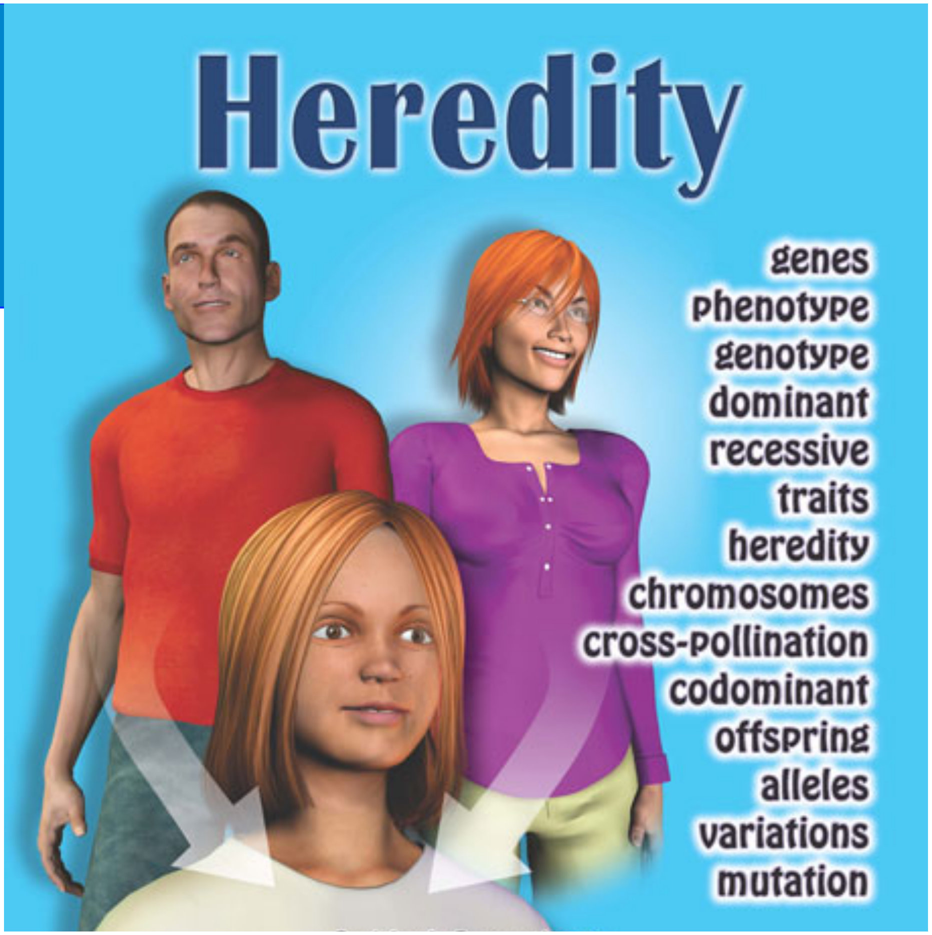
POPULATION GENETICS

G. H. Hardy

Wilhelm Weinberg

POPULATION GENETICS

- **Population** = a group of individuals of the ***same species occupying a given area*** at a certain time
- **Genetics** = the study of ***heredity***

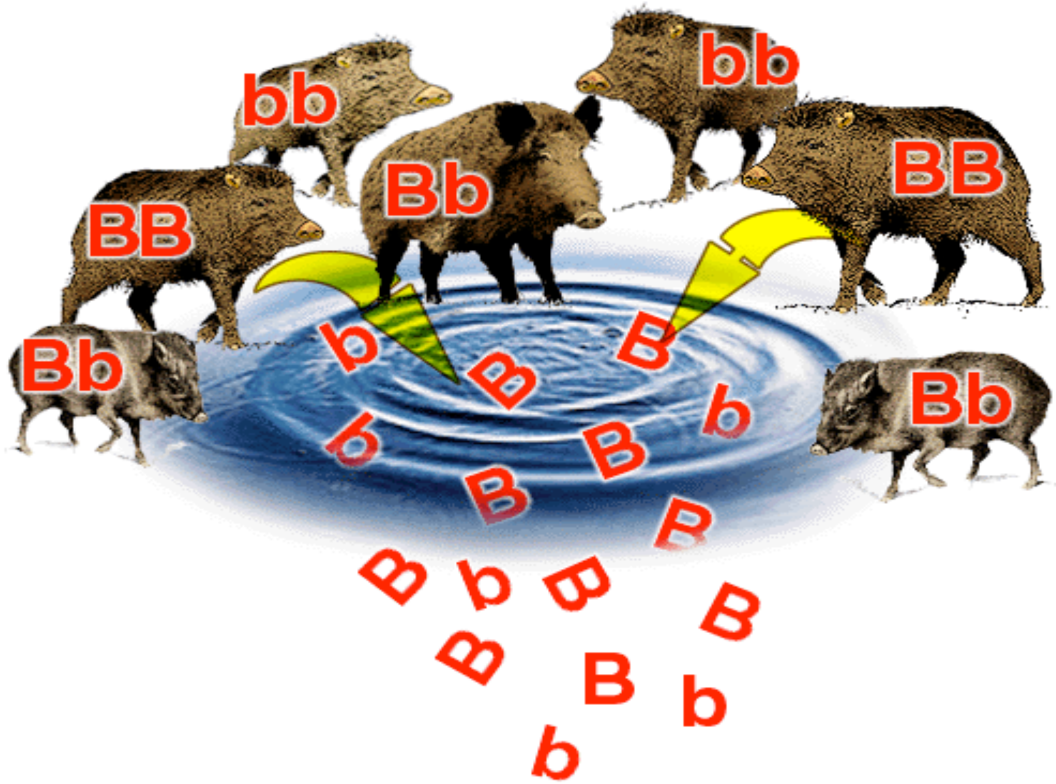


Population Genetics = the study of heredity within a given population

Species

- **Species** = a group of populations whose individuals have the potential to interbreed and produce fertile offspring in nature
  - Members of a population are more likely to breed within the population, so genes tend to stay in the **same population** for generations

