

Roadmap

- Quantitative traits
 - What kinds of variation can selection work on?
 - How much will a population respond to selection?
 - Heritability
 - How can response be restored?
- Inbreeding
 - How inbred is a population?
 - What are the consequences of inbreeding?

Genetic variability and selection

- Genetic diversity is raw material for selection
- Directional selection reduces diversity (by getting rid of bad alleles)
- If diversity runs out, selection stops working



Genetic linkage blocking selection

- If all we have is:

A b a B

all efforts to get more capital letters than small letters must fail

- If we use up the loci that have no linkage or good linkage, we could be stuck with loci like A and B
- Genetic diversity is present but selection can't use it

Variance

- Variance is calculated by:
 - Squaring the difference between each observation and the mean
 - Averaging the squared differences
- Variance measures the spread of observations around their mean
- Using variance rather than standard deviation gives useful statistical properties
 - Variability from multiple sources—you can add variances together

Partitioning the variability of a character

$$V_T = V_G + V_E$$

- V_T total variance in phenotype (how much do organisms vary?)
- V_G variance due to genes
- V_E variance due to environment

Partitioning in more detail

$$V_T = V_A + V_D + V_E + V_{GE}$$

- V_T total variance in phenotype (how much do organisms vary?)
- V_A additive genetic variance
- V_D dominance-related genetic variance
- V_E environmental variance
- V_{GE} correlation between genes and environment

Examples (for a locus named B)

- V_A – having a B causes a predictable increase
 - Each B allele adds 2 lbs to weight compared to b
- V_D – no predictable effect of B
 - Bb weighs 2 lbs more than either BB or bb
 - Males weigh 2 lbs more than females
- Both V_A and V_D – on average B causes an increase but amount varies
 - BB and Bb weigh 2 lbs more than bb

Why care about V_A ?

- It determines the response to selection
- Population where each B adds 2 lbs (all V_A):
 - Selection will increase weight until all b are gone
- Population where Bb is biggest (all V_D at equilibrium):
 - Selection will stop working leaving a population with varied sizes
- Population where BB and Bb are the same (some of both):
 - Selection will work but is less efficient
 - As b becomes rare it will hide in the heterozygote

An example of V_{GE}

- In a lowland lab, alpine plants grow shorter than lowland plants
- In a highland lab, alpine plants grow TALLER than lowland plants
- Alpine genotypes:
 - are found in the highlands
 - only make the plant taller in a highland environment
- This is a genetics x environment interaction
- (I'm mostly going to ignore these—they are difficult to deal with!)

A thought experiment

What if:

- We traveled around and collected the tallest plants we could find for breeding
- Some were highland, some were lowland
- Would we see a predictable response to selection in the next generation?
- Would it matter where our own lab was?

