

Unit 6 | QUANTITATIVE GENETICS

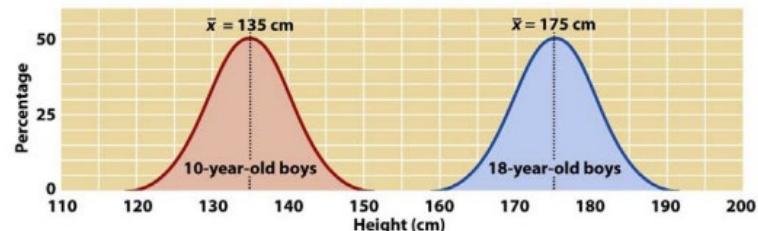


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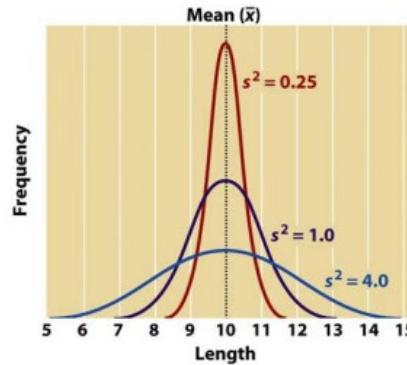
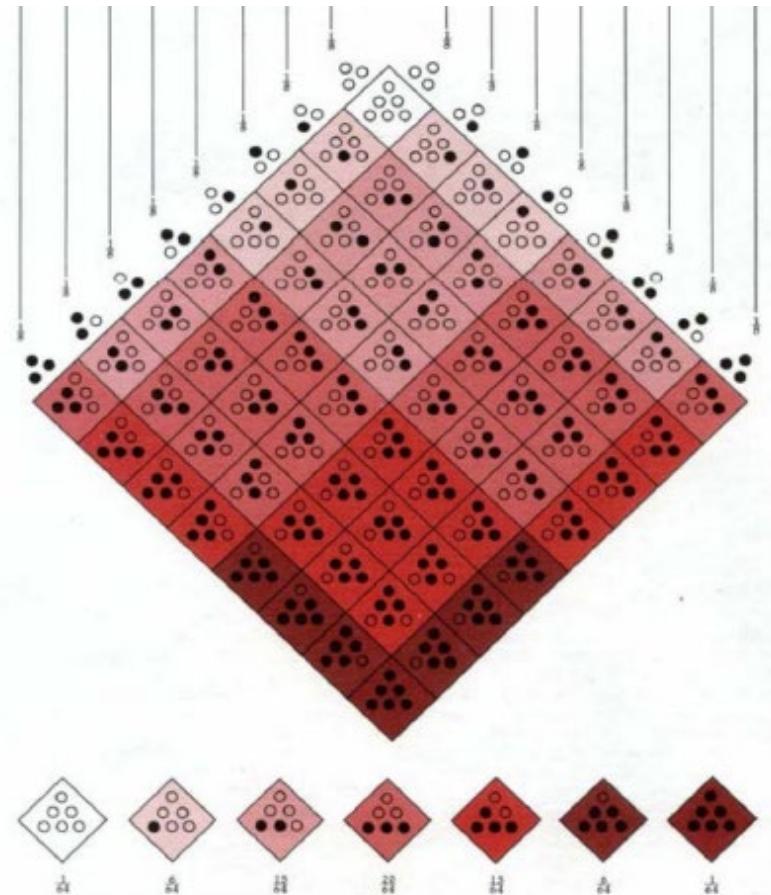


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Biometry vs Mendelian Genetics