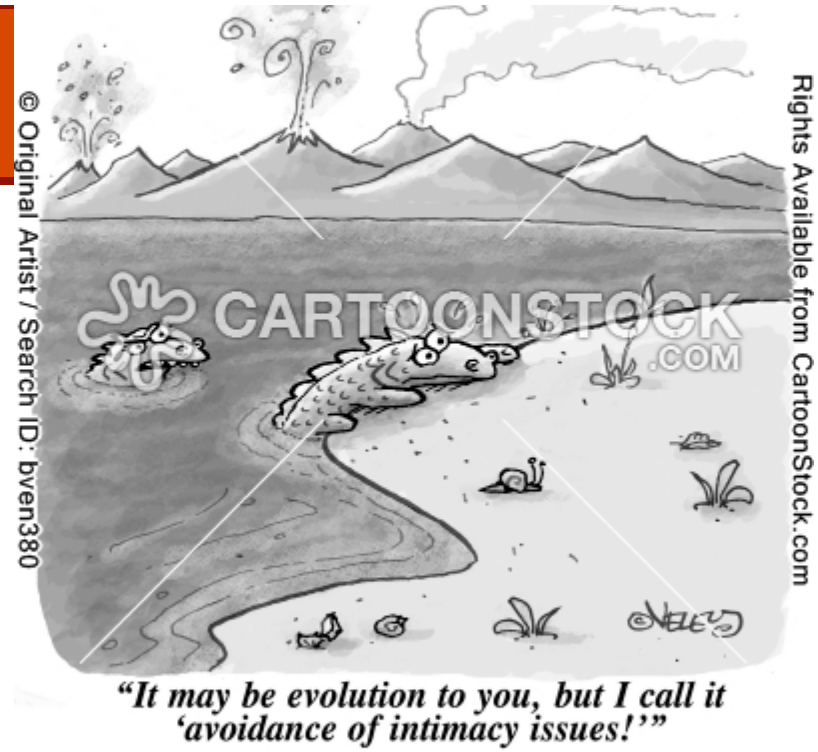
<u>Gene pool-</u> all the genes in all the members of a population at one time. Immigration increases the gene pool and emigration decreases it.



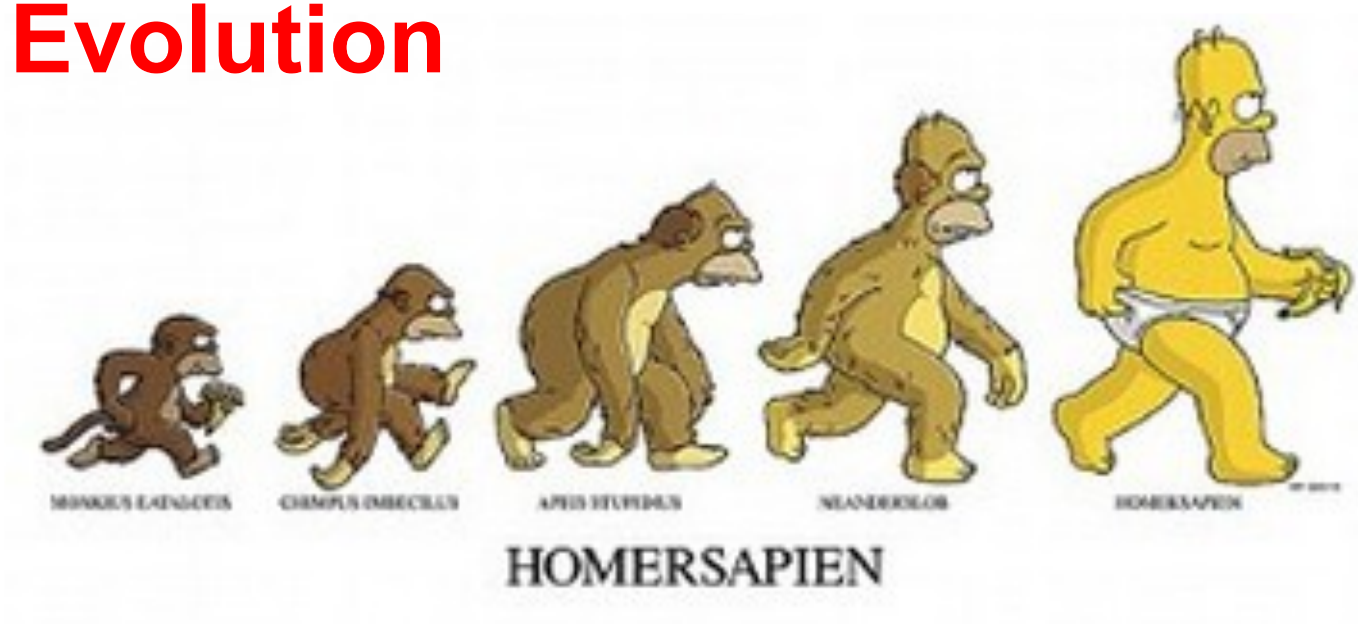
Evolution

- **Evolution** is the gradual change in the frequency of genes in a gene pool

Natural Selection is the basis for…
Evolution



"It may be evolution to you, but I call it 'avoidance of intimacy issues!'"



