

# Quantitative Trait Variation

# Variation in phenotype

- In addition to understanding genetic variation within at-risk systems, phenotype variation is also important.
  - reproductive fitness
    - traits related to the production of fertile offspring that survive to reproductive age.
  - life history traits
    - e.g. flowering time, reproductive age
  - morphology
  - disease resistance

# Phenotypic Variation



- Which population has more variation?

# The answer ...



For quantitative  
traits, we measure  
variation via

Variance ( $\sigma^2$ )

# Variance ( $\sigma^2$ )

- Character (pheno) must have numeric measurements.
- The value of  $\sigma^2$  depends on:
  1. the measured outcomes possible for that character;
  2. the probabilities of seeing those outcomes.

# Variance ( $\sigma^2$ ): tossing a die

**Case 1: a fair die (all values equally likely)**

• Outcome	probability
• 1	1/6
• 2	1/6
• 3	1/6
• 4	1/6
• 5	1/6
• 6	1/6

$\sigma^2 =$  \_\_\_\_\_

**Case 2: a loaded die (biased towards a given outcome)**

• Outcome	probability
• 1	1/10
• 2	1/10
• 3	1/10
• 4	1/10
• 5	1/10
• 6	1/2

$\sigma^2 =$  \_\_\_\_\_

# Variance ( $\sigma^2$ ): tossing a die

**Case 1: a fair die (all values equally likely)**

• Outcome	probability
• 1	1/6
• 2	1/6
• 3	1/6
• 4	1/6
• 5	1/6
• 6	1/6

$$\sigma^2 = 2.91$$

**Case 2: a loaded die (biased towards a given outcome)**

• Outcome	probability
• 1	1/10
• 2	1/10
• 3	1/10
• 4	1/10
• 5	1/10
• 6	1/2

$$\sigma^2 = 3.25$$



# Variance ( $\sigma^2$ ): tossing a die

**Case 1: a fair die (all values equally likely)**

• Outcome	probability
• 1	1/6
• 2	1/6
• 3	1/6
• 4	1/6
• 5	1/6
• 6	1/6

$\sigma^2 =$  \_\_\_\_\_

**Case 3: high probability of extreme values**

• Outcome	probability
• 1	1/2
• 2	0
• 3	0
• 4	0
• 5	0
• 6	1/2

$\sigma^2 =$  \_\_\_\_\_

# Variance ( $\sigma^2$ ): tossing a die

**Case 1: a fair die (all values equally likely)**

• Outcome	probability
• 1	1/6
• 2	1/6
• 3	1/6
• 4	1/6
• 5	1/6
• 6	1/6

$$\sigma^2 = 2.91$$

**Case 3: high probability of extreme values**

• Outcome	probability
• 1	1/2
• 2	0
• 3	0
• 4	0
• 5	0
• 6	1/2

$$\sigma^2 = 6.25$$

# Variance ( $\sigma^2$ )

- Variance is maximized ...
  - when outcomes are at their two extremes
    - low and high, with similar frequencies.
  - not when the number of likely outcomes is large.
  - diversity vs. high variance.

# Variance

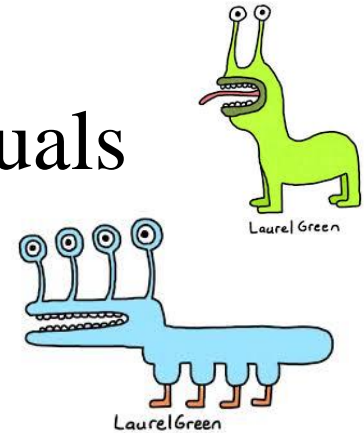
- Note that there is a difference between the model variance ...
  - $\sigma^2$ 
    - (we'll talk about this first)
- And an *estimate* of variance
  - collect data; use it to calculate sample variance.
    - (we'll talk about this second)

# Partitioning variance

- Identifying **sources of variance**
  - factors that are responsible for individuals looking different from one another.

# Sources of Variance

- Factors that are responsible for individuals looking different from one another.
- Number of eyes on a Martian ...

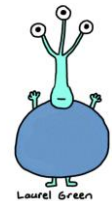


## Male Martians

<u># eyes</u>	<u>probability</u>
• 1	0.15
• 2	0.60
• 3	0.25

## Female Martians

<u># eyes</u>	<u>probability</u>
• 3	0.15
• 4	0.60
• 5	0.25



- Variance among a male-only population; a female-only population; a mixed population

# Factors that may affect variance in phenotype

- Genetic variability in populations
  - Individuals are genetically different from one another. However ...
  - not all genetic variation creates differences in phenotypes.
- Variation in environmental exposures
  - Different individuals have experienced different environmental conditions.
  - These differences also may or may not create differences in phenotypes.

# Pheno variance and selection

- If individuals in a population are monomorphic for a given trait, selection cannot act on that trait.
- If there is variation in the trait, but individuals are genetically invariant at the loci that contribute to this trait, selection cannot act on the trait.



# Pheno variance and selection

- For selection to act on a trait:
  1. genetic variation must exist in the population; AND,
  2. some variation in the trait must be due to genetic differences between individuals.

# Pheno variance and selection

- How much of the variation in the trait is due to individuals in the population having different genotypes?

# Phenotypes: what is “genetic”?

- Flamingo color ...

[www.thoughtco.com/why-are-flamingos-pink-607870](http://www.thoughtco.com/why-are-flamingos-pink-607870)

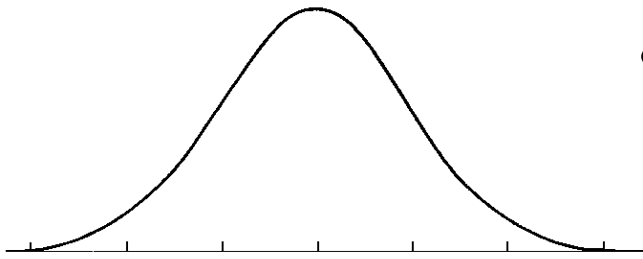


# Pheno variance and selection

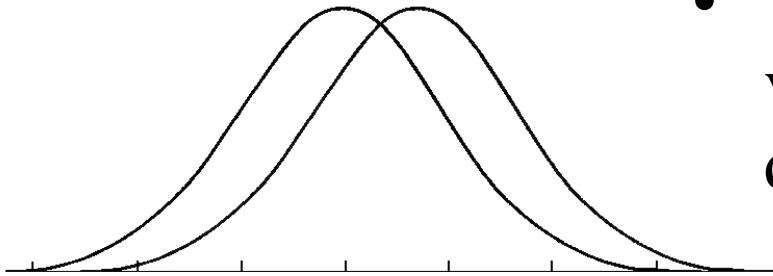
- How much of the variation in the trait is due to individuals in the population having different genotypes?
- Note: this is a different question than asking how many loci, or which loci, or are there loci at all, responsible for the development of the trait (e.g. is the trait “genetic”)
  - remember, if these loci exist, but everyone in the population carries the same alleles at these loci, then variation in this trait is not due to genetic differences (and selection cannot act on the trait).

# Heritability

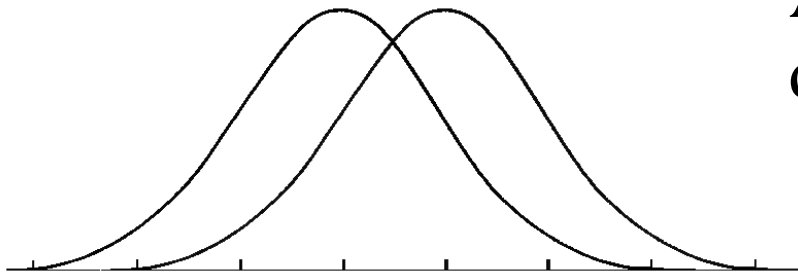
A measure of how much of the population variance of a trait is due to individuals in the population having different genotypes



- Variance of a phenotype that follows ← this distribution



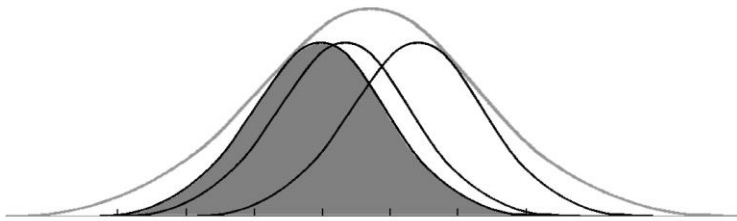
- What happens to overall variance if males and females differ slightly?



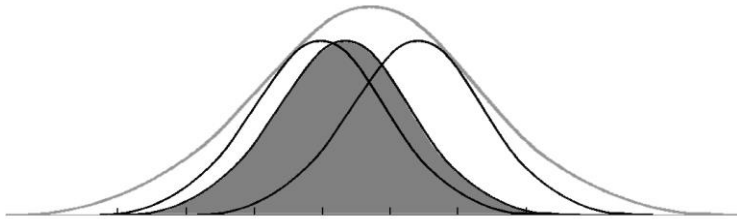
- And to overall variance if they differ even more?

