Lecture 10:

The basic model

Course 410

Molecular Evolution

“ The answer was in Mendel’s paper all the time”[1]

Hardy-Weinberg Principle

The first and most basic model in population genetics

(1) Edwards, 2008

Hardy-Weinberg Law

Weinberg

- Wilhelm Weinberg
- (1862-1937)
- German Physician

Hardy

- Godfrey H. Hardy
- (1877-1947)
- English mathematician

How did a mathematician, Hardy, get involved?

- February 28th 1908, Punnett gave a lecture on “Mendelism in relation to disease” [1].
- The lecture discusses brachydactylism.
- Brachydactylism means short-fingeredness [2].
- Shortness in fingers and toes relative to other body parts.
- The genetics disease is a dominant trait.

(1) Edwards, 2008 (2) Jewett,1914

- **Yule:** if brachydactyl is a dominant trait (assuming random mating) ➔ 3:1 brachydactyl : normal.
- Yule misinterpreted Mendel's theory. In order to get the 3:1 ratio, the gene frequency must be ½.

- **Punnett**: interpreted Yules remarks as “why the nation was not becoming ….. Brachydactylous”.
- Punnett was puzzled “why the dominant did not continually increase in frequency?”
- Hardy ….. Help!

Mendelian Proportions in a Mixed Population

• **Assumptions**: Aa is a Mendelian characters. The numbers of genotypes pure dominant (AA), heterozygotes (Aa), and pure recessives (aa) are 1:2:1 respectively.

• **Conditions**: " … suppose that the numbers are fairly large, so that mating may be regarded as random, that the sexes are evenly distributed among the three varieties, and that all are equally fertile."[1]

• Using "a little mathematics …" the allele frequencies in the next generation will be " .. unchanged after the second generation."

• " I have … considered only the very simplest hypotheses possible." [1]

(1) Hardy, 1908

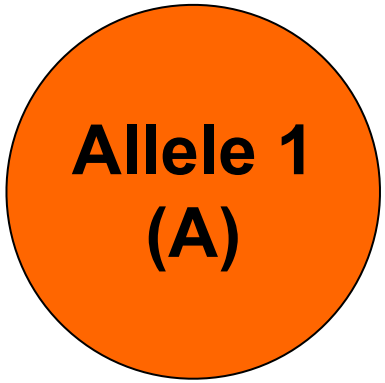
Assumptions

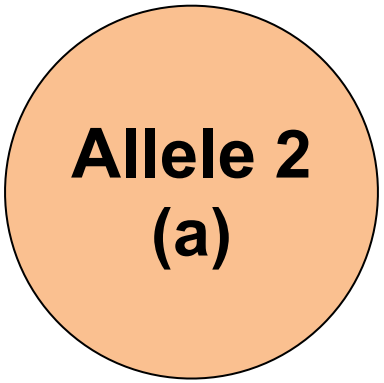
- Single locus / Biallelic locus
- Diploid organism / Equal sexes.
- **No natural selection:** equal survival rates and reproductive success
- **No mutation:** no alleles created or converted
- **No migration/gene flow:** individuals do not move into or out of the population

- No genetic drift.
- **Population is infinitely large:** sampling errors and random effects insignificant.
- No population subdivision.
- Random mating (no inbreeding).
- Non overlapping generations.

The idea

Single locus and two alleles





p = frequency of Allele 1

$$\mathbf{p} = \frac{\text{\# of allele 1}}{\text{\# allele 1+allele 2}}$$

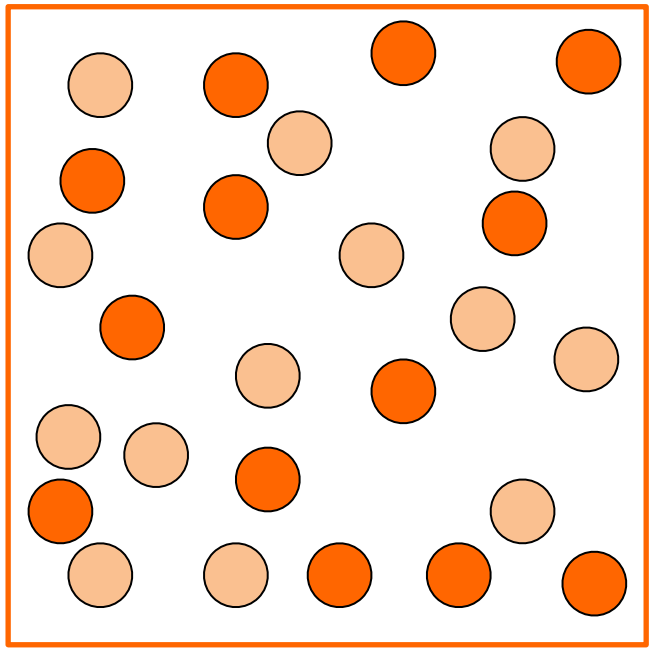
q = frequency of Allele 2

$$\mathbf{q} = \frac{\text{\# of allele 2}}{\text{\# allele 1+allele 2}}$$

What is the frequency of all alleles in a population?

$$p + q = \frac{\#\text{ of allele 1}}{\#\text{ allele 1+allele 2}} + \frac{\#\text{ of allele 2}}{\#\text{ allele 1+allele 2}} = \frac{\#\text{ allele 1+allele 2}}{\#\text{ allele 1+allele 2}}$$

Hardy-Weinberg Principle



(1) $p + q = 1$

	A (p)	a (q)
A (p)	AA (p^2)	Aa (pq)
a (q)	Aa (pq)	aa (q^2)