Natural Selection

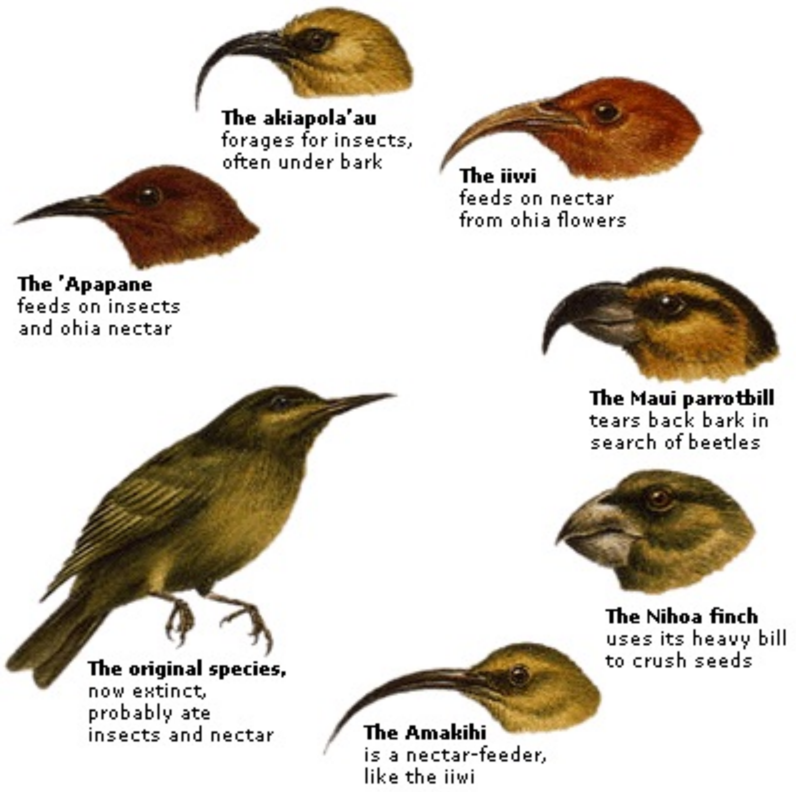
- **Natural selection** → some traits are better equipped for ***survival*** in a given environment
  - They are "naturally selected" for through ***survival of the fittest***

Example of natural selection:

Darwin's finches

•Different beak shapes for different food sources.

Each beak specialized for unique food source



The Importance of Variation

- **Variation** among organisms may be in
  - **Physical appearance**
  - **Metabolism**
  - **Fertility**
  - **Mode of reproduction**
  - **Behavior**

Variation amongst organisms is dependent on **variation in genes.**

Albatross' elaborate and unique courtship behaviour

Variation as a Means for Survival

- Certain genotypes are better equipped than others for survival
- Sexual reproduction ensures **variations get passed on to offspring**
- Leads to **natural selection** for individuals in given conditions.

Studying Human Genetic Traits

- Studying humans is problematic
    - Few offspring
    - Slow reproductive time
    - Environment affects phenotype
- How do we study human populations then?
    - **POPULATION SAMPLING!!**

Population Sampling

1. Select **a small sample** of individuals from the population
2. Find the **gene frequencies** for a particular genetic trait in that sample
3. Apply gene frequencies to the **whole population**
4. This allows scientists to analyze

trends over time

Electro-shocking for population sample

Gene Frequency

•Geneticists have used **gene frequencies** to study changes in the human population

•Example Blood type in North America

-Type 0 blood is most common, whereas Blood type AB is considered rare

-Recessive Rh- alleles are found only in 15 % of Canadians

Types of Gene Frequency

Genotype Frequency -is the proportion of a population with a particular **genotype** (expressed as a decimal)

Phenotype Frequency -is the proportion of a population with a particular **phenotype** (expressed as a decimal or %)

