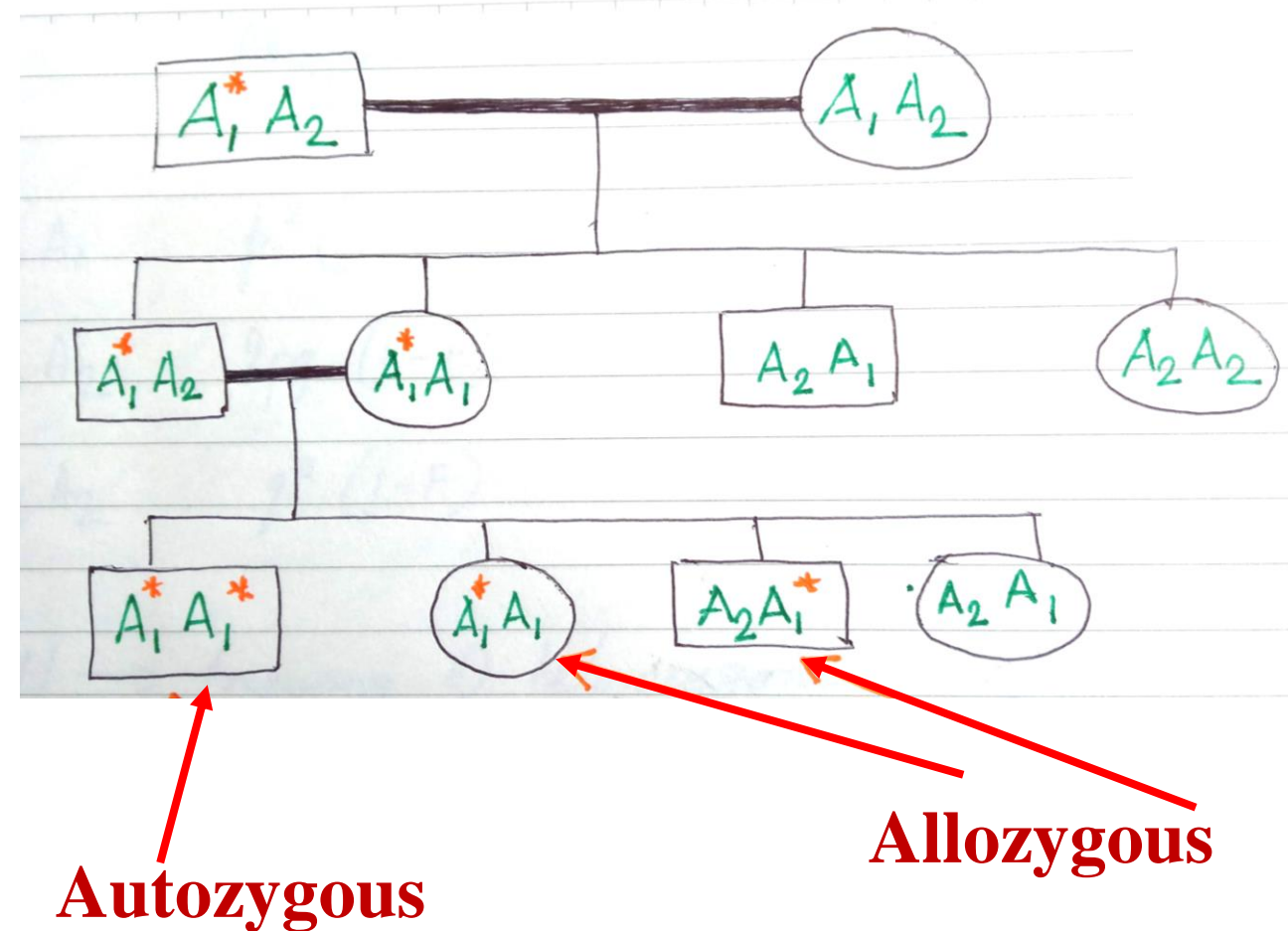
The frequency of heterozygotes is $H = H_0(1-F)$, where H_0 is the heterozygote frequency expected if the locus were in HWE, and F is the **inbreeding coefficient**.

