Exam on Friday 2pm

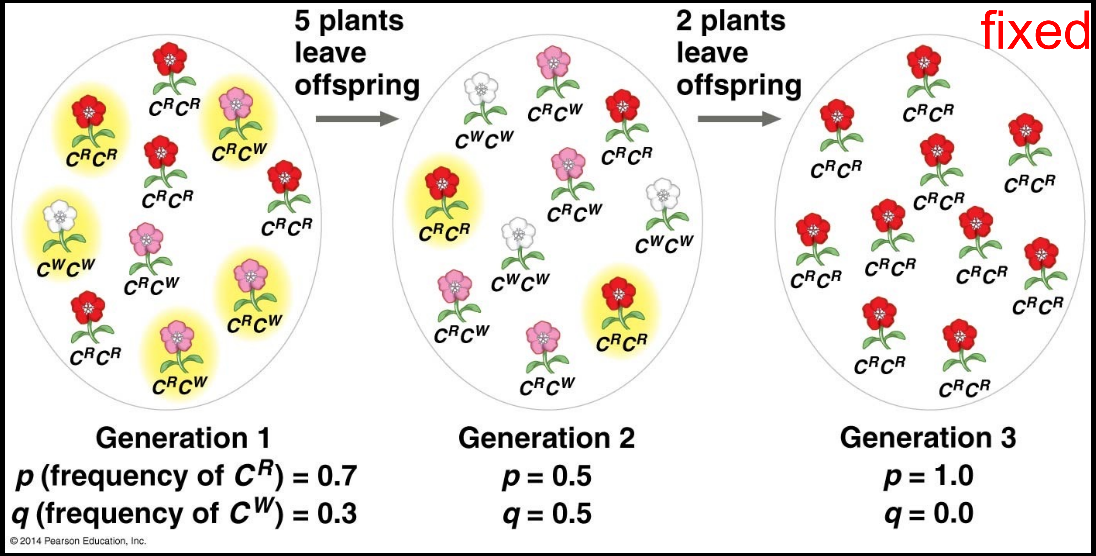
- Bring a pencil and a pen (black or blue)
- student ID (just in case)
- See announcement

Intro to population genetics

- How do alleles and genotypes behave in whole populations?

Gene pool

- A concept denoting the entire genetic makeup of a population i.e. all the alleles present in an interbreeding population
- If only one allele is present, this is allele said to be fixed (this can happen by natural selection, or genetic drift)



Intro to population genetics

- Genotype frequency: the frequency of individuals with a specific genotype (e.g. homozygous for white C^wC^w)
- Allele frequency: the number of copies of one specific allele divided by number of copies of all alleles in that population (for that gene, remember two copies for each individual)
- Allele frequencies in a population; typically denoted by lowercase letters (e.g. alleles C^R, C^w could have allele frequencies p, q)
- If there are only two alleles present, then p+q = 1 (100%)
- If there is only one allele for a given locus, then it is said to be fixed (e.g. if all individuals are white C^wC^w)

Intro to population genetics

- E.g. a population of 100 plants has two alleles for flower color, C^w and C^R. 10 individuals are homozygous for white, 20 are heterozygous C^wC^R, and 70 are homozygous red.

- Number of white alleles: (10x2) + (20x1) = 40. Allele frequency: <u>q= 40 / 2x100 = 0.2</u>

- Red allele frequency: <u>p= 1- 0.2 = 0.8</u>

Intro to the Hardy Weinberg principle

- Population = a localized group of interbreeding individuals of the same species
- Hardy and Weinberg studied the allele frequencies in stable populations

Hardy Weinberg principle

- Given a series of assumptions, allele frequencies and genotype frequencies in a population will not change between generations, and <u>genotype frequencies follow a set pattern</u>

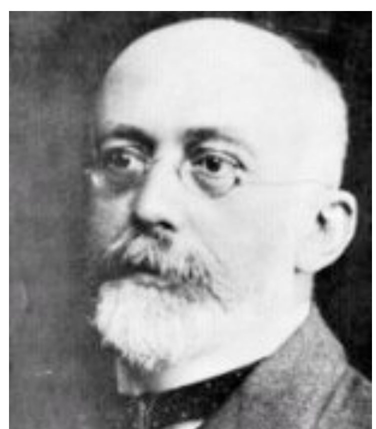
$$\boxed{p^2 + 2pq + q^2 = 1}$$

Hardy-Weinberg Equilibrium

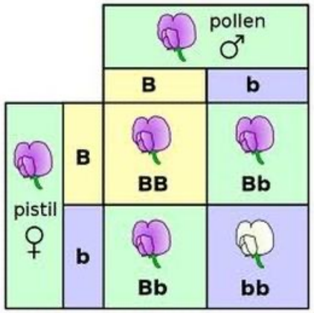
If the following 5 assumptions/conditions are met, the frequency of each allele will remain exactly the same from generation to generation (i.e. no evolution):