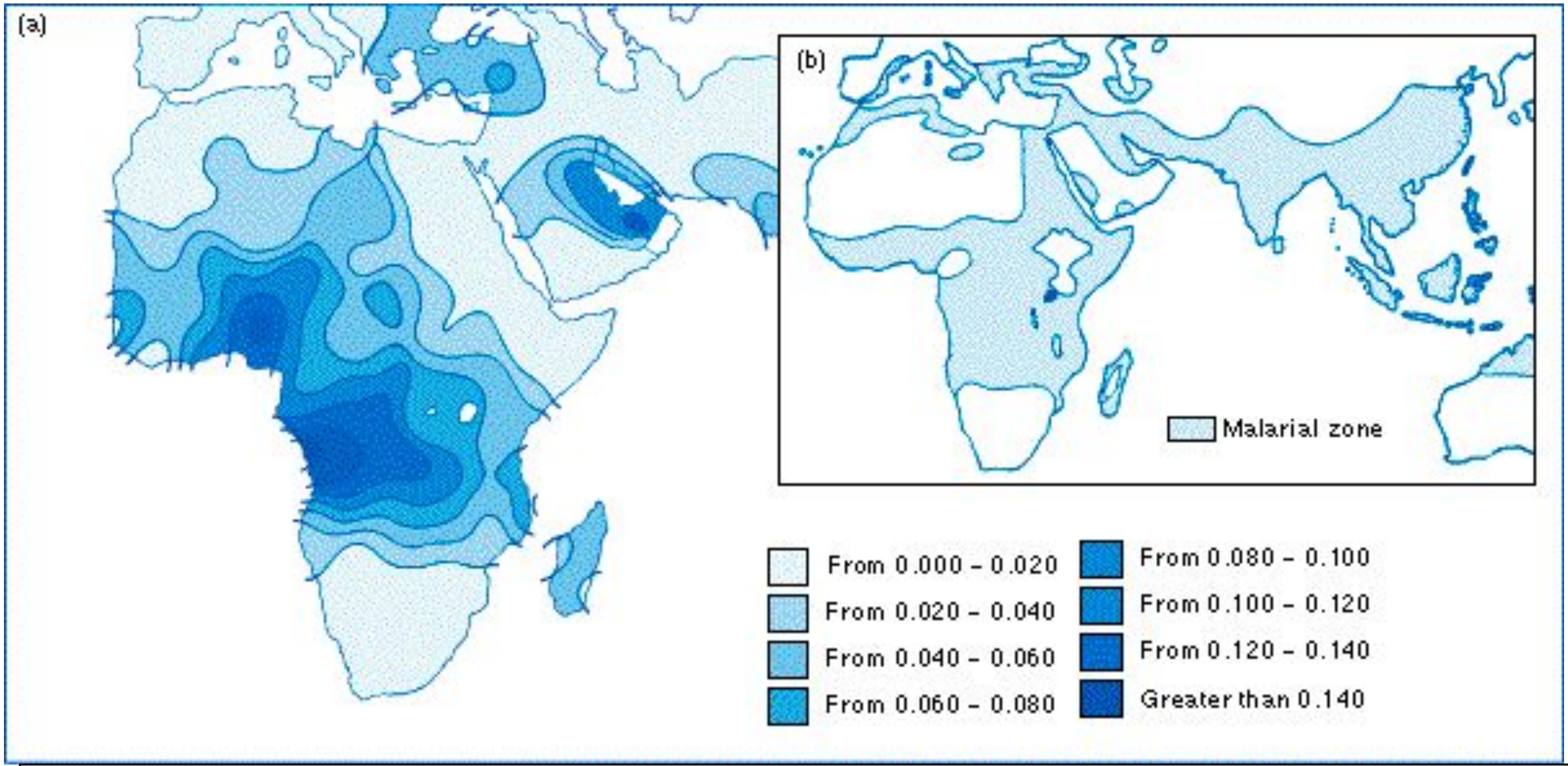
Allele Frequency -is the rate of occurrence of a particular allele in a population with respect to a particular gene. (usually expressed as decimal)

Gene Frequencies

- Sometimes, gene frequencies are associated with certain populations
  - Swedish people mostly blond haired and blue eyed
  - We say frequency of blond hair and blue eyes is **HIGH** in this population

Gene Frequencies

The recessive allele that causes Sickle cell anemia has different frequencies throughout Africa. The heterozygous condition provides immunity to malaria.

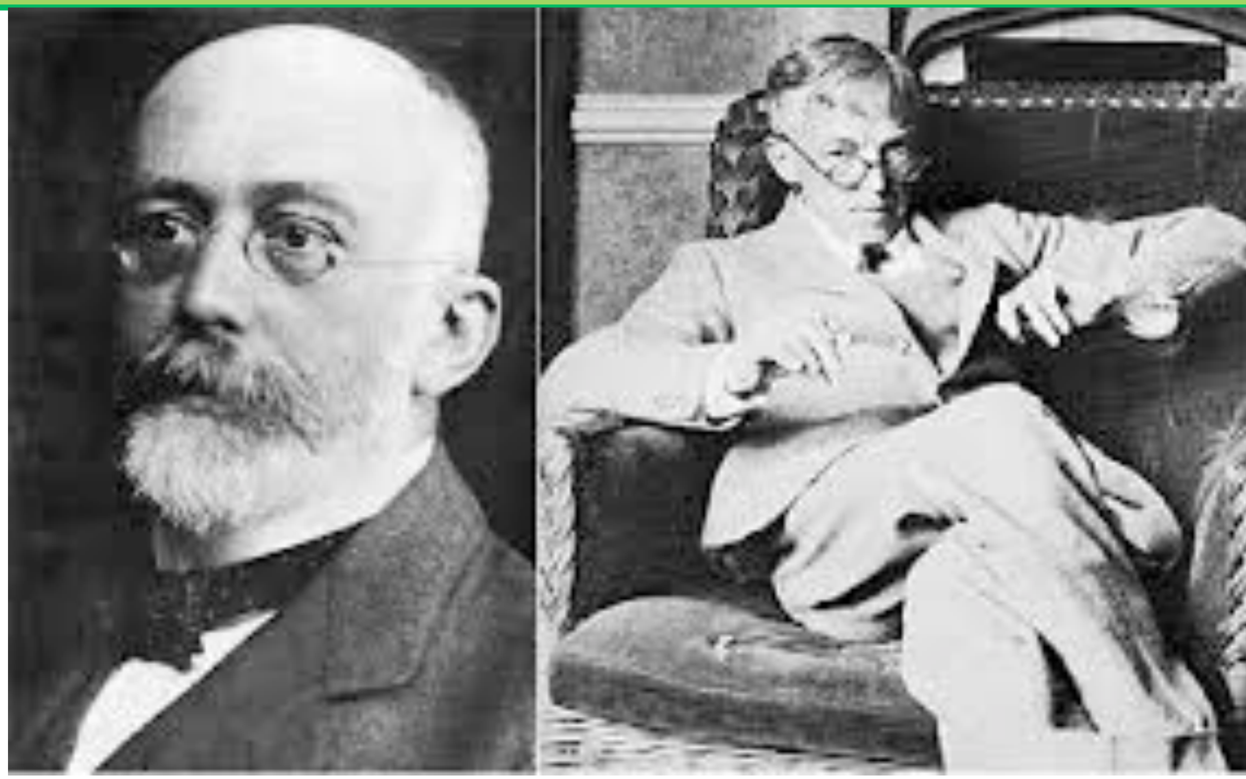


Sickle cell trait is hypothesized to have evolved because of the vital protection from malaria it provides.