Inbreeding

Self fertilization is the most extreme form of inbreeding, which occurs in many species of plants.

Inbreeding coefficient (F) is the probability that an individual taken as random from the population will be autozygous.

In a completely outbred population, $F = 0$.



Inbreeding

| | Allozygous | Autozygous | Genotype frequency | |
|----------|-------------|------------|--------------------|-------|
| A_1A_1 | $p^2 (1-F)$ | $+ pF$ | $= p^2 + Fpq$ | $= D$ |
| A_1A_2 | $2pq (1-F)$ | $+ 0$ | $= 2pq (1-F)$ | $= H$ |
| A_2A_2 | $q^2 (1-F)$ | $+ qF$ | $= q^2 + Fpq$ | $= R$ |

$H \rightarrow$ frequency of heterozygotes in the inbred population

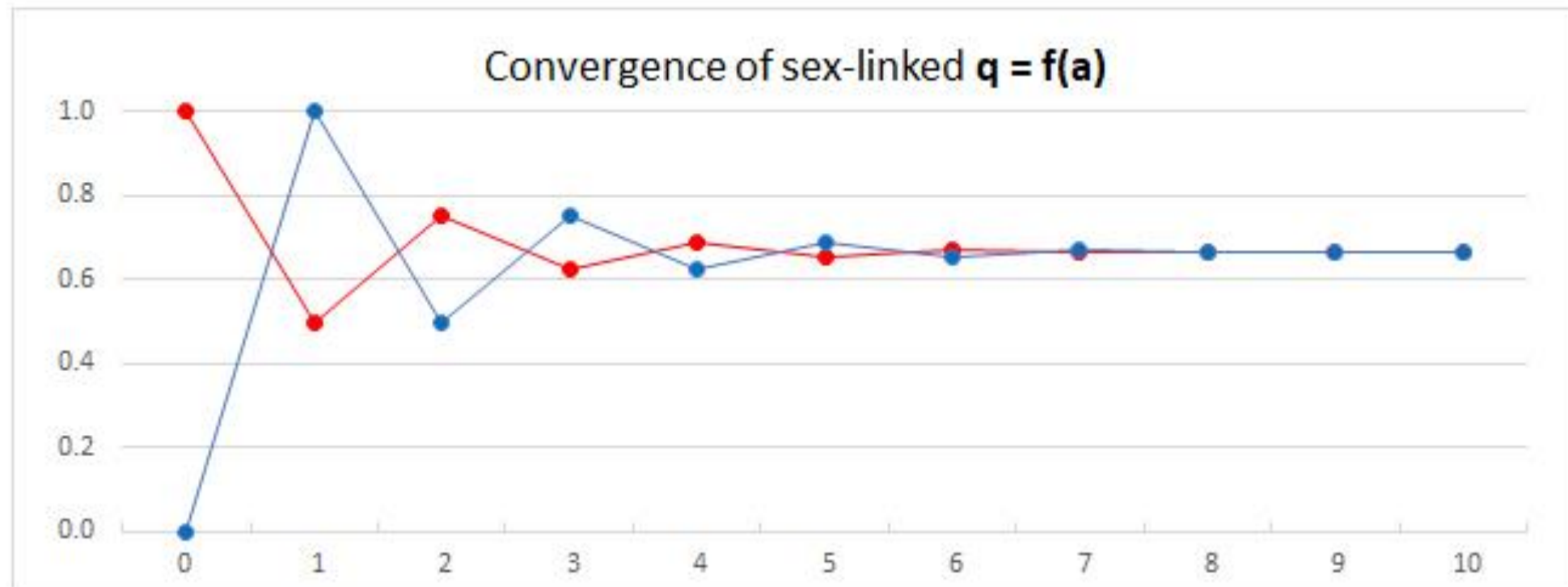
$H_0 = 2pq \rightarrow$ expected frequency of heterozygotes in the population in HWE