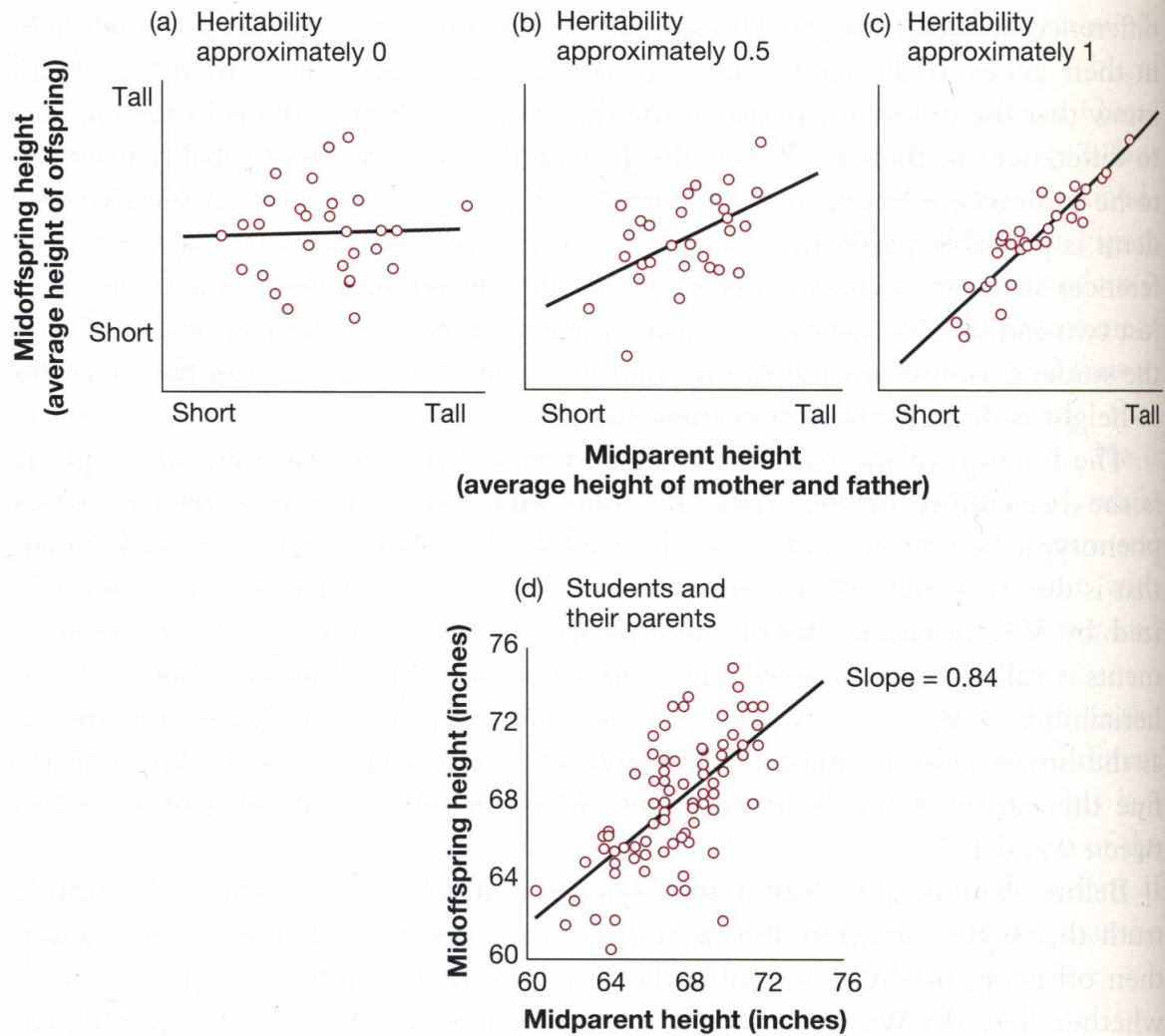
Heritability

$$\text{Heritability} = h^2 = \frac{V_A}{V_T}$$

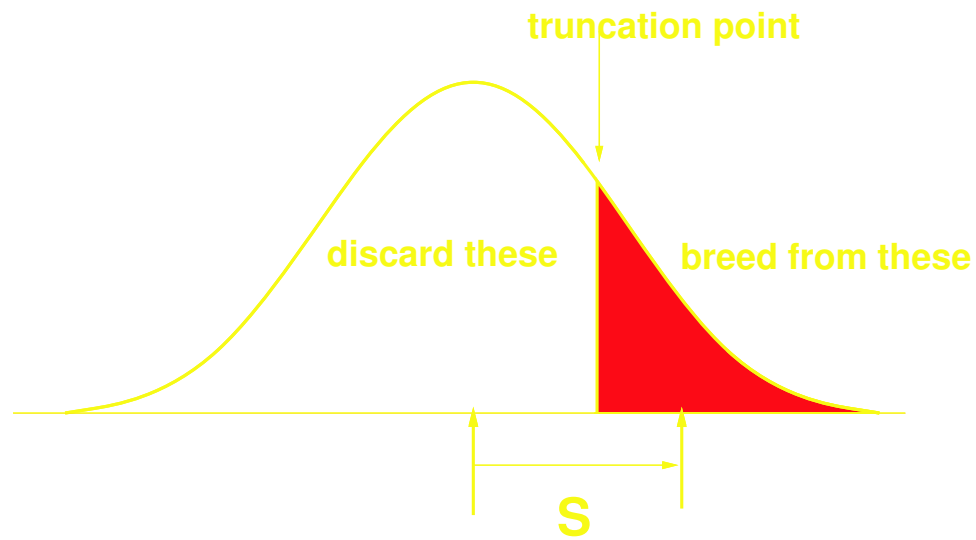
- Heritability:
 - Ratio between additive genetic variance and total variance
 - Measures the potential of the population to respond to selection
- *Only meaningful for a specific population and environment*

Figure 6.12 Scatterplots showing offspring height as a function of parent height

Each of the top three scatterplots shows data for a hypothetical population, and each includes a best-fit line through the data. (a) In this population, offspring do not resemble their parents. (b) In this population, offspring bear a moderate resemblance to their parents. (c) In this population, offspring strongly resemble their parents. (d) This graph shows data for an actual population of students in a recent evolution course at a university in the Pacific Northwest, USA.