When studying GENE FREQUENCIES…

Must use…

HARDY WEINBERG EQUILIBRIUM!!!



Mathematician and Geneticist

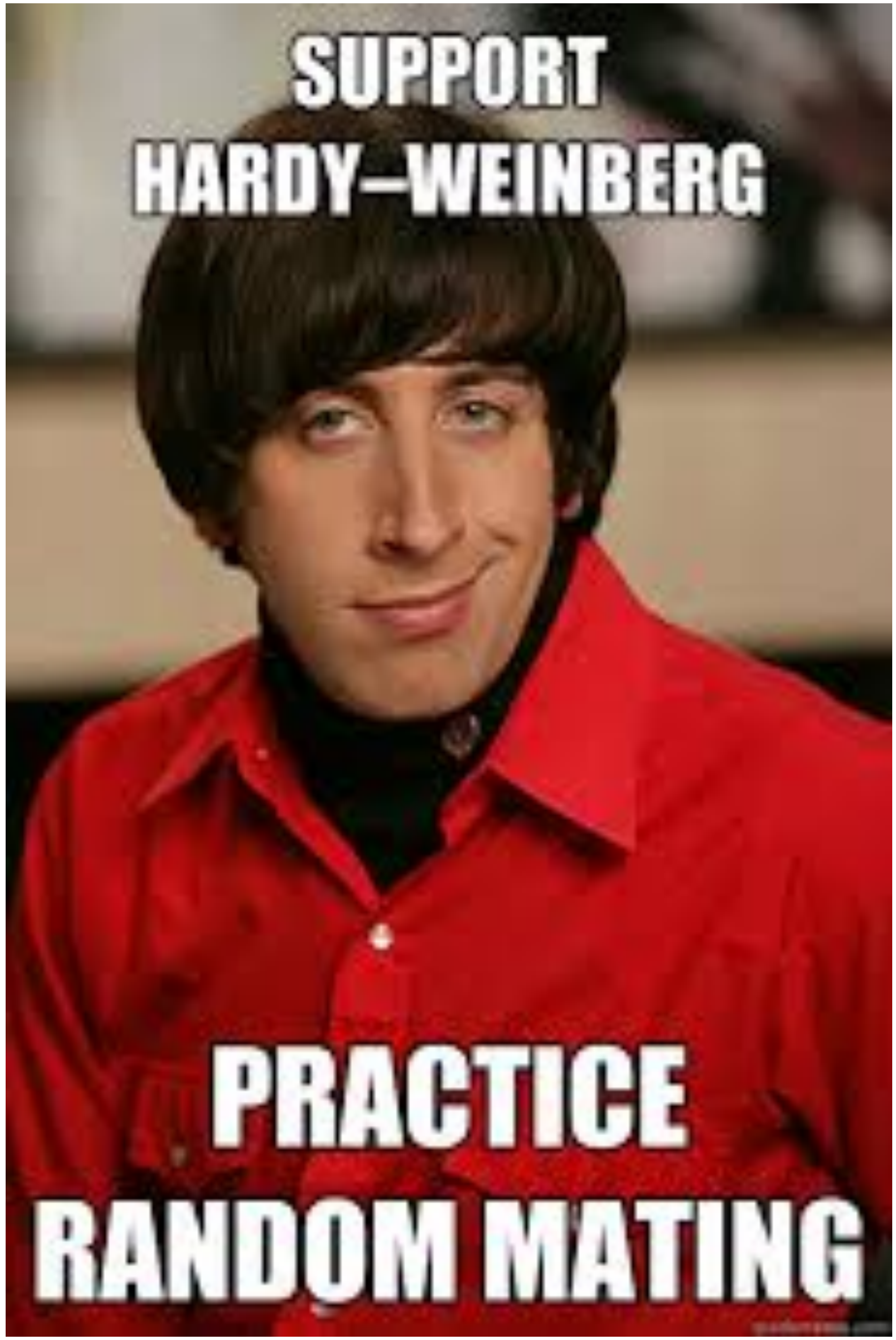
Hardy Weinberg Equilibrium

Under specific conditions…

allele and genotype frequencies(gene pool) in a population will remain stable from generation to generation in the absence of other evolutionary influences

Conditions necessary for HW equilibrium:

1. Large population
2. Random mating
3. No genetic drift (disappearance of genes due to individuals dying or not reproducing)
4. No gene flow - migration
5. No natural selection
6. No mutations


SUPPORT
HARDY–WEINBERG
PRACTICE
RANDOM MATING

RULE 1: The sum of all alleles must equal 1

Frequency of Dominant Allele	p	A, B, Z
Frequency of Recessive Allele	q	a, b, z

$$p + q = 1$$

In a population the frequency of which "p" (dominant) occurs plus the frequency that "q" (recessive) occurs must add up to 1

Just as many R as r with means each allele is 50% of the total amount of alleles
.5 + .5 = 1
So… p + q = 1

	R	r
R	RR	Rr
r	Rr	rr

(eg) .6 + .4 = 1 or .6 + .4 = 1

So out of 120 people, 48 would have the recessive allele
.4 X 120 = 48

So out of 120 people, 72 would have the dominant allele
.6 X 120 = 72 or…
120 – 48 = 72

GENOTYPE FREQUENCY

Frequency of **HOMOZYGOUS DOMINANT** ALLELE
"**AA**"

Frequency of **HOMOZYGOUS RECESSIVE** ALLELE
"**aa**"