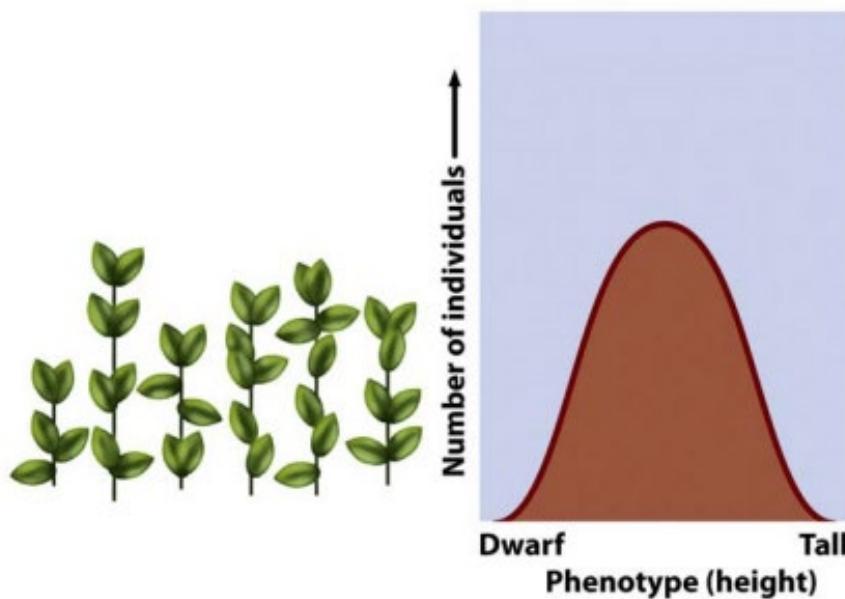
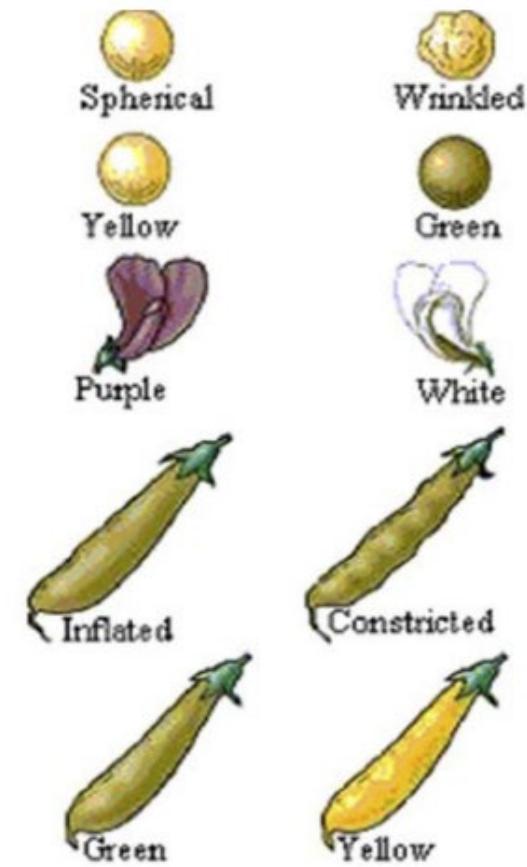