- Note that it's not enough just to have both males and females in the population – they need to differ.
- And note that there's no effect on variance if males and females differ, but there are only males or only females in the population.

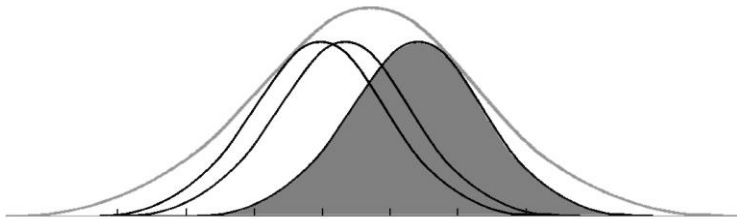
# A genetic locus



bb



Bb



BB

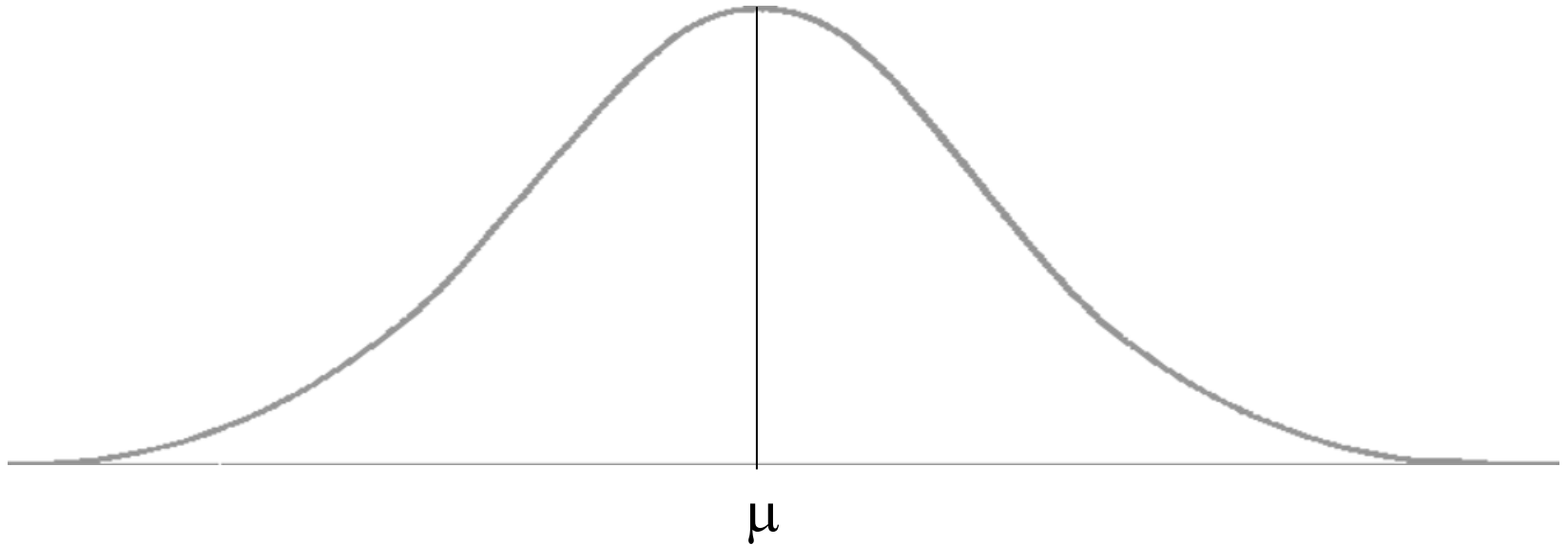
- If there are different genotypes present in the population,
- and, if they have different effects on the phenotype,
- This will create larger variance in phenotype
  - than in a monomorphic population
  - or than there would be if all genotypes had the same effect on phenotype.

# Effect of genotypes on variance

- The question is: how much of the variance in phenotype can be explained by the presence of different genotypes in the population?
  - Quantify *how* different individuals are *because* they have different genotypes.
- Total phenotype variance:  $\sigma^2_P$
- Phenotype variance that can be explained by individuals having different genotypes:  $\sigma^2_G$