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

Frequency of **HETEROZYGOUS ALLELE**
"**Aa**"

THE WHY

Heterozygous Punnett

	R **(p)**	r **(q)**
R **(p)**	RR **(p^2)**	Rr **(pq)**
r **(q)**	Rr **(pq)**	rr **(q^2)**

FORMULA SUMMARY

Alleles: $p + q = 1$
A a

Genotypes: $p^2 + 2pq + q^2 = 1$
AA Aa aa

p = frequency of dominant **allele** in a population. **(A)**

q = frequency of recessive **allele** in a population. **(a)**

p^2 = frequency of homozygous dominant **genotype**. **(AA)**

$2pq$ = frequency of heterozygous **genotype**. **(Aa)**

q^2 = frequency of homozygous recessive **genotype**. **(aa)**

WHY USE THE HARDY-WEINBERG EQUILIBRIUM?

Alleles: $p + q = 1$
A a

Genotypes: $p^2 + 2pq + q^2 = 1$
AA Aa aa

These formulae allow scientists to determine whether evolution has occurred.

Changes in the gene frequencies over time indicates evolution.

No change in the gene frequencies indicates stability (no evolution).

FINDING THE HARDY-WEINBERG EQUILIBRIUM

	R	r
R		
r		

R = Tongue rolling

r = non-tongue rolling

	R (p)	r (q)
R (p)	RR (p^2)	Rr (pq)
r (q)	Rr (pq)	rr (q^2)

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

p = **dominant** allele (R)

q = **recessive** allele (r)

Let p → DOMINANT ALLELE frequency= 0.6

Let q → RECESSIVE ALLELE frequency= 0.4

R = Tongue rolling

r = non-tongue rolling

	R (p) **0.6**	r (q) **0.4**
R (p) **0.6**	RR (p^2) **0.36**	Rr (pq) **0.24**
r (q) **0.4**	Rr (pq) **0.24**	rr (q^2) **0.16**

	R (p) **0.6**	r (q) **0.4**
R (p) **0.6**	RR (p^2) **0.36**	Rr (pq) **0.24**
r (q) **0.4**	Rr (pq) **0.24**	rr (q^2) **0.16**

What do the numbers add up to?

0.36 + 0.24 + 0.24 + 0.16 = 1

If we express this in algebra form, we get…

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

Extra practice: Visit the following sites. Read the brief explanations and answer the questions. Answer keys are provided.
http://www.ksu.edu/parasitology/biology198/hardwein.html
http://www.mansfield.ohio-state.edu/~sabedon/biol1509.htm

RULE 2: THE KEY TO HARDY-WEINBERG PROBLEMS IS THE HOMOZYGOUS RECESSIVE ALLELS

The reason is we can literally COUNT THEM…

We can count homozygous recessive “rr” phenotypes because they only occur if the genotype is “rr”. But the phenotype of “Rr” and “RR” will look the same.

Remember the black sheep example…we know a sheep is homozygous recessive “ww” because it is black. A white sheep can be “Ww” or “WW” – we don’t know which one a sheep might be

If we know the frequency of the **dominant** allele (p) and the frequency of the **recessive** allele (q), then we can calculate the frequency of the genotypes **AA** (p^2), **Aa** ($2pq$) and **aa** (q^2) and vice versa.

Steps to Solving Hardy Weinberg Questions

Step 1: Write down your known and unknowns

Step 2: Follow these steps:

(Write this on the left side of your page.)

NOTE

To get q^2 we multiply q x q. or in reverse to get q we square root q^2

aa q^2 → Recessive trait or **genotype**

a $q = \sqrt{q^2}$ → Recessive **allele**

A $p = 1 - q$ → Dominant **allele**

AA $p^2 = p \times p$ → Dominant trait or **genotype**

Aa $2pq = 2 \times p \times q$ → Heterozygous trait or **genotype**

Example: Find all the Hardy Weinberg frequencies for the following.

7 out of 9 mice are brown. Brown is dominant to white.

Solution:

Since brown is the dominant trait, start by subtracting 7 from 9 to get the number of white mice, or the recessive individuals, which is **2**.

Divide so we get the "aa" frequency (Number of **recessive** individuals divided by total)

aa	q^2	$= 2/9 = 0.22222$
a	q	$= \sqrt{q^2} = \sqrt{0.2222} = 0.47140452$
A	p	$= 1 - q = 1 - 0.47140452 = 0.528595479$
AA	p^2	$= p \times p = 0.528595479 \times 0.528595479 = 0.27941318$
Aa	$2pq$	$= 2 \times p \times q = 2 \times 0.528595479 \times 0.47140452 = 0.498364596 = 0.50$

THEY ALL ADD UP TO 1

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

PROOF: .27941318 + .498364596 + .22222 = **pretty dang close to 1**

Hardy Weinberg

QUICK REFERENCE table

Allele / Genotype	Variable	How To Calculate Variable	Other Calculation Tips
aa	q^2	**# recessive indiv.** / **Total # indiv.**	To get q^2 from q multiply $q \times q$
a	q	$\sqrt{q^2}$	
A	p	$1 - q$	To get p from p^2 take the square root of p^2
AA	p^2	$p \times p$	
Aa	$2pq$	$2 \times p \times q$	

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

Example 1

In a population of flying pigs, some pigs have stubby wings. If there were 50 pigs in total and 8 have the recessive trait of stubby wings, how many are homozygous for the dominant allele?