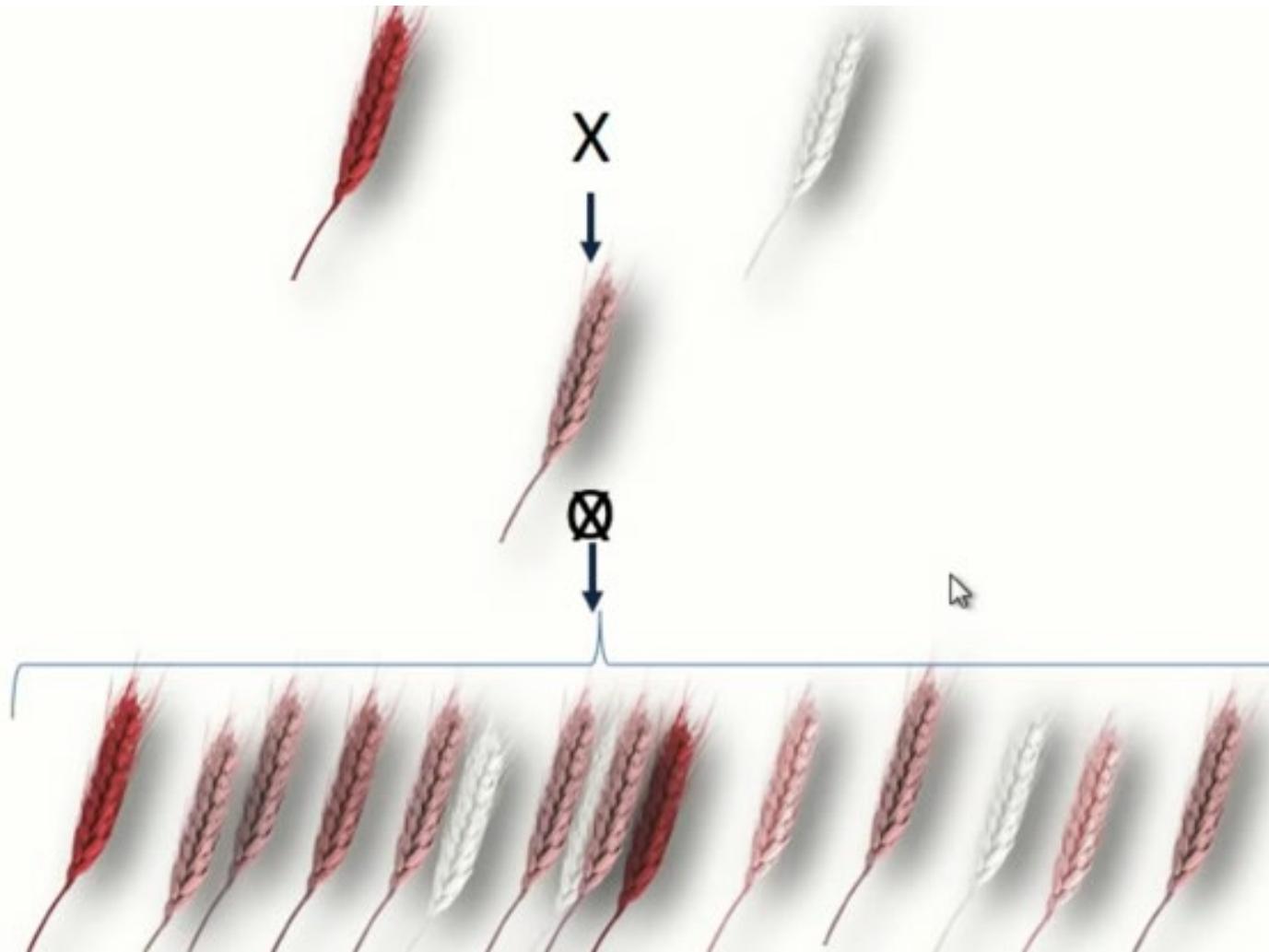
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Continuous variation