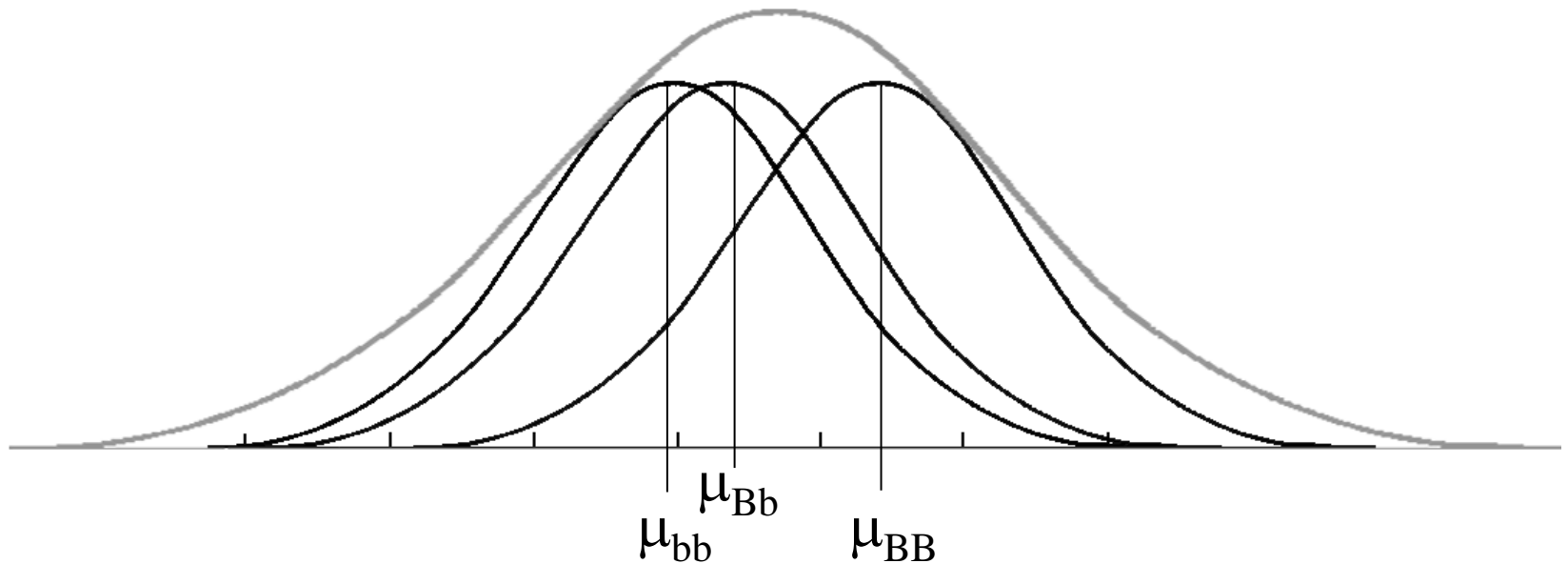


# Total phenotype variance, $\sigma^2_P$



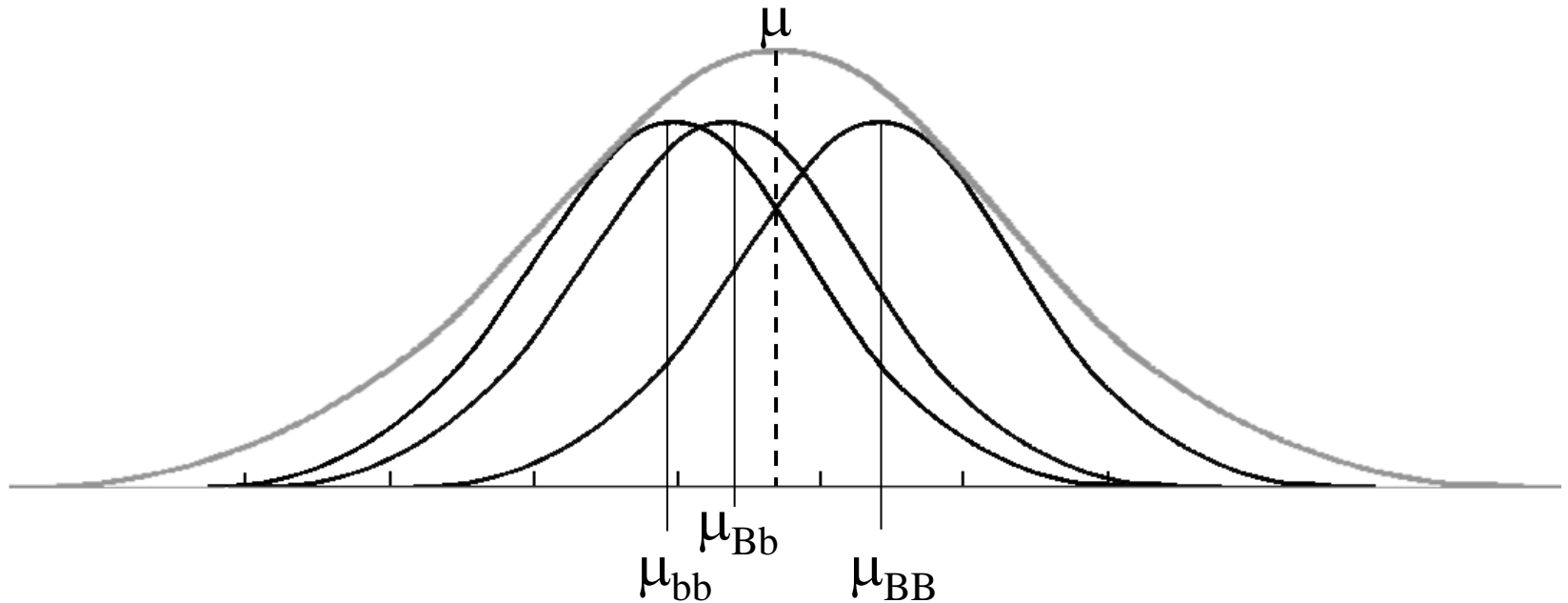
- $\sigma^2_P = E [ (X - \mu)^2 ]$

# Genotypes with different effects on the phenotype



- $\mu_{bb}$     $\mu_{Bb}$     $\mu_{BB}$
- Genotype values: mean phenotype for all individuals with that genotype

# Genotypes with different effects on the phenotype



- $\sigma^2_G = E[ (\text{difference between the genotype values and the population mean})^2 ]$
- genotype values:  $\mu_{bb}$   $\mu_{Bb}$   $\mu_{BB}$   
(mean phenotype for all individuals with that genotype)

# Variance in phenotype caused by genetic variation: $\sigma^2_G$

- $\sigma^2_G = E[ (\text{difference between the genotype values and the population mean})^2 ]$

- $\sigma^2_G = E [ (X - \mu)^2 ] =$   
 $p^2_B (\mu_{BB} - \mu)^2 + 2p_B p_b (\mu_{Bb} - \mu)^2 + p^2_b (\mu_{bb} - \mu)^2$

# Broad sense heritability, $H^2$

- $\sigma^2_P$  : variance in phenotype
  - measures how different individuals are from one another ...
  - for any reason.
- $\sigma^2_G$  : variance in phenotype
  - measures how different individuals are from one another ...
  - *because* they have different genotypes.
- $\sigma^2_G \leq \sigma^2_P$ 
  - because  $\sigma^2_G$  is probably only one part of  $\sigma^2_P$ .

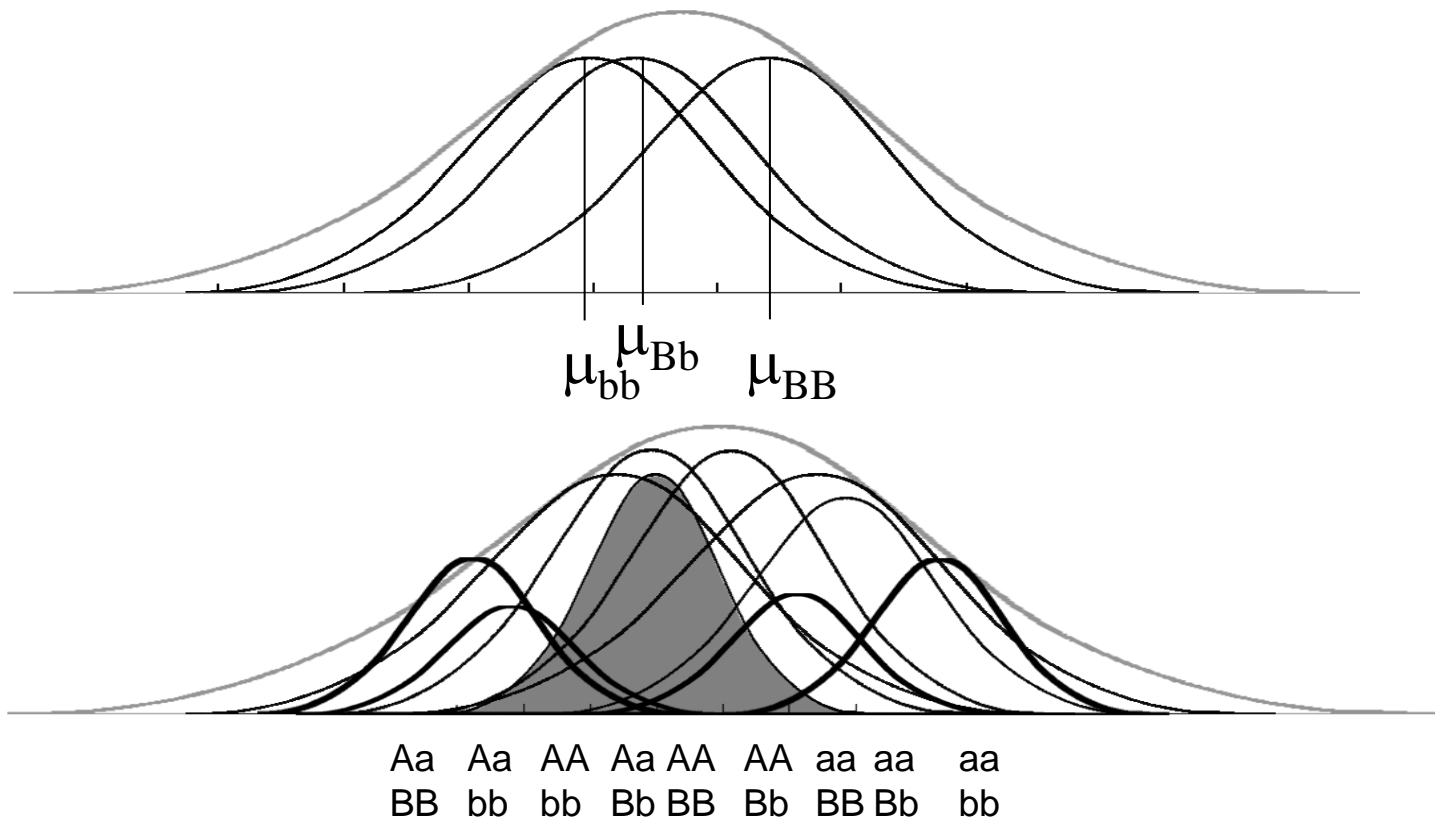
$$H^2 = \sigma^2_G / \sigma^2_P$$

- $\sigma^2_P$  : variance in phenotype
  - measures how different individuals are from one another ...
  - for any reason.
- $\sigma^2_G$  : variance in phenotype
  - measures how different individuals are from one another ...
  - *because* they have different genotypes.
- $\sigma^2_E$  : variance in phenotype
  - measures how different individuals are from one another ...
  - because they have experienced different environmental exposures.

$$H^2 = \frac{\sigma_G^2}{\sigma_P^2} = \frac{\sigma_G^2}{\sigma_G^2 + \sigma_E^2}$$

# Broad sense heritability, $H^2$

- Of all the phenotypic variance in the population, what proportion of it is because individuals have different genotypes.



# Flamingos

- Discussion exercise ...
  - What population characteristics would yield low  $H^2$  ?
  - What about high  $H^2$ ?



# Heritability

- *Proportion* of phenotypic variance that can be explained by variation in genotypes.
- Depends on allele frequencies.
  - ➔ Varies from population to population.
    - different allele frequencies.
    - different environmental conditions.
  - ➔ Varies over time within a population.
    - changing allele frequencies.
    - changing environmental conditions.

# Narrow sense heritability, $h^2$

- To understand  $h^2$ , you need to understand additive genetic variance
  - $\sigma^2_A$  (“A” for additive)
  - phenotype variance that can be inherited from one generation to the next.
    - Can be predicted for offspring based on parents.
- To understand  $\sigma^2_A$ , it’s helpful to understand additive effects of alleles
  - $\alpha_i$  (“i” for allele i)

# Additive effects of alleles

- Phenotypic values associated with alleles.
- Theoretical constructs
  - (alleles themselves don't have phenotypes)
- Needed because individuals don't pass along their genotypes to their offspring, they pass along their alleles.
  - The pheno component that is inherited from a parent to an offspring is connected to the values of alleles.