Find: **AA** $= p^2$

Given: **aa** $= q^2 = \frac{8}{50} = 0.16$

a $q = \sqrt{q^2} = \sqrt{0.16} = 0.4$

A $p = 1 - q = 1 - 0.4 = 0.6$

FIND → **AA** $p^2 = 0.6^2 = 0.36$

Aa $2pq = 2 \times p \times q$

Don't need

Multiply the frequency of P^2 by the total number…its like figuring out the GST on a purchase

0.36 x 50 = 18 pigs

Example 2

If the frequency of the recessive allele for blue eyes in a population is 30% and there are 500 individuals in the population, how many blue-eyed people would there be?

Find: aa = q^2

FIND	aa	q^2	$= (.30)^2 = 0.09$
Given:	a	q	= .30 (converted from 30 percent)
	A	p	= don't need
	AA	p^2	= don't need
	Aa	2pq	= don't need

Like before, but this time use the q^2 frequency multiplied by the number of individuals to get how many are blue eyed in the whole population

0.09 x 500 = 45 blue-eyed people

Example 3

The dominant allele T controls the ability to taste PTC. Individuals with the T allele find PTC bitter, while tt individuals find it tasteless. In a sample of 320 students, 240 were tasters. Determine the number of heterozygous individuals in this population.

Find: Tt = 2pq

Given: **TT and Tt = $p^2 + 2pq$ = 240 taster students**

tt $q^2 = 320-240 = 80$ **(recessive people)**

$$= \frac{80}{320} = 0.25$$

t $q = \sqrt{q^2} = \sqrt{0.25} = 0.5$

T $p = 1-q = 1-0.5 = 0.5$

FIND **Tt** $2pq = 2(0.5)(0.5) = 0.5$

Again we were looking for how many individuals are "Tt" out of the total.

$0.5 \times 320 =$ **160 students are heterozygous**

Example 4

In cats, yellow eyes are controlled by a dominant allele and green eyes are the recessive trait. If 90 cats out of 250 have green eyes, how many cats have at least one recessive allele?

Find: $Aa = 2pq$ AND $aa = q^2$

Given:

$$q^2 = \frac{90}{250} = 0.36$$

$$q = \sqrt{q^2} = \sqrt{0.36} = 0.6$$

$$p = 1 - q = 1 - 0.6 = 0.4$$

$$2pq = 2\,(0.4)\,(0.6) = 0.48$$

FIND

$$2pq + q^2 = 0.48 + 0.36 = 0.84$$

$$0.84 \times 250 = \boxed{210 \text{ cats}}$$

Try this link for an online quiz for Hardy-Weinberg equilibrium

5 questions

http://www.phschool.com/science/biology_place/labbench/lab8/quiz.html?radio1=c&radio2=b&radio3=d&radio4=d&radio5=d&x=76&y=9

Another quiz

http://people.cst.cmich.edu/swans1bj/hwe/hwetutorial.html

Bozeman Hardy Weinberg Equation 9:23

Why do we always start with q^2?

http://www.youtube.com/watch?v=oEBNom3K9cQ

Bozeman:

Solving Hardy Weinberg Problems 11:07

http://www.youtube.com/watch?v=xPkOAnK20kw

Example 1:

1. A population has only two alleles, R and r, for a particular gene. The allele frequency of R is 20%. What are the frequencies (use a whole number percentage) of the homozygous dominant and recessive genotypes, as well as the heterozygous genotype?

R = 20%(dominant)

Solution:

FIND → q^2 = homozygous recessive = $(0.80)^2$ = 0.64

rr — q = recessive allele = 1- 0.2 = 0.80

R — p = dominant allele = 20% = .20

R / RR — FIND → p^2 = homozygous dominant = $(0.20)^2$ = 0.04

Rr — FIND → 2qp = heterozygous genotype = 2(0.80)(0.20) = 0.32

Check your Answers!

$p^2+2pq+q^2$ = 1 → .04 + .32 + .64 = 1

- **Example 2:** Cystic Fibrosis is a recessive condition that affects about 1 in 2500 people in the Caucasian population of Canada. Calculate the following:

a) The population frequencies for the dominant (C) and recessive (c) alleles

b) The percentage of the population that is a carrier of the recessive allele

c) The number of students at a school (2400 students) that are likely to be carriers of the cystic fibrosis allele

C = dominant
c= recessive
recessive condition = cc
recessive condition = 1/2500
Carriers = Aa

Solution:

– 1/2500 = 0.0004 = q^2

cc q^2 = homozygous recessive = 0.0004

c q = recessive allele = √0.0004 = 0.0200

C p = dominant allele = 1 - 0200 = 0.98

CC p^2 = homozygous dominant = .98 x .98 = .9604

Cc 2pq = heterozygous genotype = 2(0.0200)(0.98) = 0.0392 = **4.0%**

c) Number of students = ~ 2400 X 0.004 = ~ **94.08 = 94 students**

Additional Example H-W Problems. Example #3

5. In a randomly mating population of snakes, one out of 100 snakes counted as albino, a recessive trait. Determine the theoretical percentage of each of the genotypes in the population.

Plan your attack!

- what is required?
- What is given?
- Plan your strategy.
- Act on your strategy.
- Check your solution.

1 / 100 = albino(recessive)
"aa"

aa	q^2 =	1/100 = .01	= 1%
a	q =	$\sqrt{.01}$ = .1	
A	p =	1 - .1 = .9	
AA	p^2 =	.9 x .9 = .81	= 81%
Aa	$2pq$ =	2 x .9 x .1 = .18	=18%

CHECK: .18 + .81 + .01 = 1

Example #4

A single pair of alleles codes for one of the genes that controls wing length in fruit flies. The long wing allele (L) is dominant to the short wing allele (l). If 40 fruit flies out of 1000 are counted to have short wings, how many fruit flies out of 1000 would be expected to be heterozygous?