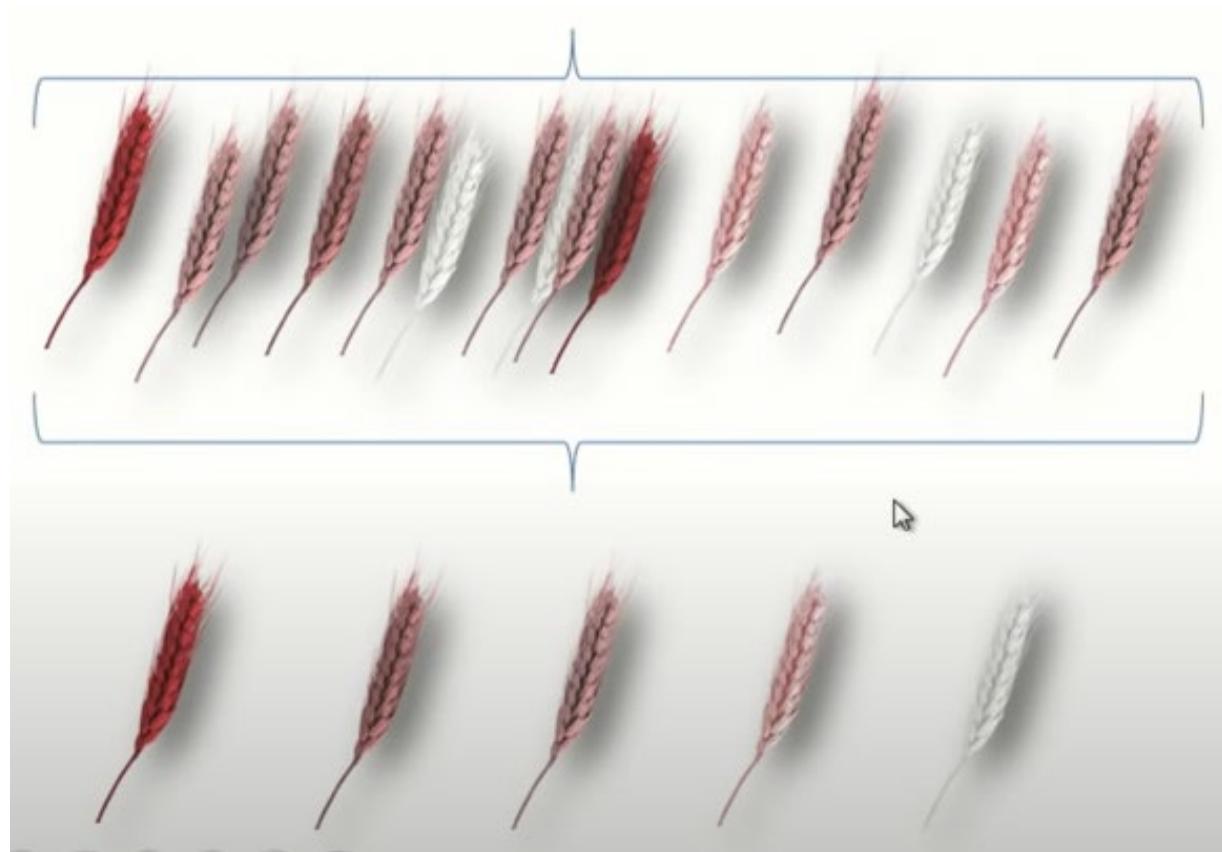


Discontinuous variation

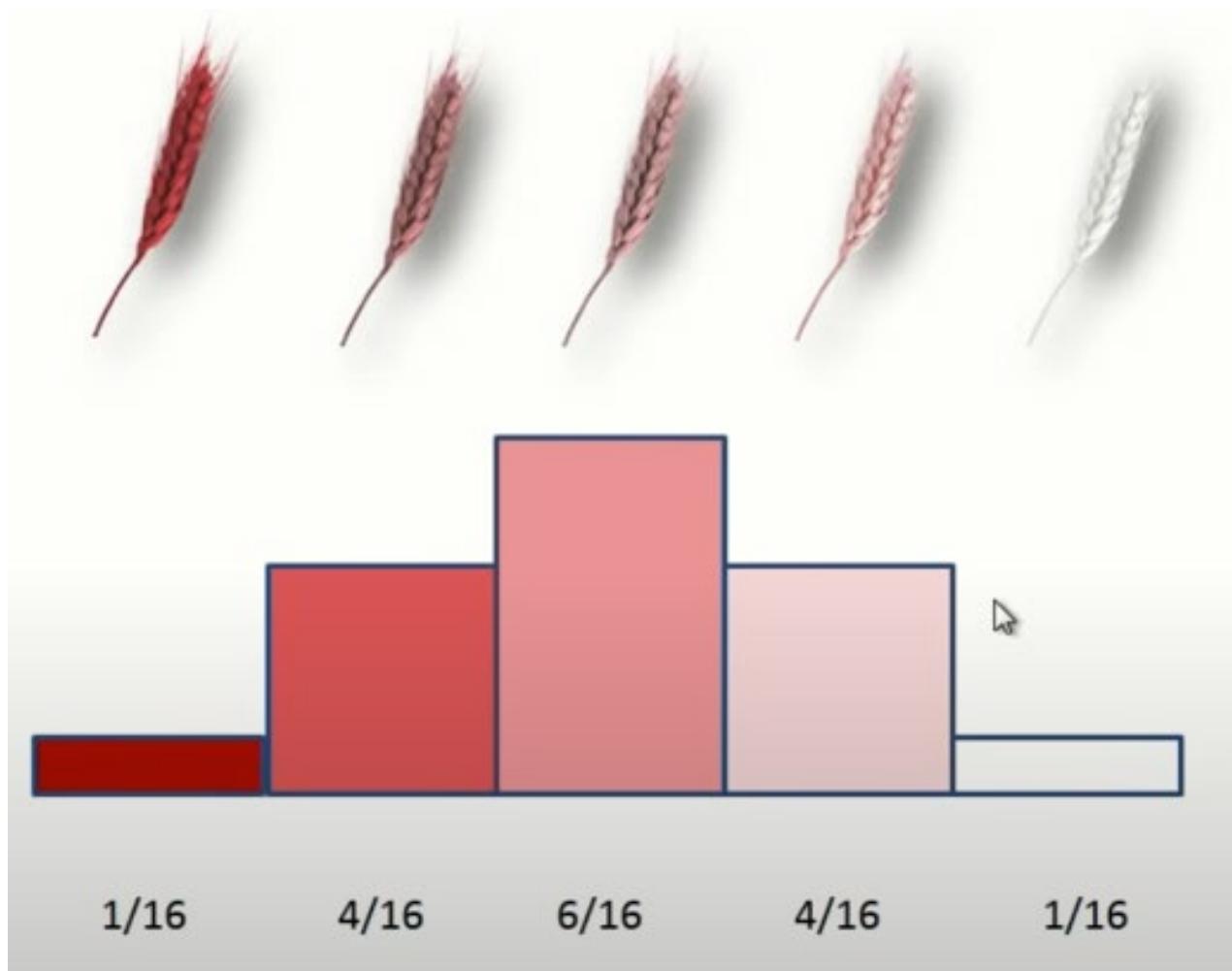
Nilsson-Ehle Experiment (1909)