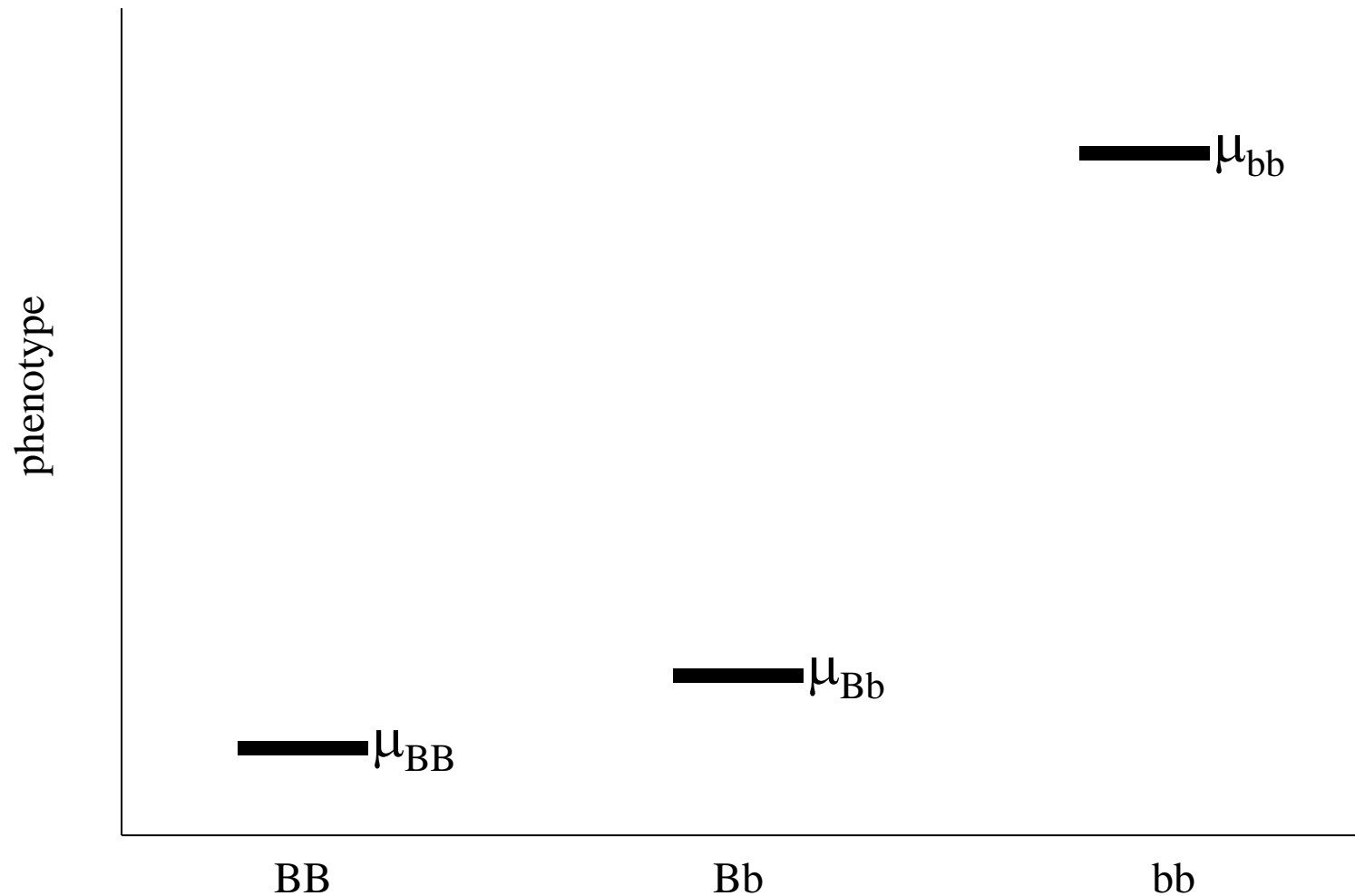
# Additive effects of alleles

- $\alpha_B =$   
E [phenotype for all BB and Bb indivs]  $- \mu$  .
  - How much do individuals who carry at least one B allele differ from the overall population mean?
- $\alpha_B = p_B \mu_{BB} + p_b \mu_{Bb} - \mu$ .
  - depends on allele frequencies
  - If the b allele is rare, then there will be a lot more BB individuals than Bb
    - which affects the mean phenotype of the group of BB and Bb individuals.

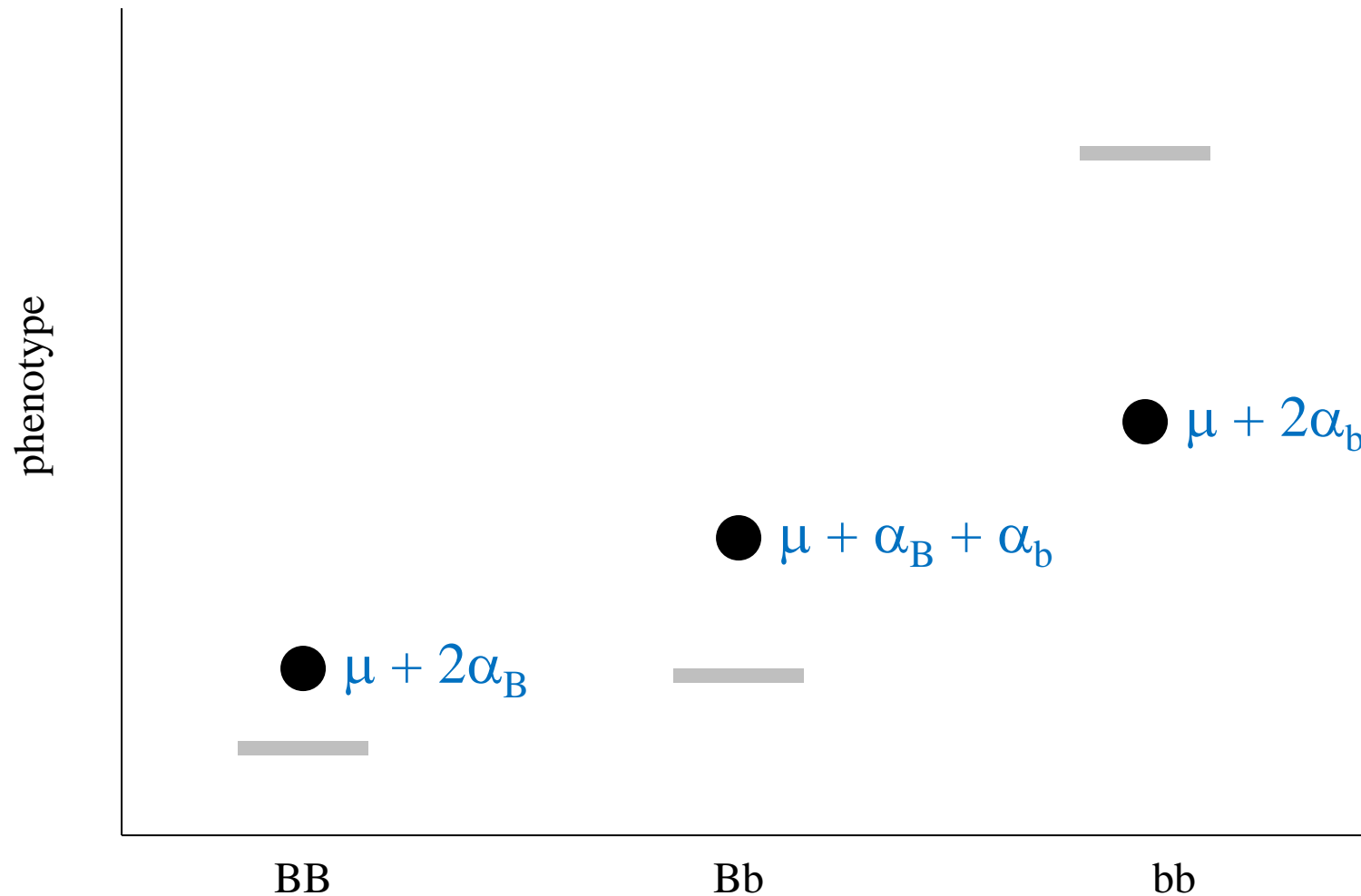
# Genotype values & additive effects

- What if all you knew about a phenotype were the additive effects of alleles?
  - (for all alleles affecting the trait; let's assume one locus, two alleles for now)
- Could you describe the genotype values in terms of the additive effects of alleles?
  - $\mu_{BB} = \mu + \alpha_B + \alpha_B = \mu + 2\alpha_B$
  - $\mu_{Bb} = \mu + \alpha_B + \alpha_b$
  - $\mu_{bb} = \mu + \alpha_b + \alpha_b = \mu + 2\alpha_b$
- Is this reasonable? Would it be perfect?

