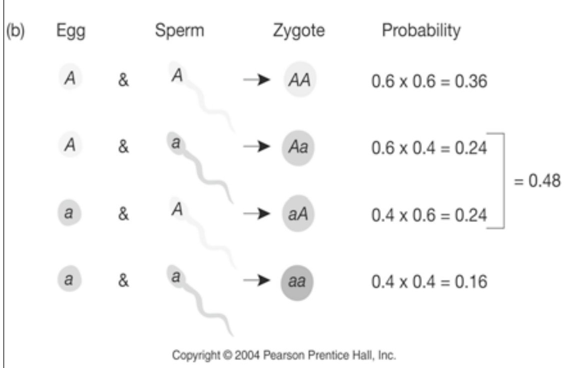
Population Genetics

- Integrates Darwin's evolution with Mendelian genetics
- Changes in traits in a population are related to changes in allele frequencies
- Evolution is change in allele frequencies across generations

Probabilities

- The combined probability that 2 independent events (a particular egg meets a particular sperm) is equal to the product of their individual probabilities
- So let's say our population has 60% *A* And 40% *a* alleles. The probabilities of getting particular genotypes in the offspring are as follows……



Hardy-Weinberg Equilibrium

1. The allele frequencies in a population will not change generation after generation: $p + q = 1$
2. If the allele frequencies are p and q, the genotypic frequencies are
   1. P^2 = AA
   2. 2pq = Aa
   3. Q^2 = aa

Assumptions

1. There is no selection: all members of the model population survive at equal rates and contribute equal numbers to the gene pool
2. There is no mutation: no new alleles are created
3. There is no migration: no one moves into or out of the population

More Assumptions

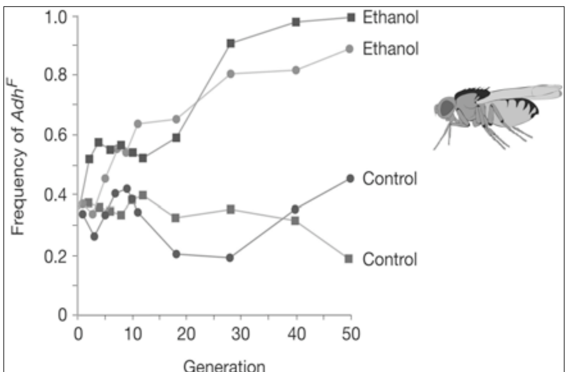
- 4. The population is infinitely large: there can be no genetic drift by chance
- 5. Individuals choose their mates at random

So what?

- By providing a list of conditions under which evolution won't occur, the Hardy-Weinberg model identifies the forces that CAUSE evolution in the real world
- It serves as a null hypothesis
  - We'll look at a few examples of how this is useful

Assumption 1: Selection

- If selection is occurring, some phenotypes are more fit than others and have more offspring
- If we assume that phenotypes are directly controlled by genotypes, these fitter individuals contribute more of their alleles to future generations.
- H-W should not hold if there is selection



Another example…

- The Fore people of Papua New Guinea went through a long epidemic of kuru, a fatal neurological disorder
- It's a spongiform encephalopathy, or spongy brain disease, like Mad Cow
  - Do you remember how did the cows got mad cow in the 1980's?

The Test

- At the height of the epidemic, it was killing ~1% of the Fore annually
- The PrP gene on chromosome 20 influences a person's susceptibility to Creutzfeldt-Jakob disease, sort of the human version of Mad Cow
  - One allele produces the amino acid methionine, the other, valine
  - So you can be met/met, met/val, or val/val

The Outcome

- The gene frequencies in the surviving populations are:
  - Met/met: 0.13
  - Met/val: 0.77
  - Val/val: 0.10