$$H = H_0 (1-F)$$

$$F = (H_0 - H) / H_0$$

Sex-linked loci

| | 0 | 1 | 2 | 3 | 4 | 5 | 6 | 7 | 8 | 9 | 10 |
|--------|--------|--------|--------|--------|--------|--------|--------|--------|--------|--------|--------|
| $f(f)$ | 1.0000 | 0.5000 | 0.7500 | 0.6250 | 0.6875 | 0.6563 | 0.6719 | 0.6641 | 0.6680 | 0.6660 | 0.6670 |
| $f(m)$ | 0.0000 | 1.0000 | 0.5000 | 0.7500 | 0.6250 | 0.6875 | 0.6563 | 0.6719 | 0.6641 | 0.6680 | 0.6660 |
| mean' | 0.6667 | 0.6667 | 0.6667 | 0.6667 | 0.6667 | 0.6667 | 0.6667 | 0.6667 | 0.6667 | 0.6667 | 0.6667 |



Sex-linked loci

Consider a sex-linked locus in a species where females are XX (homogametic) and males are XY (heterogametic). Suppose allele frequencies in females and males are initially unequal.

The frequency of the allele $f(a)$ in females of generation n necessarily determines $f(a)$ in males in generation $n+1$. The male $f(a)$ therefore "chases" the female $f(a)$ in the preceding generation until they reach approximate equality.

Because each female in generation $n+1$ receives an X chromosome for each parent in generation n , the female $f(a)$ is the mean of the male and female $f(a)$ in the preceding generation.

Sex-linked loci

Calculation of allele frequencies for an X-linked locus requires a bit of caution, as males are hemizygous for this locus. But the same rules apply. Simply count males as having only one allele for each frequency calculation.

EXAMPLE:

In a population of squirrels, a recessive allele of an X-linked locus (R) codes for a white star on the forehead (r).

Females:

XRXR - no star

XRXr - no star

XrXr - star

Sex-linked loci

The dominant allele occurs in
RR females x 2 (since each one carries two alleles)
heterozygous females (each of whom carries one R allele)
starless males (each of whom carries one R allele)

The recessive allele occurs in:
rr females x 2 (since each one carries two alleles)
heterozygous females (each of whom carries one r allele)
starred males (each of whom carries one r allele)

In our population of 1000 squirrels, there are (conveniently!) 500 females and 500 males.

Sex-linked loci

But unlike an autosomal trait, which would have 2000 copies in this population, the X-linked trait has only 1500 copies due to the hemizyosity of the males.

In our population, we counted:

460 unstarred females ($XRX-$)

40 starred females ($XrXr$)

300 unstarred males (XRY)

200 starred males (XrY)

Sex-linked loci

In the recessive homozygous females, $q^2 = 40/1000$ (0.04), so $q = 0.2$.

In the hemizygous males, the frequency of q is $200/1000$ (0.2).

The summed frequency of q in the expressing individuals is $(0.2 + 0.2 = 0.4)$.

Solving for p , the expected frequency of the dominant allele should be $1.0 - 0.4 = 0.6$

Sex-linked loci

Since the total number of alleles in the population is only 1500, this means that the expected relative frequencies of R and r should be:

1500×0.6 , or 900 R alleles

and

1500×0.4 , or 600 r alleles

Sex-linked loci

You know from your census that

40 starred females carry 80 r alleles

200 starred males carry 200 r alleles

→ for a total of 280 of the 600 r alleles in the population.

That means the remaining unaccounted 320 r alleles ($600 - 280 = 320$) must be "hiding" in the heterozygous females. Therefore, 320 of your 460 unstarred females are expected to be heterozygous (each has one copy of r) for the recessive "starring" allele (r) if the population is in HW equilibrium.