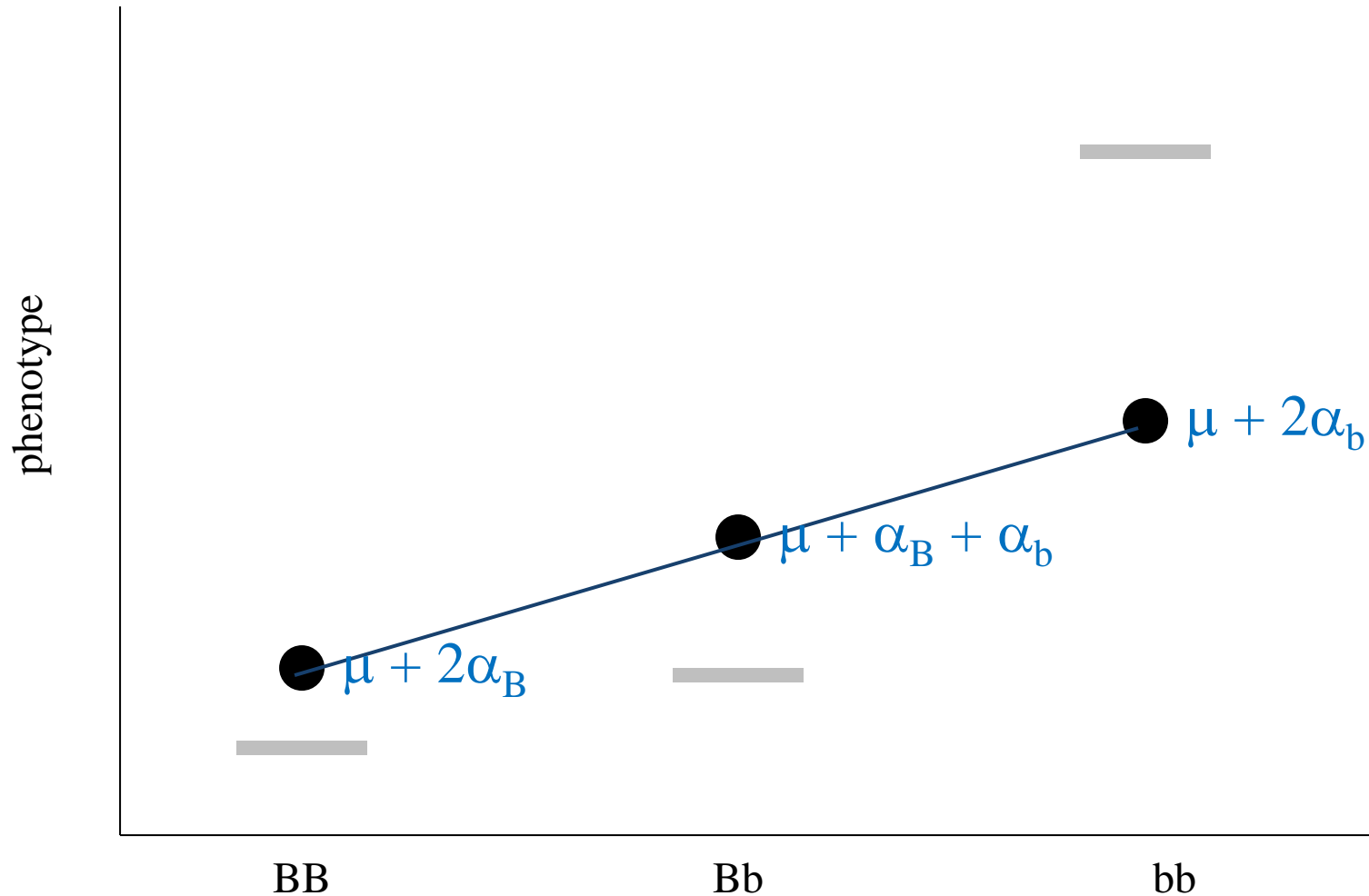
# Genotype values ( $\mu_{BB}$ , $\mu_{Bb}$ , $\mu_{bb}$ )