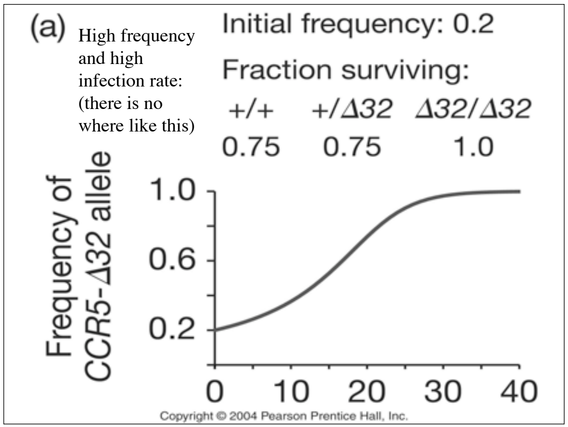
Survival was not random! Heterozygotes resist kuru and were selected for

So…..

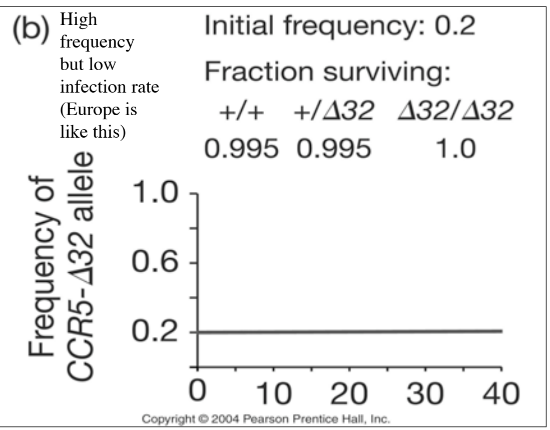
Back to the AIDS question

- Can we use H-W to figure out whether the AIDS epidemic will increase the frequency of CCR5-d32 in the human population?
- Scientists have used the H-W model to calculate allele frequency over generations using 3 different scenarios

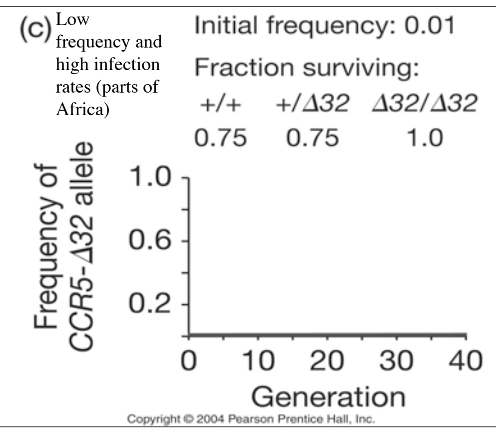
High frequency and high infection rate: (there is no where like this)



High frequency but low infection rate (Europe is like this)

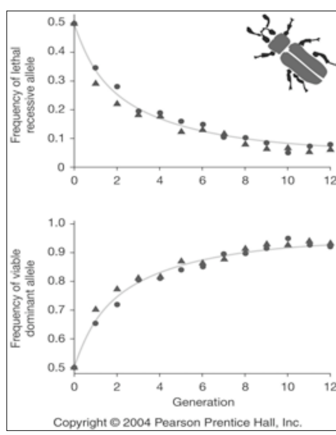


Low frequency and high infection rates (parts of Africa)



Testing the H-W Model

- Flour Beetles: Dawson identified the L-gene which is either + or l; l-homozygotes die
- Dawson predicted that based on the H-W model, over generations, the l-allele frequency should decrease
- He took 2 populations of heterozygotes and let them reproduce for 12 generations