Nilsson-Ehle Experiment (1909)



Nilsson-Ehle Experiment (1909)



Nilsson-Ehle Experiment (1909)

Methods

Cross wheat having white kernels and wheat having purple kernels. Intercross the F₁ to produce F₂.

P generation

$$A^+A^+B^+B^+ \times A^-A^-B^-B^-$$



Results

F₁ generation

$$A^+A^-B^+B^-$$

Break into simple crosses

$$A^+A^- \times A^+A^-$$

$$B^+B^- \times B^+B^-$$

$$\begin{array}{cccccc} \frac{1}{4} A^+A^+ & \frac{1}{2} A^+A^- & \frac{1}{4} A^-A^- & \frac{1}{4} B^+B^+ & \frac{1}{2} B^+B^- & \frac{1}{4} B^-B^- \end{array}$$

Combine results

F₂ generation

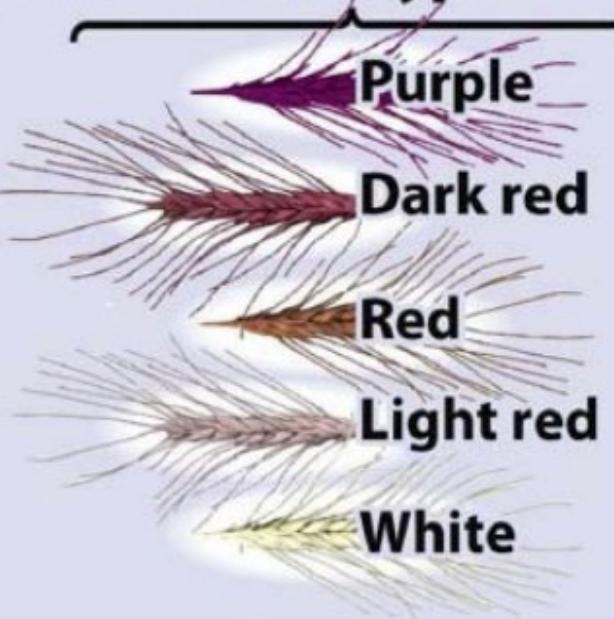
Number of pigment genes

Phenotype

| | | | | |
|----------------------|---|---|--------|-----------|
| $\frac{1}{4} B^+B^+$ | $\frac{1}{4} \times \frac{1}{4} = \frac{1}{16}$ | 4 | Purple | |
| $\frac{1}{4} A^+A^+$ | $\frac{1}{2} B^+B^-$ | $\frac{1}{4} \times \frac{1}{2} = \frac{2}{16}$ | 3 | Dark red |
| | $\frac{1}{4} B^-B^-$ | $\frac{1}{4} \times \frac{1}{4} = \frac{1}{16}$ | 2 | Red |
| $\frac{1}{2} A^+A^-$ | $\frac{1}{4} B^+B^+$ | $\frac{1}{2} \times \frac{1}{4} = \frac{2}{16}$ | 3 | Dark red |
| | $\frac{1}{2} B^+B^-$ | $\frac{1}{2} \times \frac{1}{2} = \frac{4}{16}$ | 2 | Red |
| | $\frac{1}{4} B^-B^-$ | $\frac{1}{2} \times \frac{1}{4} = \frac{2}{16}$ | 1 | Light red |
| $\frac{1}{4} A^-A^-$ | $\frac{1}{4} B^+B^+$ | $\frac{1}{4} \times \frac{1}{4} = \frac{1}{16}$ | 2 | Red |
| | $\frac{1}{2} B^+B^-$ | $\frac{1}{4} \times \frac{1}{2} = \frac{2}{16}$ | 1 | Light red |
| | $\frac{1}{4} B^-B^-$ | $\frac{1}{4} \times \frac{1}{4} = \frac{1}{16}$ | 0 | White |