# Genotype values predicted from additive effects

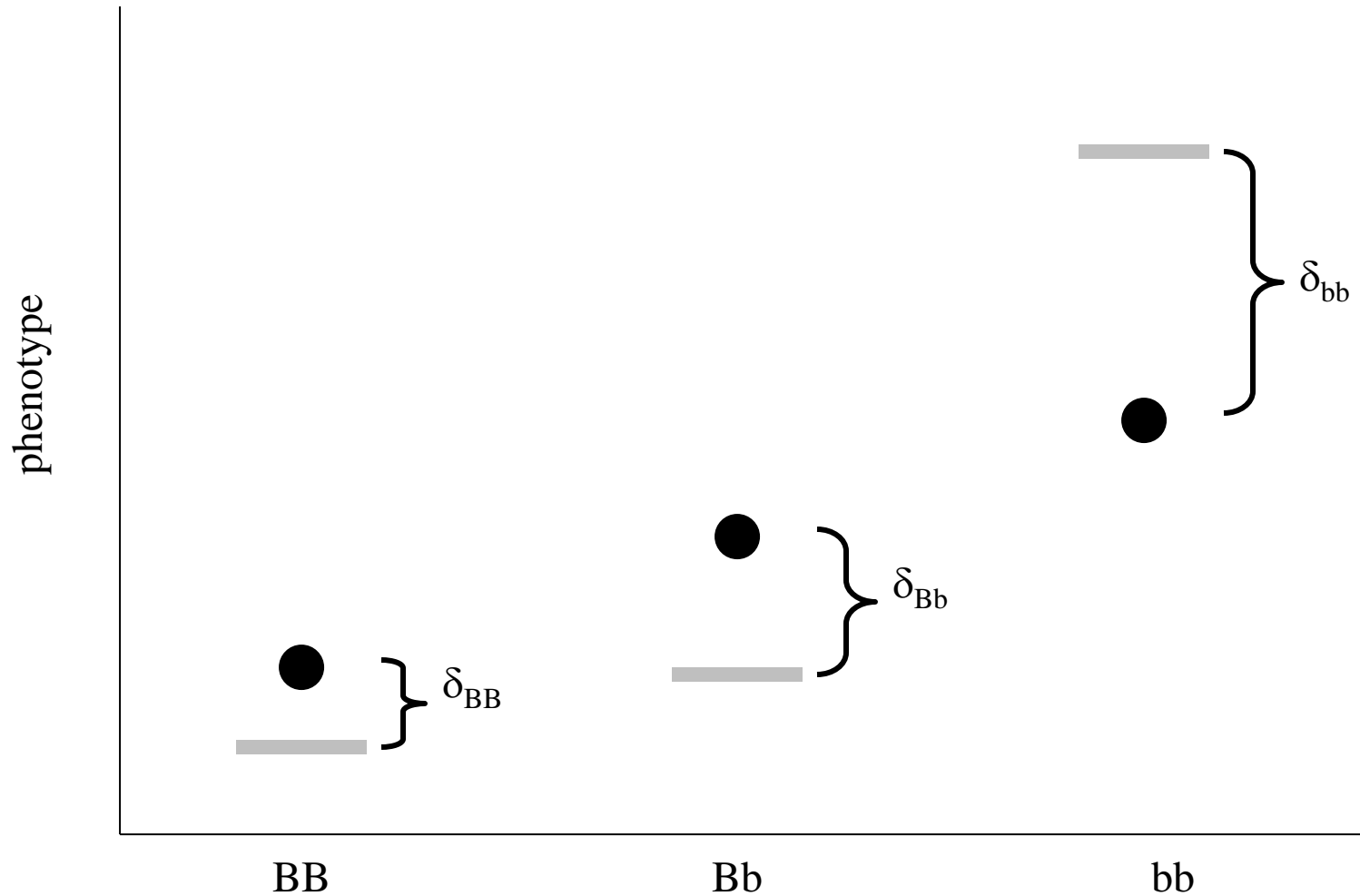


# Genotype values predicted from additive effects

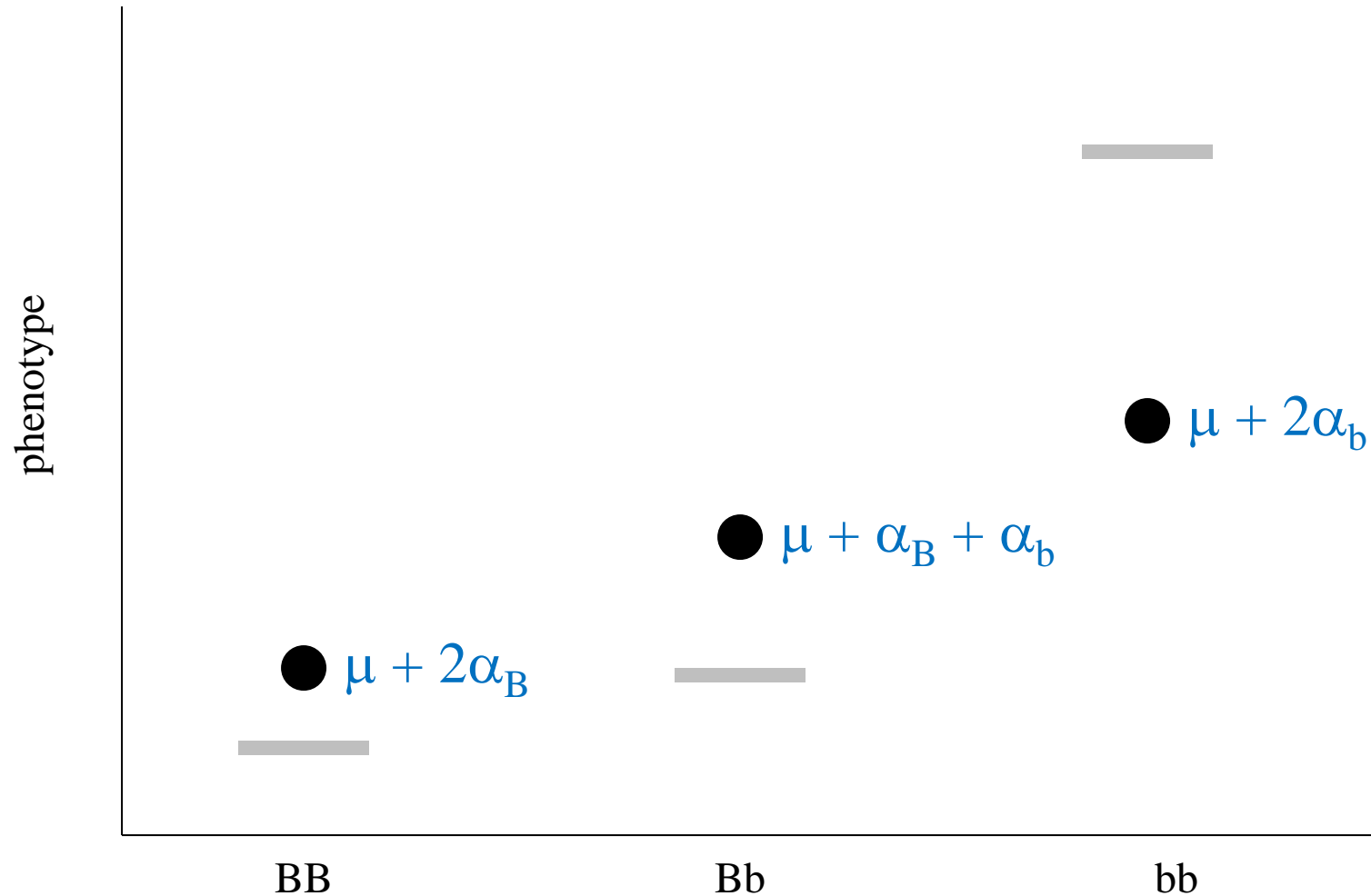




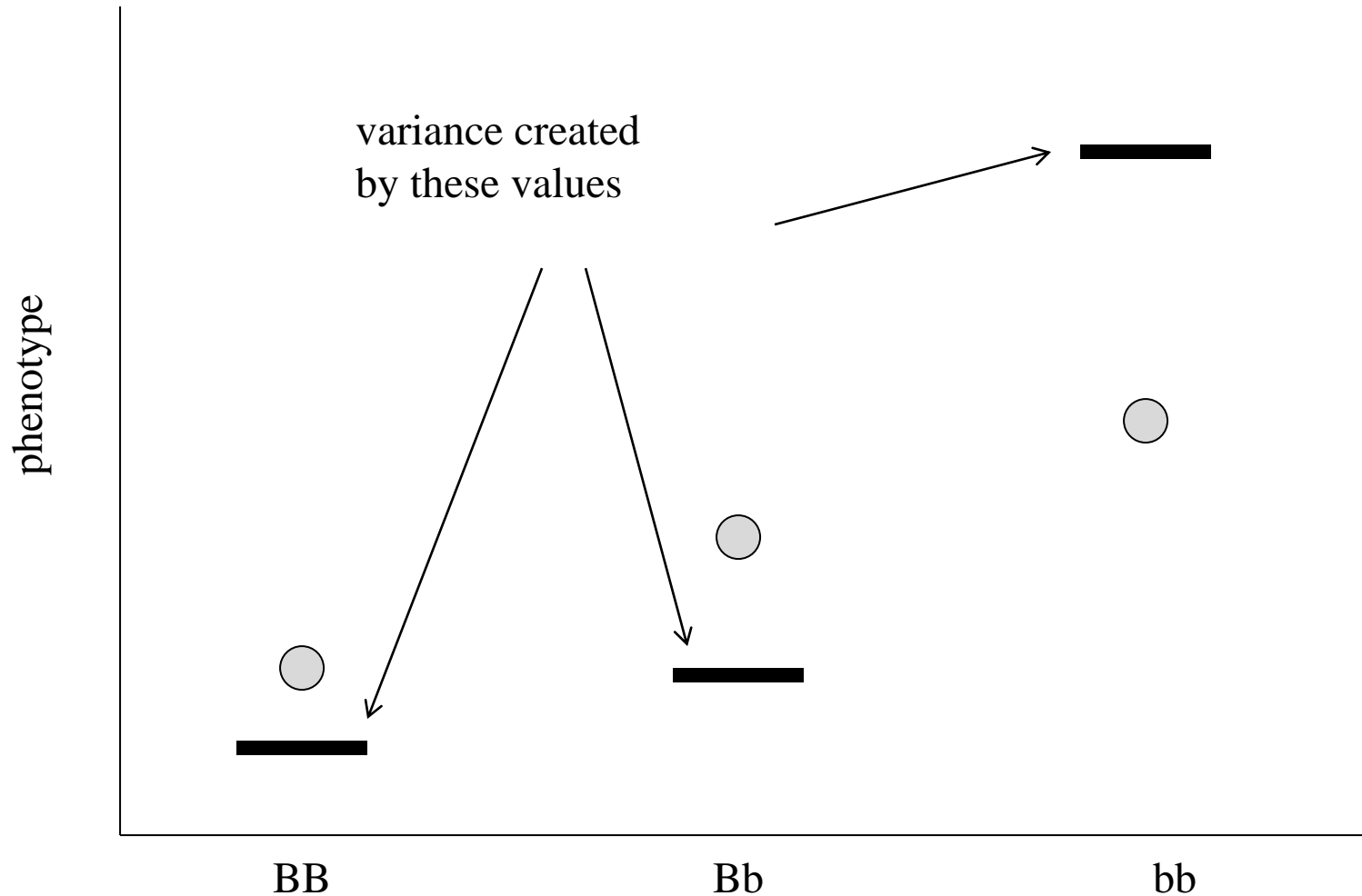
# Dominance deviations



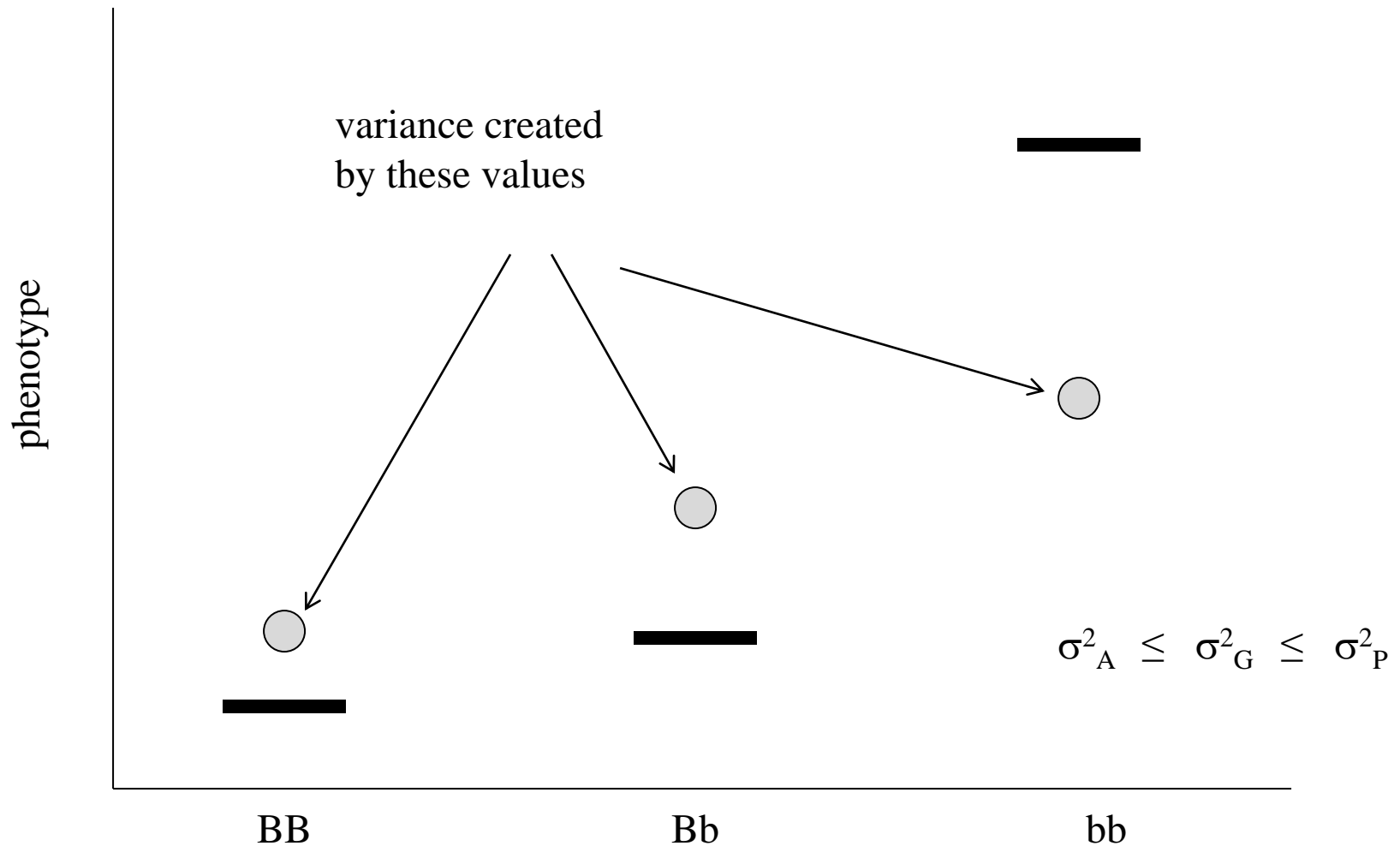
# Genotype values predicted from additive effects