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

Predictions

(1) Allele frequency **do not** change over time

(2) Genotype frequencies can be calculated

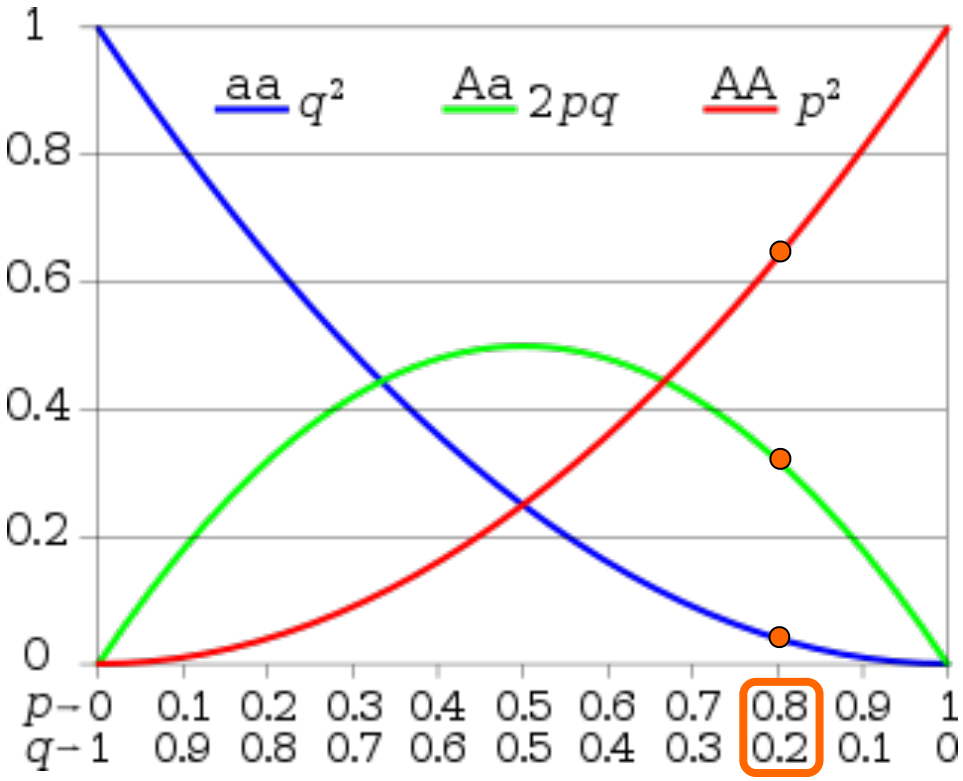
Relationship between genotype and allele frequency [1]

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

p2 = $(0.8)^2$ = 0.64

2pq = 2*(0.8)*(0.2) = 0.32

q2 = $(0.2)^2$ = 0.04



Population is at HW equilibrium at this locus

(1) Klug et al., Concepts of Genetics

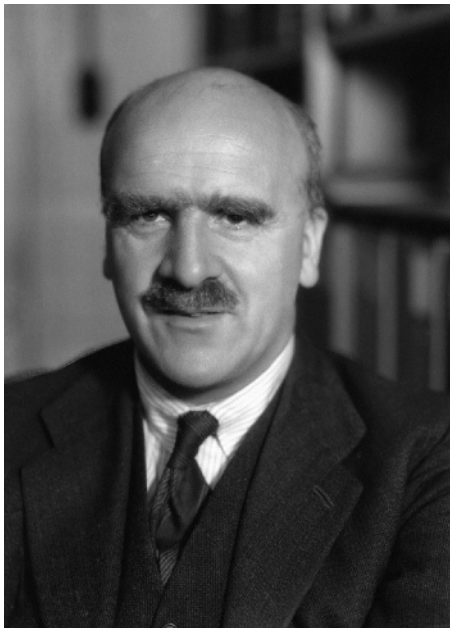
What happens in frequencies <u>ARE NOT</u> in HW equilibrium?

We test which assumption of HW was violated and may, as a result, explain the evolution of the population at this particular locus

Let’s do beanbag genetics

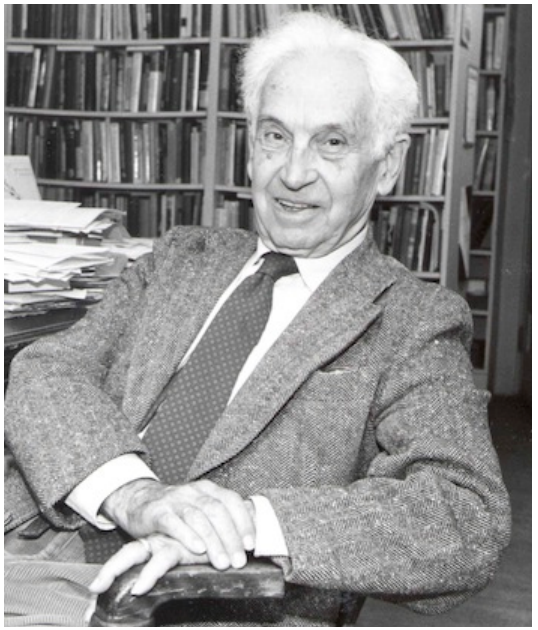
A Defense of Beanbag Genetics*

JBS Haldane



My friend Professor Ernst Mayr, of Harvard University, in his recent book *Animal Species and Evolution*[1], which I find admirable, though I disagree with quite a lot of it, has the following sentences on page 263.

> The Mendelian was apt to compare the genetic contents of a population to a bag full of colored beans. Mutation was the exchange of one kind of bean for another. This conceptualization has been referred to as ''beanbag genetics''. Work in population and developmental genetics has shown, however, that the thinking of beanbag genetics is in many ways quite misleading. To consider genes as independent units is meaningless from the physiological as well as the evolutionary viewpoint.



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