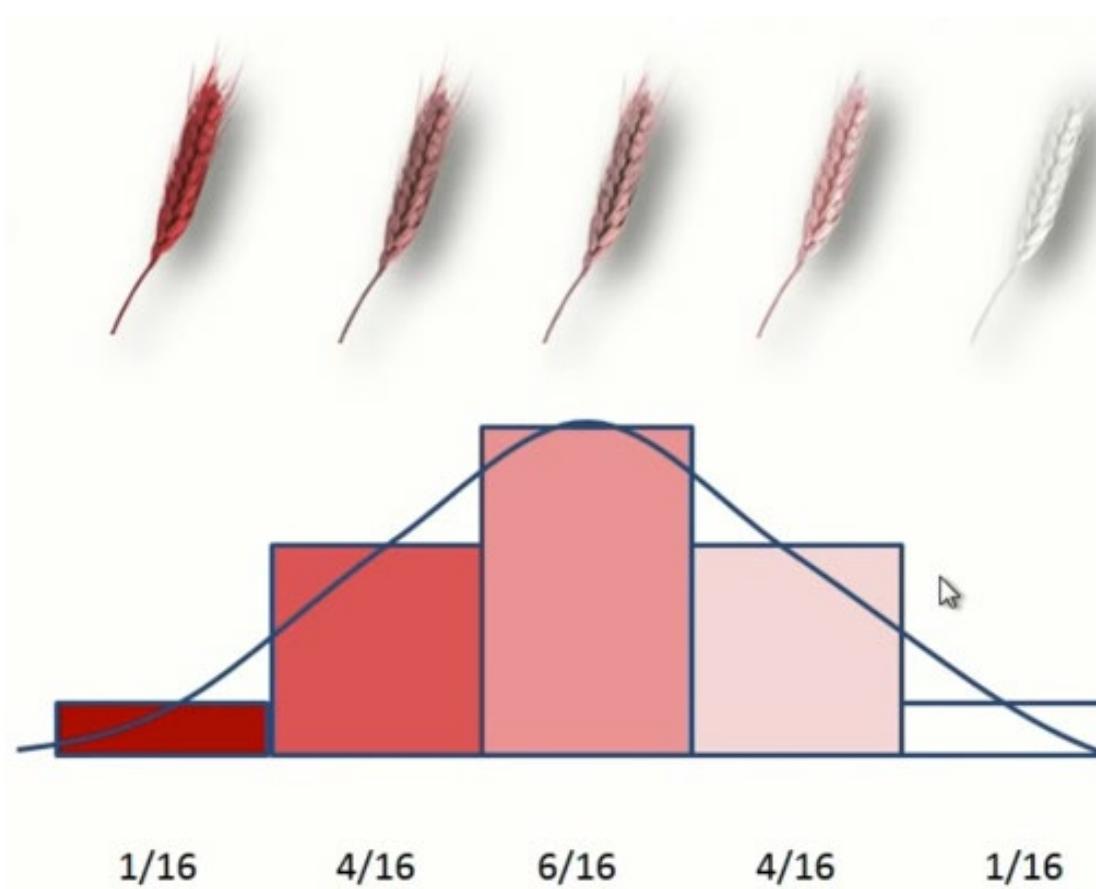
Nilsson-Ehle Experiment (1909)

| F₂ ratio | Number of Frequency pigment genes | Phenotype |
|----------------------------|--|------------------|
| $1/16$ | 4 | Purple |
| $4/16$ | 3 | Dark red |
| $6/16$ | 2 | Red |
| $4/16$ | 1 | Light red |
| $1/16$ | 0 | White |