$\sigma^2_G$  : variance in phenotype due to variation in genotype



# $\sigma^2_A$ : Additive genetic variance



# $\sigma^2_A$ : Additive genetic variance

- $\sigma^2_G =$

$$p^2_B (\mu_{BB} - \mu)^2 + 2p_B p_b (\mu_{Bb} - \mu)^2 + p^2_b (\mu_{bb} - \mu)^2$$

- $\sigma^2_A = E [ (X - \mu)^2 ] =$

$$p^2_B (\mu + 2\alpha_B - \mu)^2$$

$$+ 2p_B p_b (\mu + \alpha_B + \alpha_b - \mu)^2$$

$$+ p^2_b (\mu + 2\alpha_b - \mu)^2$$

- $\sigma^2_A = p^2_B (2\alpha_B)^2 + 2p_B p_b (\alpha_B + \alpha_b)^2 + p^2_b (2\alpha_b)^2$

# $\sigma^2_A$ : Additive genetic variance

- $\sigma^2_A = p_B^2 (2\alpha_B)^2 + 2p_B p_b (\alpha_B + \alpha_b)^2 + p_b^2 (2\alpha_b)^2$ 
  - [lots of algebra]
- $\sigma^2_A = p_B \alpha_B^2 + p_b \alpha_b^2$ 
  - [more algebra]
- $\sigma^2_A = 2p_B p_b \alpha^2$ 
  - $\alpha =$  additive effect of allele substitution ( $\alpha_B - \alpha_b$ )

# Narrow-sense heritability: $h^2$

- $h^2 = \sigma^2_A / \sigma^2_P$
- Of all the variance in phenotype in the population, what proportion of it is explained by the effects of the alleles that are segregating in the population.
- Explains what proportion of variance in phenotype can be inherited from one generation to the next
  - Because *alleles* are transmitted from one generation to the next, not genotypes.

# Narrow-sense heritability: $h^2$

- $h^2 = \sigma^2_A / \sigma^2_P$
- Of all the variance in phenotype in the population, what proportion of it is explained by the additive effects of the different alleles that are segregating.
  - what proportion of it can be inherited from one generation to the next
    - can be predicted for offspring by knowing the parents.
- Because *alleles* are transmitted from one generation to the next, not genotypes.



# Estimating heritability

- If one of the reasons individuals look different from each other in a population is because individuals are genetically different
  - (meaning genetic variation is a source of phenotype variation)
- Then, groups of individuals who are more genetically similar to each other (relatives) should also be more phenotypically similar.

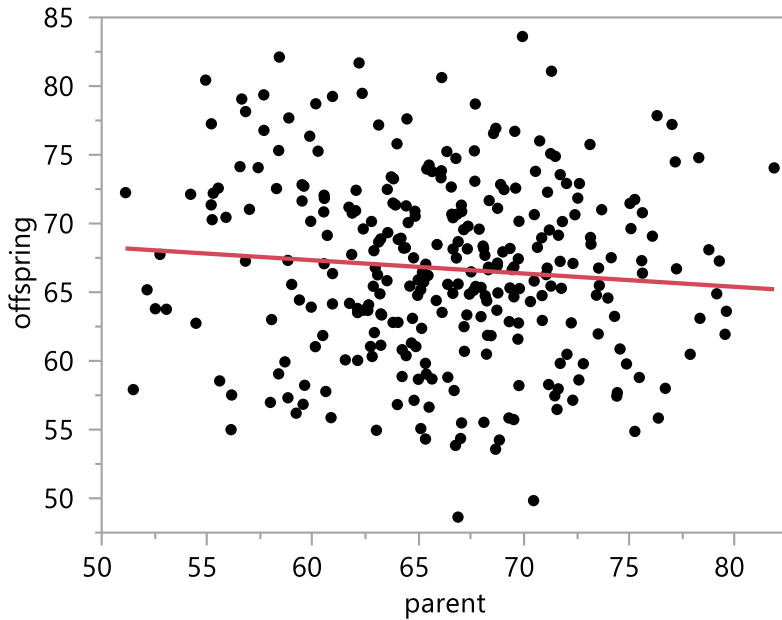
# Flipping this around

- If relatives resemble each other more than unrelated individuals do ...
- then this is an indication that some of the variation in phenotype in the population is due to genetic variation in the population.
  - which indicates that heritability is greater than zero.
- Use this to estimate heritability.

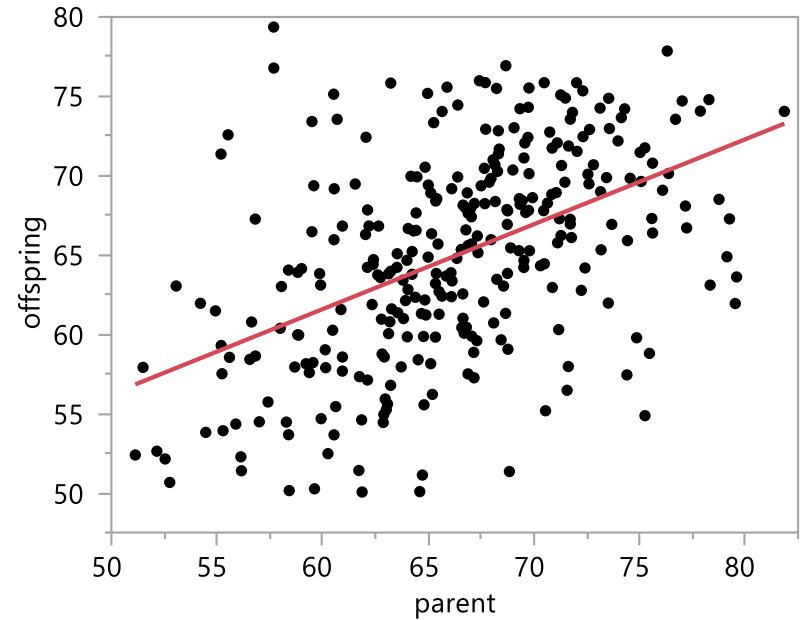
# Estimating $h^2$ : parent-offspring

- We know a parent shares one allele with its offspring IBD (at each locus).
- If offspring resemble their parents more closely than offspring resemble random individuals
  - tall parents have tall offspring
  - short parents have short offspring
- then allelic variation segregating in the population contributes to pheno var.

# Estimating $h^2$ : parent-offspring



- Offspring don't resemble their parents more closely than they resemble unrelated individuals.
- Individuals may look different from each other, but not because of genetic differences.
- Allelic variation does not contribute to pheno variance.
  - heritability is low to zero.