L = long wing(dominant)
l = short wing(recessive)
40 / 1000 = "ll"

Has to be "ll" because if there was a "L" then the recessive trait would not show

ll q^2 = **40/1000 = .04**

l q =

L p = **1 - .2 = .8**

LL p^2 = **.8 x .8 = .64** **√.04 = .2**

Ll $2pq$ = **2 x .8 x .2 = .32** → Change to # of individuals → 1000 x .32 = <u>320</u>

FIND

CHECK: .04 + .64 + .32 = 1

Check for Understanding:

1. What are the conditions of the Hardy-Weinberg principle?
2. Using the Hardy-Weinberg equation, distinguish between p and p^2 as used to describe a population.
3. How is it possible to find the number of heterozygotes in a sample population, given the allele frequencies?
4. Fill in the following chart:

p^2	+	$2pq$	+	q^2	=	1
Frequency of:		Frequency of:		Frequency of:		All of the individuals in the population (100%)

Use the following information to answer the next two questions.

A high percentage of purebred dogs have genetic defects. Some examples of these defects follow.

1 Hip dysplasia, a defect in the hip joints that can cripple a dog, occurs in 60% of golden retrievers.

2 Hereditary deafness, due to a recessive autosomal disorder, occurs in 30% of Dalmatians.

Recessive trait = aa = q^2

3 Retinal disease, which may cause blindness, occurs in 70% of collies.

4 Hemophilia, an X-linked recessive disorder, is common in Labrador retrievers. Dwarfism is also common in this breed of dog.

What is the frequency of the abnormal allele that causes hearing defects in Dalmatians?

Answer: .55

$q^2 = .3$ (was 30%)

$q = \sqrt{.3} = .55$

$p = .45$

$p^2 = .2$

$2pq = .5$

Check $.3 + .2 + .5 = 1$

125. The breeding of purebred dogs for certain characteristics related to appearance is blamed for the disturbing number of genetic defects in these animals. These defects are **most likely** the result of

- **A.** natural selection
- **B.** non-random mating
- **C.** geographic isolation
- **D.** high rates of mutation

Ocular albinism is a recessive genetic disorder. The frequency of ocular albinism in Northern Ireland is 1 in 10 000.

The Hardy–Weinberg expression that represents the frequency of the allele for ocular albinism in Northern Ireland is

- **A.** $p^2 = 0.01$
- **B.** $q^2 = 0.01$
- **C.** $p = 0.01$
- **D.** $q = 0.01$

aa = q^2 = 1/10 000 = .0001

a = q = √.0001 = .01

Use the following information to answer the next question.

> A community of Pima Indians in the American Southwest has a very high rate of diabetes in their adult population. Of the population of adults over the age of 35, 42% to 66% develop diabetes. The recessive trait that causes diabetes in this population is a distinct disadvantage to individuals whose diets are rich in carbohydrates.
>
> —from *Cummings, 1993*

Numerical Response

4. If 42% of the population have diabetes, then the percentage of the population who are carriers is calculated to be ___46___%.

(Record your answer **as a whole number** in the numerical-response section on the answer sheet.)

Diabetes = recessive trait = aa
42 % of population

Carriers are Aa

q^2 = .42 (was 42%)

q= √.42 = .65

p= 1 - .65 = .35

p^2 = .35 x .35 = .1225

2pq= 2 x .65 x .35 = .455 (46%)

Check .42 + .1225 + .455 = 1

Flower Phenotype Distribution of 87 Snapdragon Plants in a Garden

Red flowers	43
Pink flowers	36
White flowers	8

RED = dominant = RR = p^2
WHITE = dominant = WW = p2
PINK would be RW = 2pq

Note: Flower colour in snapdragon plants is a single gene trait controlled by two alleles: the red flower colour allele and the white flower colour allele.

Which of the following rows identifies the frequency of the two alleles in this garden?

Row	Red Allele	White Allele
A.	0.91	0.09
B.	0.70	0.30
C.	0.49	0.51
D.	0.40	0.60

	RED	WHITE
q^2 =		
q=		
p=	√ .494=.70	= √ .092 =.30
p^2	= 43/87=.494	=8/87 = .092
2pq=		

Hereditary hemochromatosis (HHC) is an autosomal recessive disorder characterized by elevated levels of iron in the blood. HHC is one of the most common genetic disorders. In the Caucasian population in the United States, approximately 5 in 1 000 people carry two copies of the mutated hemochromatosis allele.

—based on *DOEgenomes.org*, 2002

2 recessive copies = aa

The frequency of the dominant allele associated with HHC in the Caucasian population of the United States is

A. 0.005
B. 0.071
C. 0.864
D. 0.929

Recessive trait = 5/1000 = aa = q^2

$q^2 = 5/1000 = .005$

$q = \sqrt{.005} = .071$

$p = 1 - .071 = .929$

$p^2 = .929 \times .929 = .863$

$2pq = 2 \times .929 \times .071 = .131919$

Check $.863 + .1319 + .005 = 1$

In people of Northern European descent, the frequency of the mutated hemochromatosis allele is 0.05.

—based on Feder et al., 1996

The frequency of carriers of HHC in people of Northern European descent is

A. 0.003
B. 0.050
C. 0.095
D. 0.950

Carriers = Aa = 2pq
Allele = .05 = a = q

q^2 = .05 x .05 = **.0025**

q= .05

p= 1 - .05 **= .95**

p^2 = .95 x .95 **= .9025**

2pq= 2 x .95 x .05 = **.095**

Check .9025 + .095 + .0025 = 1