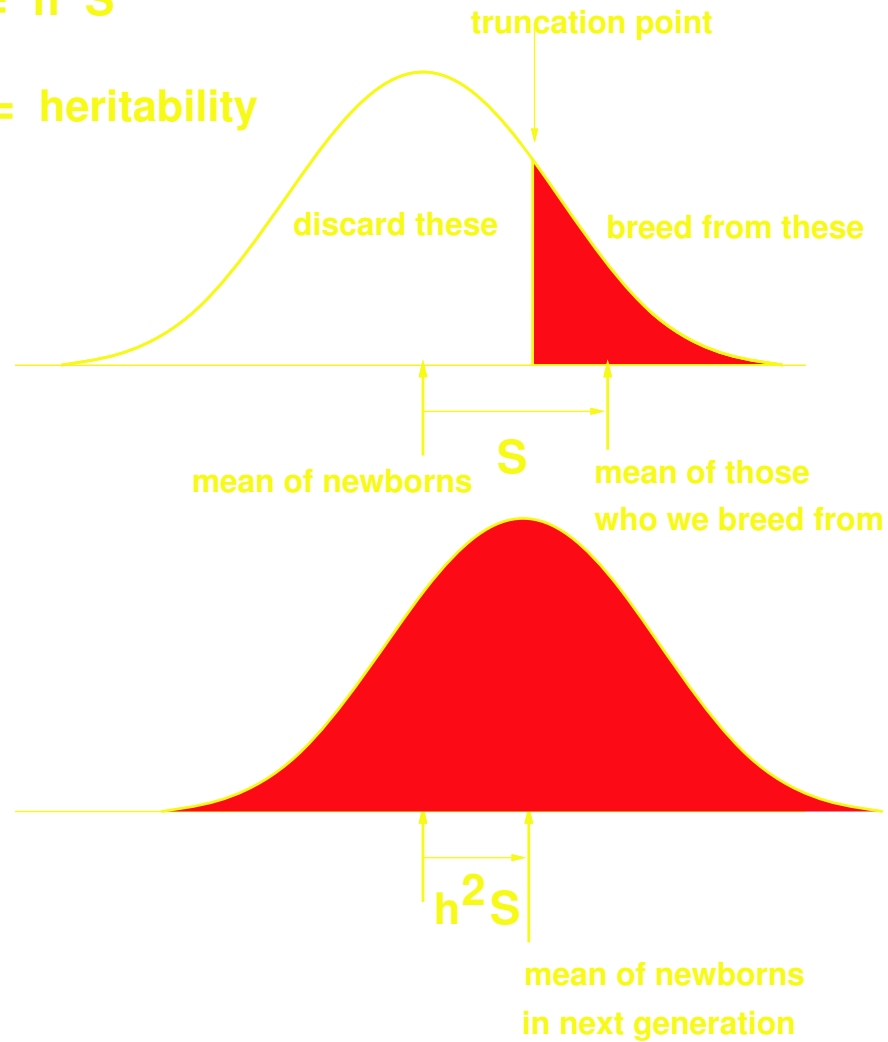
Selection differential S



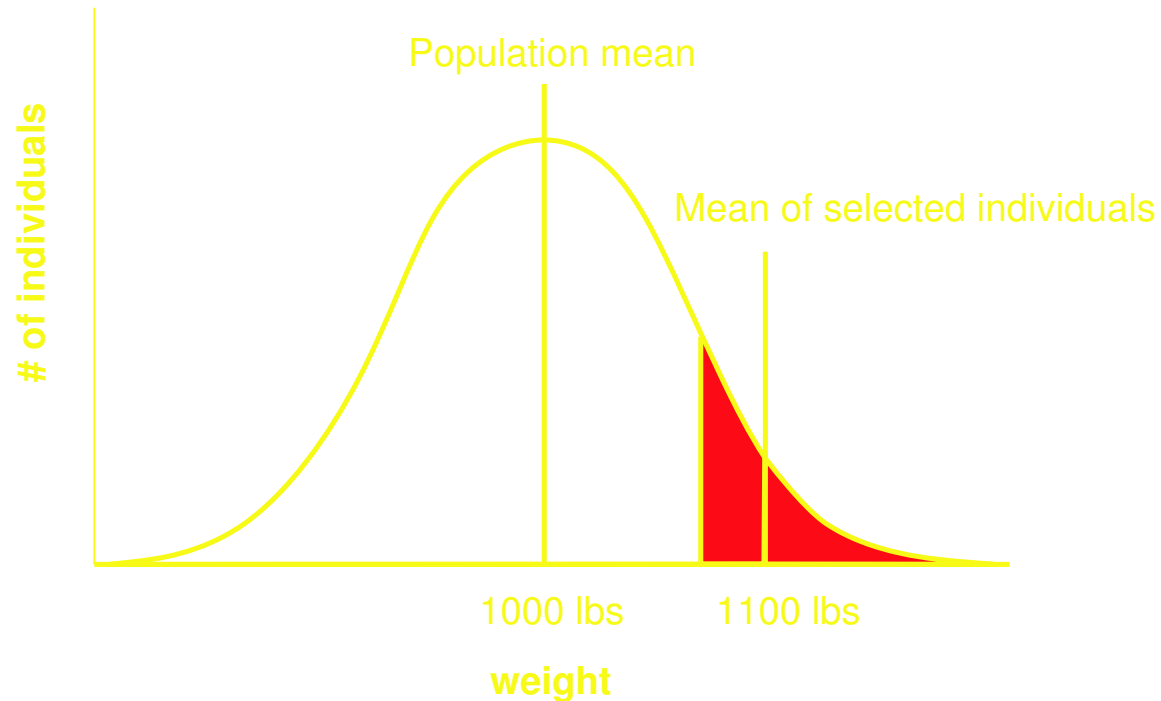
Response to artificial selection

$$R = h^2 S$$

h^2 = heritability



Possible gain per generation



If heritability = 0.4

S = selection differential

= mean of selected individuals - population mean = 100 lbs

R = gain

= $h^2 S$ = 40 lbs

This is the expected gain in one generation

Practice problem

- Sample wild corn with a mean of 100 kernels/ear and considerable variation
- Breed from ears with a mean of 150 kernels/ear
- Heritability of kernels/ear is 75%
- How many kernels/ear do I expect in 1 generation?

Practice problem

- We gain 37.5 kernels in 1 generation
- In 100 generations will we have corn with 3850 kernels/ear?
- Is there something wrong with our reasoning?

Heritability $h^2 = V_A/V_T$

Heritability can be different for different populations:

- Directional selection decreases V_A (alleles are being eliminated)
- Environmental differences affect V_T which changes h^2
- Different alleles and allele frequencies will affect V_A which changes h^2
- Heritability is generally low for traits that are very important to the organism, because there is little genetic variation in such traits

A mysterious exception

- Major exception: genes involved directly in sexual reproduction
 - Egg functions like egg membrane makeup
 - Sperm functions like egg membrane penetration
 - Seminal fluids
- Possible factors:
 - Evolutionary conflict between males and females