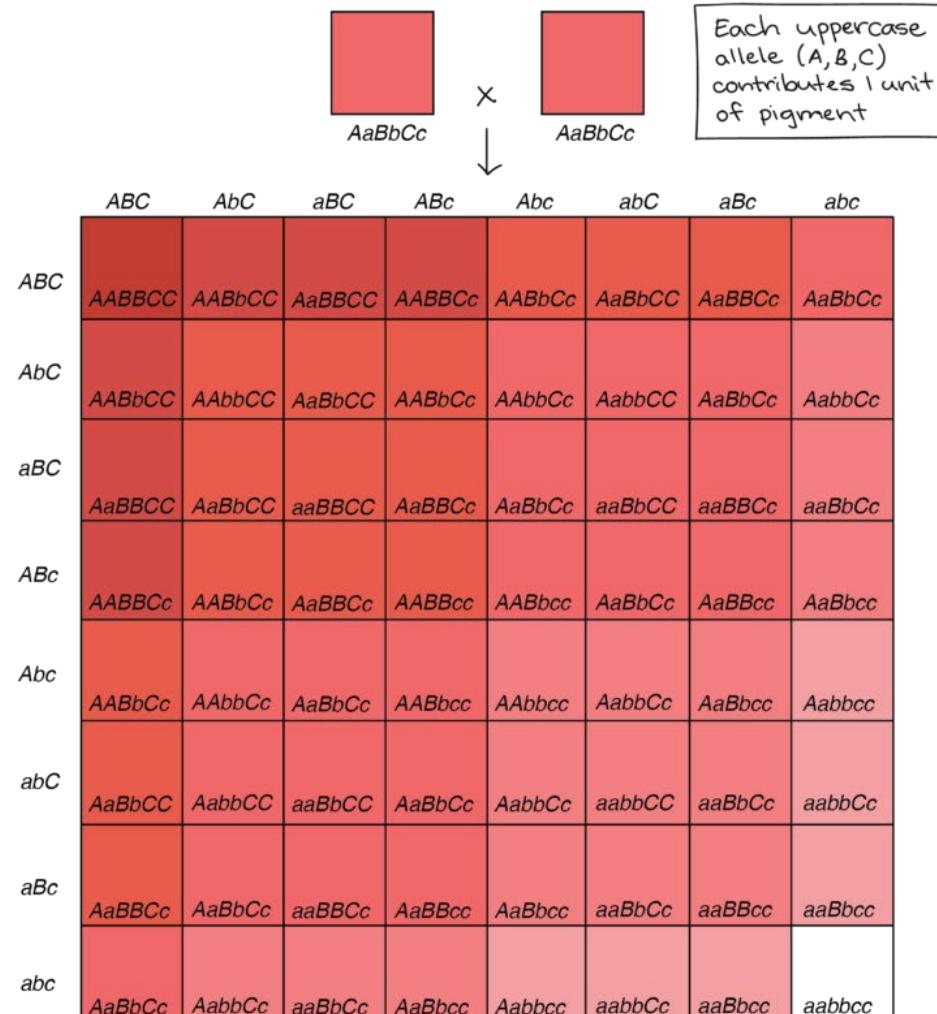
The illustration shows five wheat ears arranged vertically, each with its芒 (glumes) visible. The ears represent the different phenotypes based on the number of pigment genes:
1. Purple: The ear has the darkest, most intense purple color.
2. Dark red: The ear has a deep red color.
3. Red: The ear has a bright red color.
4. Light red: The ear has a pale, light red color.
5. White: The ear is completely white, with no visible red or purple pigmentation.

Nilsson-Ehle Experiment (1909)