Rate of Evolution
In Flour Beetles

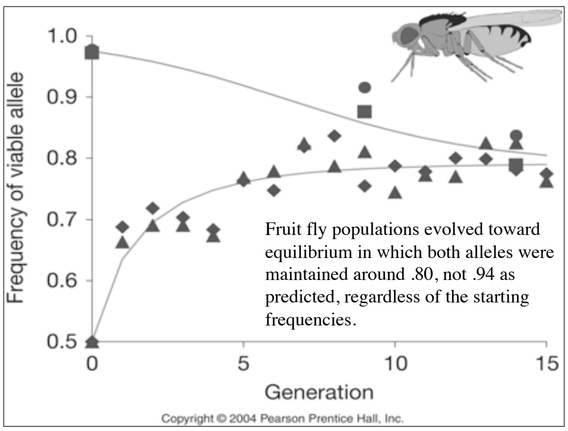
(gray lines are the model)

result

- Dominance and allele frequency interact to determine the rate of evolution
  - If the recessive allele is common, evolution by natural selection is rapid (the flour beetles)
  - If the dominant allele is common, evolution by natural selection is slow (CCR5-Δ32)

Heterozygote Superiority

- At a particular locus in fruit flies, V is viable and L is lethal if homozygous
- Same experiment as with the beetles
- It was predicted that the same thing would happen if you started with a population of heterozygotes
  - V allele should reach a frequency of 94% by 15 generations



What happened?

- **Heterozygote superiority**
- The heterozygotes have higher fitness than either homozygote
- At some point, the lethality of the lethal homozygote exactly balances its advantage as a heterozygote and equilibrium is achieved
- This could maintain genetic diversity

Frequency-Dependent Selection

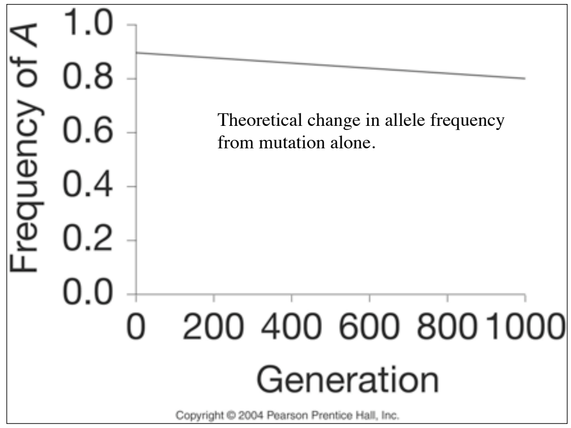
- Elderflower orchids can be yellow or purple and yellow is a little more common
- The colors attract bees for pollination, but the bees don't get any nectar from these flowers
- Bees alternate flower colors for awhile and then give up

Why isn't one color selected against until it disappears?

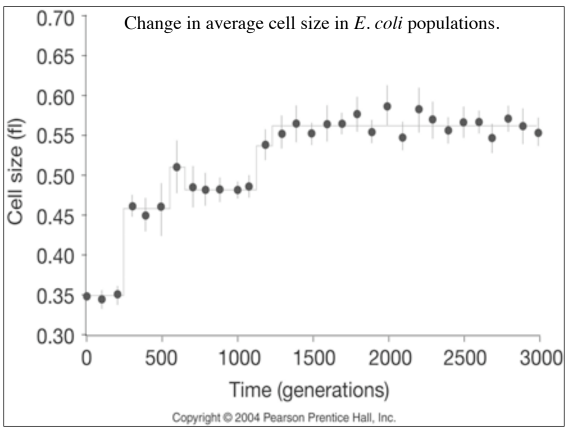
- If bees are alternating flowers, the more rare flower gets visited more
- If more visits = better pollination = more reproductive success, the rarer color will be selected for as long as it is rare
- Selection favors yellow until it becomes too common and then it favors purple
- **Frequency-dependent selection**

Mutation and H-W

- In the H-W model there is no mutation
- How effective is mutation at changing allele frequencies?
- If we assume a mutation rate of 1 in 1000 (A to a)---quite a high rate…..



Theoretical change in allele frequency from mutation alone.



Change in average cell size in *E. coli* populations.

Mutation-Selection Balance

- Most mutations are deleterious and selection removes them from populations but they are continuously occurring
- If the mutation rate balances the rate at which they are being eliminated by selection, this is **mutation-selection balance**
  - Something else that maintains genetic diversity