1. No mutations
2. No gene flow
3. No natural selection
4. Random mating
5. Large population

$$\boxed{p^2 + 2pq + q^2 = 1}$$



THE HARDY-WEINBERG EQUATION



Punnett Square assuming complete dominance

$$p + q = 1$$

p = frequency of the dominant allele in the population

q = frequency of the recessive allele in the population

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

p^2 = freq. of homozygous dominant individuals (genotypes)

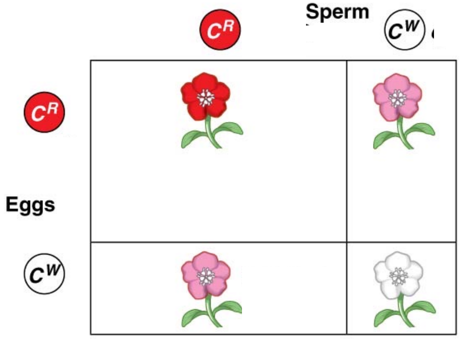
q^2 = freq. of homozygous recessive individuals (genotypes)

$2pq$ = freq. of heterozygous individuals (genotypes)

Deriving the H-W equation:

Use the Punnett square:

A species of flower occurs in red, pink and white varieties in nature, the alleles for color are all on one locus: what kind of dominance do you think this is?

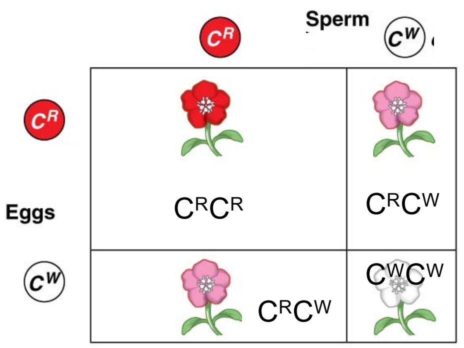


Let's study this example:

A species of flower occurs in red, pink and white varieties in nature:

-Incomplete dominance

1. C^R= allele for red
2. C^W = allele for white
3. C^RC^W genotype will produce pink flowers, C^RC^R red, C^WC^W white.



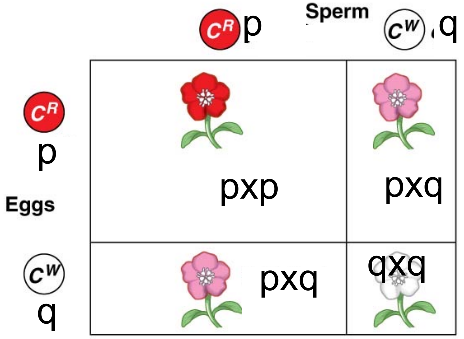
-Note: this is a stable, relatively unchanging population in nature, NOT based on experiments with true breeding etc.

Deriving the H-W equation:

Allele frequencies:

Frequency of C^R=p

Frequency of C^W=q



Deriving the H-W equation:

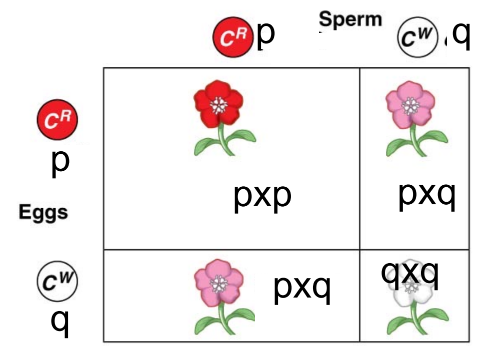
Assume random mating.