 - Pressure for reproductive isolation between species
- This is a hot area of research including Willie Swanson in this department

An error to avoid

- Suppose a trait has high heritability
- Does this mean it can never be affected by the environment?

An error to avoid

- Consider the $pk-$ mutation
- High phenylalanine (normal) environment:
 - $pk- / pk-$ individuals mentally retarded
 - Selection for IQ would reduce allele frequency of $pk-$
- Novel low phenylalanine environment:
 - $pk- / pk-$ have normal IQ
 - Selection for IQ would not reduce allele frequency of $pk-$
- pk would contribute to h^2 in one environment but not the other

An error to avoid

- The reverse of this: does low heritability mean that a trait is not genetic?
- Let's brainstorm a list of genetic traits with low heritability

Is this trait genetic or environmental?

- Not a well defined question:
 - Almost all traits have some genetic component
 - Almost all traits are influenced by environment
- This question is only meaningful for:
 - A particular gene pool or pools
 - A particular environment or range of environments

Two points of terminology

1. I defined $h^2 = V_A/V_T$ as “heritability”
 - Some authors call this “narrow sense heritability” and define “broad sense heritability” as V_G/V_T
 - Narrow sense heritability is the part that responds to breeding efforts, and to natural selection
2. The label h^2 is a square for historical reasons (like χ^2)
 - Its square root doesn't mean anything in particular!