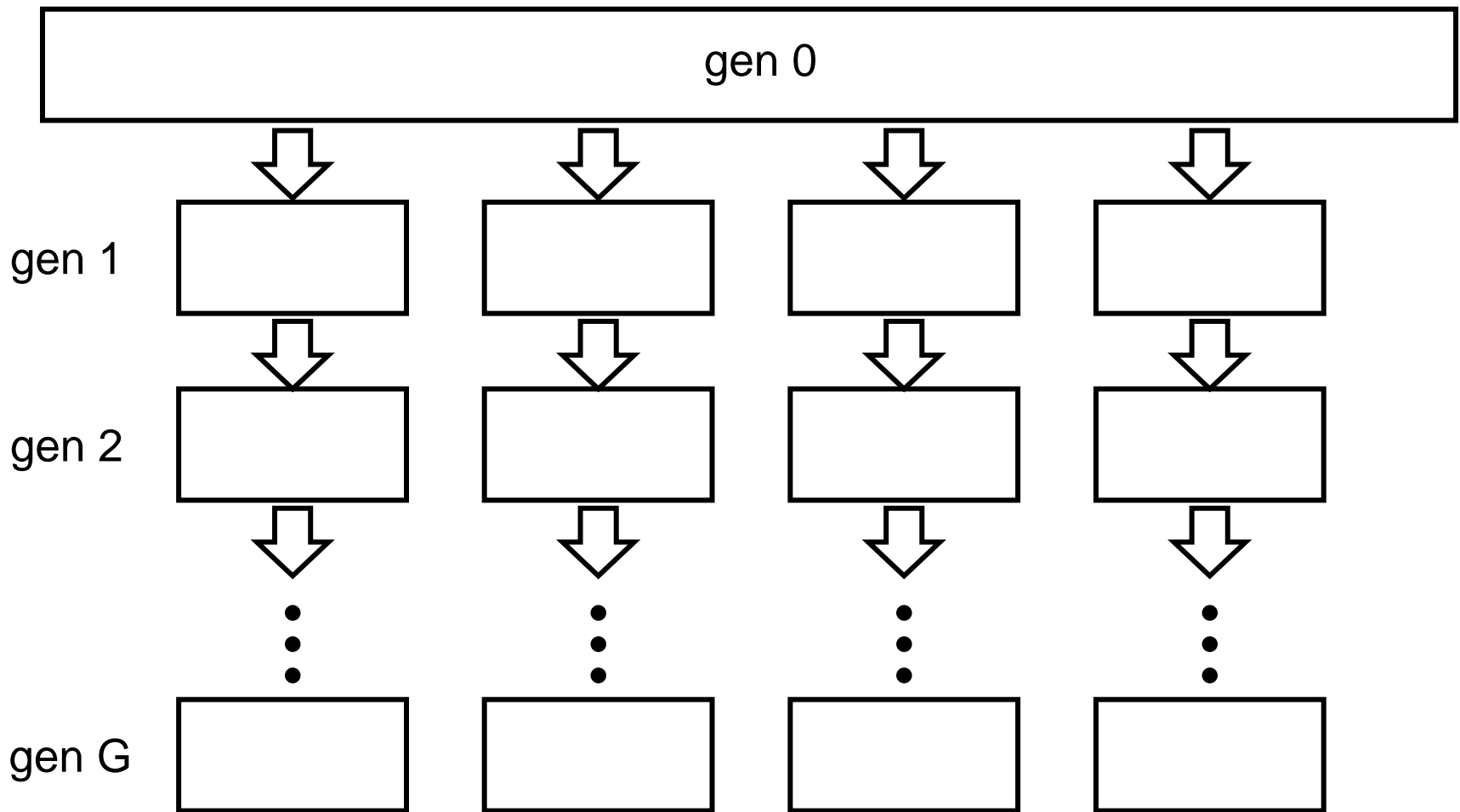
- Offspring's phenotypes are correlated with their parents' phenotypes.
- One of the reasons individuals look different from one another is because of genetic differences.
- Allelic variation is contributing to pheno variance.
  - heritability is high.

# Narrow-sense heritability: $h^2$

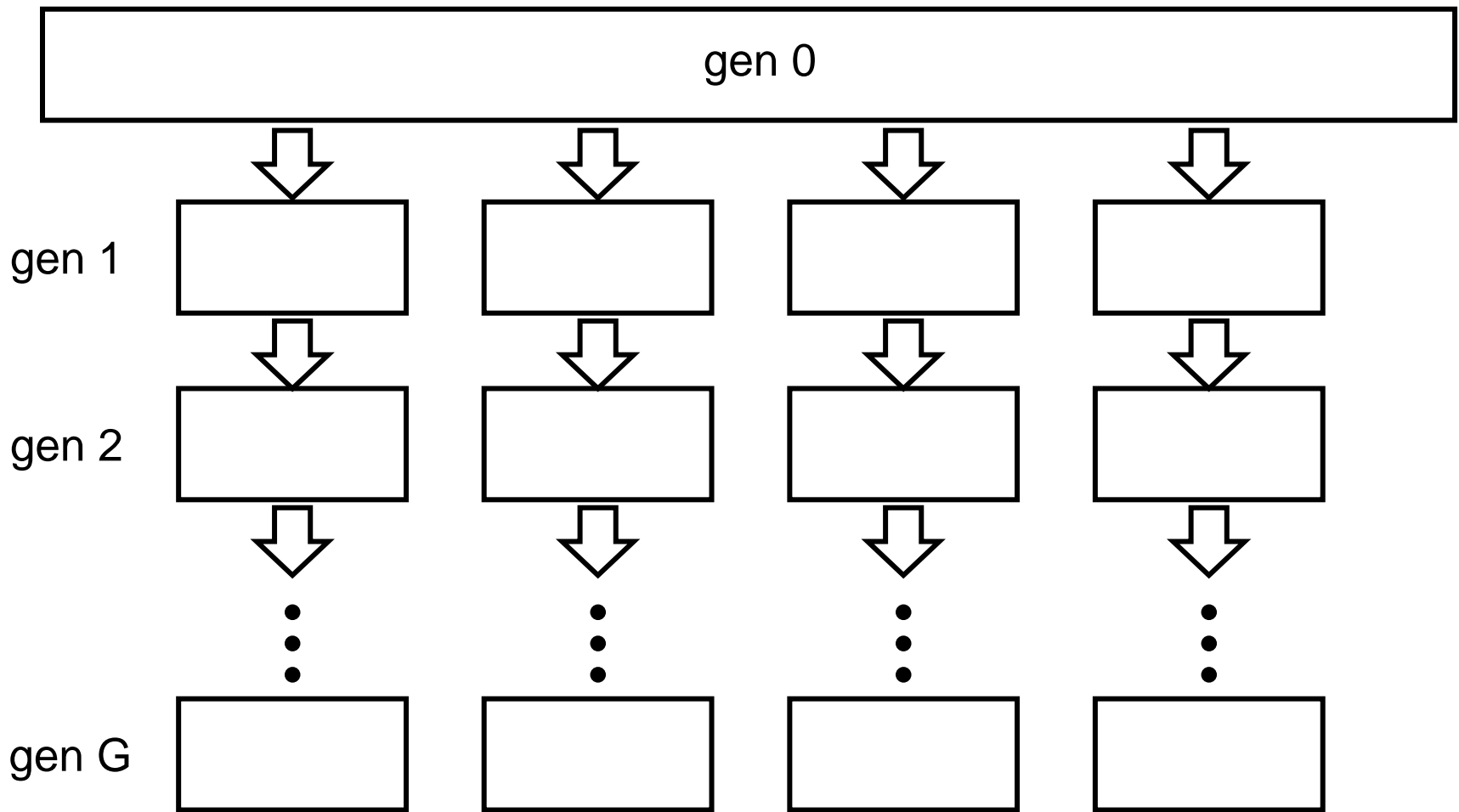
- It is the measure that predicts how well a trait responds to selection.
- Fitness traits: high selection pressure.
- Expectation that  $h^2$  will generally be low for traits associated with fitness.

# Quantitative trait variation in fragmented populations

- Remember the consequences for genetic variation in fragmented populations ...



Early in the process (before much time has passed after fragmentation), the subpopulations' genetic composition will be similar to each other. Allele frequencies will be similar (variance will be low).



Later in the process, genetic variation will start to be lost within the individual subpopulations. Which alleles become rarer in each subpopulation is random. Variation in  $p$  gets larger.



# Quantitative trait variation in fragmented populations

- Alleles underlying quantitative trait variation are also undergoing drift.
  - Allele frequencies increasing/decreasing.
- Genetic variation is being lost within subpopulations.
- Variance in phenotype ( $\sigma^2_G$ ) is being lost within subpopulations relative to the total population.

# Quantitative trait variation in fragmented populations

- Alleles underlying quantitative trait variation are undergoing drift.
- Selection for fitness traits is also potentially occurring.
  - Selection pressures may be similar across subpopulations.
  - Or different pressures may exist for different subpopulations.
    - driving differences between them.

# Quantitative trait variation in fragmented populations

- $Q_{ST}$  measures quantitative trait differentiation between subpopulations.

- $$Q_{ST} = \frac{\sigma_B^2}{\sigma_B^2 + \sigma_W^2}$$

- $\sigma_B^2$  = variance in phenotype explained by differences between subpopulations.

- $\sigma_W^2$  = variance in phenotype of individuals (or families) within subpopulations.

# Quantitative trait variation in fragmented populations

- $Q_{ST}$  measures quantitative trait differentiation between subpopulations.
- Measures how much variance in phenotype can be explained by individuals being members of different subpopulations.

# $Q_{ST}$ and $F_{ST}$

- These parameters have been shown to be equivalent under neutral subdivided population models:
  - Drift is the only force affecting allele frequencies.
- If  $Q_{ST} \neq F_{ST}$ , this could be a signature of selection.
  - Stabilizing selection:  $Q_{ST} < F_{ST}$
  - Diversifying selection:  $Q_{ST} > F_{ST}$