Genotype frequencies, based on Punnett square:

Frequency of homozygous dominant genotype = **p^2**

Frequency of homozygous recessive genotype = **q^2**

Frequency of heterozygous genotype = **pxq+pxq= 2pq**



Deriving the H-W equation:

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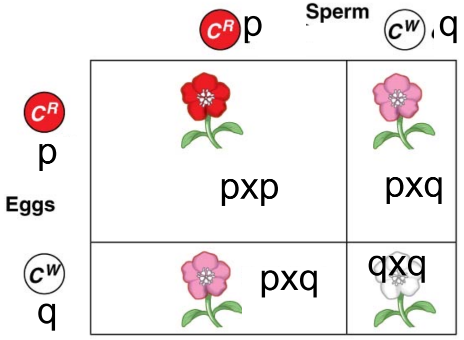
Genotype frequencies, based on Punnett square:



Frequency of homozygous dominant genotype = **p^2**

Frequency of homozygous recessive genotype = **q^2**

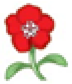
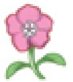
Frequency of heterozygous genotype = **pxq+pxq= 2pq**

These are all the available genotypes, so they should add up to 1 (=100%)

$$\mathbf{p^2+2pq+q^2 = 1}$$

Let's study this example further:

In this case we have 500 individual plants (1000 alleles, remember that each individual has two alleles, when counting allele frequencies):

1. 320 individual flowers are red 
2. 160 are pink 
3. 20 are white 
4. Number of red alleles = (320x2)+(1x160)= 800
5. Frequency of C^R = p = 800/1000 = 0.8
6. Number of white alleles = 1000-800 =200
7. Frequency of C^W = q = 200/1000 = 0.2

Let's study this example:

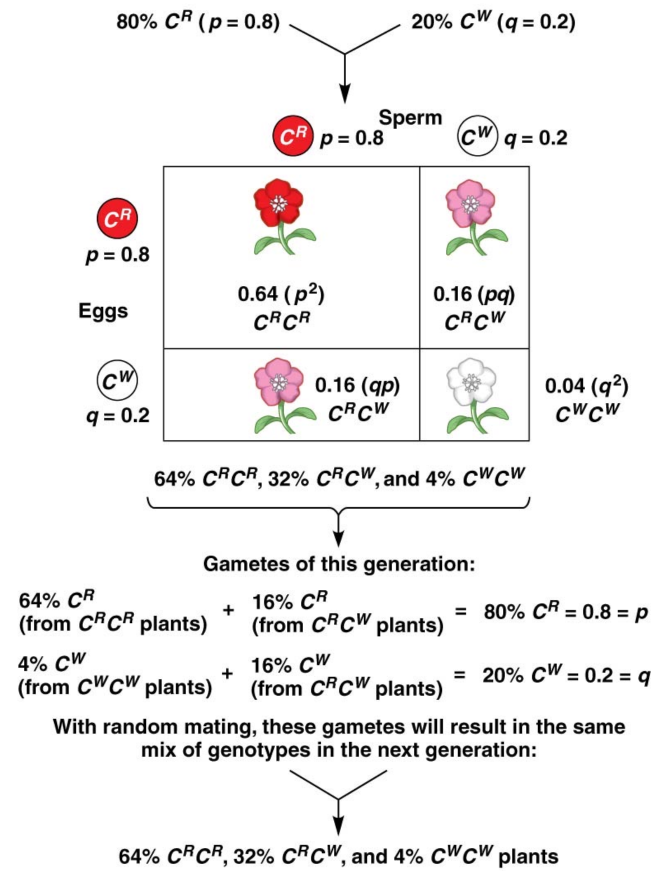
Frequency of genotypes:

1. 320 are red, frequency 320/500= 0.64 (same as H-W p^2)
2. 20 are white, frequency 20/500= 0.04 (same as H-W q^2)
3. 160 are pink = 160/500=0.32 (same as H-W 2pq)

All genotype frequencies should add up to one:

$0.64 + 0.32 + 0.04 = p^2 + 2pq + q^2 = 1$

Conclusion: The population is in Hardy Weinberg equilibrium



Don't forget the easy solutions

$p+q=1$

$q=\sqrt{q^2}$ =sq.root of frequency of homozygous recessive genotype.

E.g. eye color:

B = brown (dom.)

b= blue (rec.)

You are told that the frequency (q) of blue eyed (bb) people is 0.25. What is the frequency of the recessive allele (assuming H-W)?

$$\boxed{p^2 + 2pq + q^2 = 1}$$

A simple example….

- Imagine a population made up of 100 individuals, assume H-W equilibrium: A is dominant, a is recessive.
- 4 individuals show the recessive trait
- The allele frequency of A is?
- The allele frequency of a is?

A simple example….

- Imagine a population made up of 100 individuals, assume H-W equilibrium: A is dominant, a is recessive.
- 4 individuals show the recessive trait
- The allele frequency of A is? 0.8
- The allele frequency of a is? 0.2

So what?

- H-W forms the basis of microevolutionary studies
- It serves as a null model: if a population does not conform to the equilibrium equation, we can look at which assumptions are violated (i.e., which evolutionary forces are acting)
- The Hardy-Weinberg Principle can be used to test whether a population is evolving