How can we get unstuck?

A population that stops responding to selection can begin responding again:

- A new allele arises by mutation
- A new allele enters by migration
- Recombination breaks up an unhelpful linkage association
- The environment changes

Selection and linkage disequilibrium

- If there is no LD, selection on one gene changes allele frequencies at that gene
- With LD it can also affect linked genes
- Example:
 - Gene A codes for coat color – AA or Aa brown, aa gold
 - Gene B codes for epilepsy – BB or Bb normal, bb epileptic
 - Without LD, you can simply select for gold color
 - What if genes A and B are tightly linked and have LD?

Selection and linkage disequilibrium

- Suppose population contains mostly AB and ab haplotypes
- We select for gold color – aa genotypes
- What will happen to B locus?
- What could help: we need an aB haplotype
 - Recombination
 - Gene flow (if another population has it)
 - Mutation (could be a long wait)
- Recombination only works if B still present in population

Inbreeding

- Inbreeding is preferential mating with kin. The most severe form is self-fertilization.
- Inbreeding reduces heterozygosity
- We can define an inbreeding coefficient f representing the strength of inbreeding

A warning

- Is a small subpopulation “inbred” ?
 - It will have lower diversity
 - It will have more homozygotes
- BUT
 - If small population has random mating it will be in H-W
 - No preferential mating with kin *within the population*
- It depends on your perspective (whole population vs. subpopulation)

Inbreeding

$f = 1$ is complete self-fertilization

$f = 0$ randomly mating population

- We can think of inbreeding as dividing the population into two parts:
 - a fraction f which receive their two alleles from a single gene copy in an ancestor
 - a fraction $(1 - f)$ which receive their two alleles at random from the gene pool
- Inbreeding always increases homozygotes

Genotype frequencies with inbreeding

$$\text{frequency}(AA) = p^2 + f pq$$

$$\text{frequency}(Aa) = 2pq - 2f pq$$

$$\text{frequency}(aa) = q^2 + f pq$$

- Inbreeding does not itself change allele frequencies
- Selection on an inbred population:
 - Focuses more attention on homozygotes
 - May have a different outcome because of this

Effects of inbreeding

- With random mating any heterozygote advantage defines a stable polymorphism
- With increasing inbreeding the population is pushed toward one of the homozygotes
- At $f = 1$ the more fit homozygote will fix
- Intermediate values of f make the area of stable polymorphism smaller
 - Heterozygote has to be better to make up for their relative rareness
 - If heterozygote not good enough, best homozygote will fix

Inbreeding depression

Inbreeding depression is an observed loss of fitness in inbred organisms

- Heterozygote advantage
- Recessive deleterious alleles
- Excessive similarity among individuals (epidemic risk)

Do humans avoid inbreeding?

- Most cultures actively avoid brother/sister matings
- Some cultures forbid cousin marriages but others prefer them
- About 10% of marriages worldwide are first-cousin; up to 50% in some areas
- Outbred risk of birth defects: 3%
- First-cousin risk of birth defects: 4%
- This is comparable to the increased risk of having a child at age 40 rather than 30

Inbreeding and the gene pool

- It is often said that inbreeding is “bad for the gene pool”
- Small population size can be bad for the gene pool because of drift
- Inbreeding itself does not change allele frequencies, only genotype frequencies (pushes away from H-W)
- Homozygosity due to inbreeding disappears after one generation of random mating

Inbreeding and the gene pool

- Inbreeding coupled with selection can be good or bad
- It allows quicker loss of harmful recessives
- However, it also loses helpful overdominants
- Inbreeding is a useful tool in animal breeding to weed out bad alleles and fix good ones
- Outbreeding often produces more vigorous organisms—overdominance?

Inbreeding versus small size

- Inbreeding:
 - Increases homozygosity
 - Does not directly change allele frequencies
 - Does not eliminate alleles
 - Reveals recessives to selection (good or bad)
- Small population size:
 - Leads to rapid drift in allele frequencies
 - Can eliminate alleles or make them frequent
 - Diminishes the power of selection (good or bad)

One-minute responses

- Tear off a half-sheet of paper
- Write one line about the lecture:
 - Was anything unclear?
 - Did anything work particularly well?
 - What could be better?
- Leave at the back on your way out