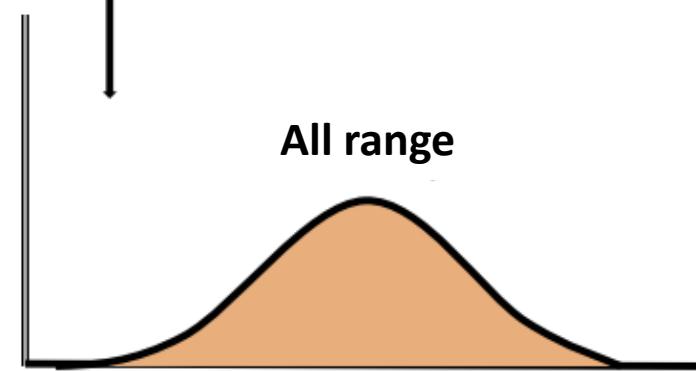
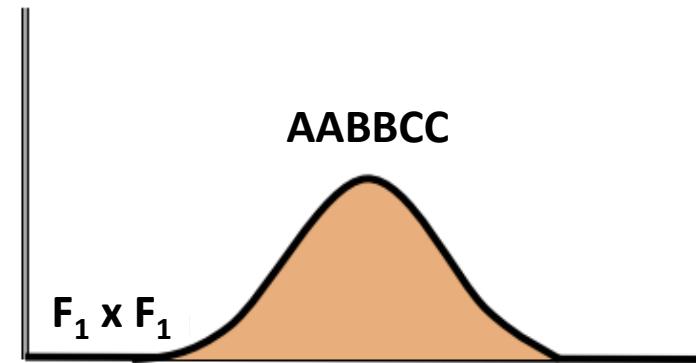
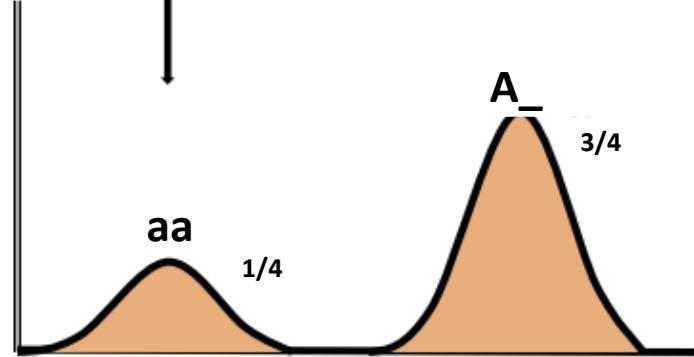
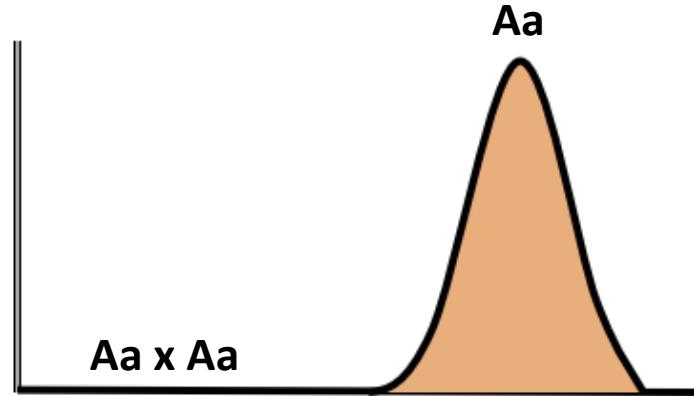
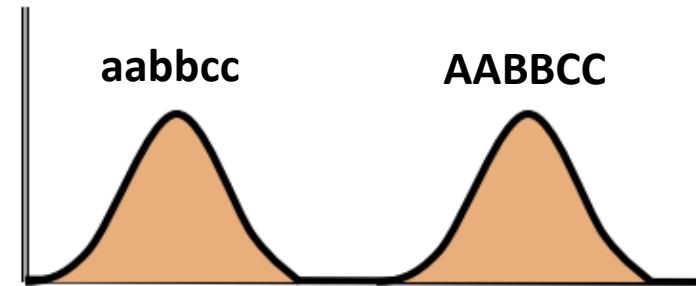
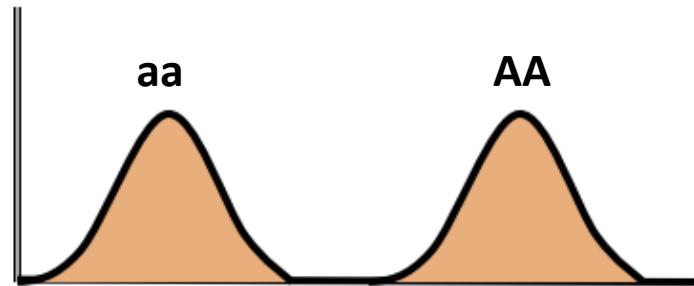


Additive Model:

- Trait due to several genes: polygenic.
- There are additive and non-additive alleles (**additive alleles** contribute to the expression of the trait; **non-additive alleles** don't).
- The contribution of additive alleles is similar and small.



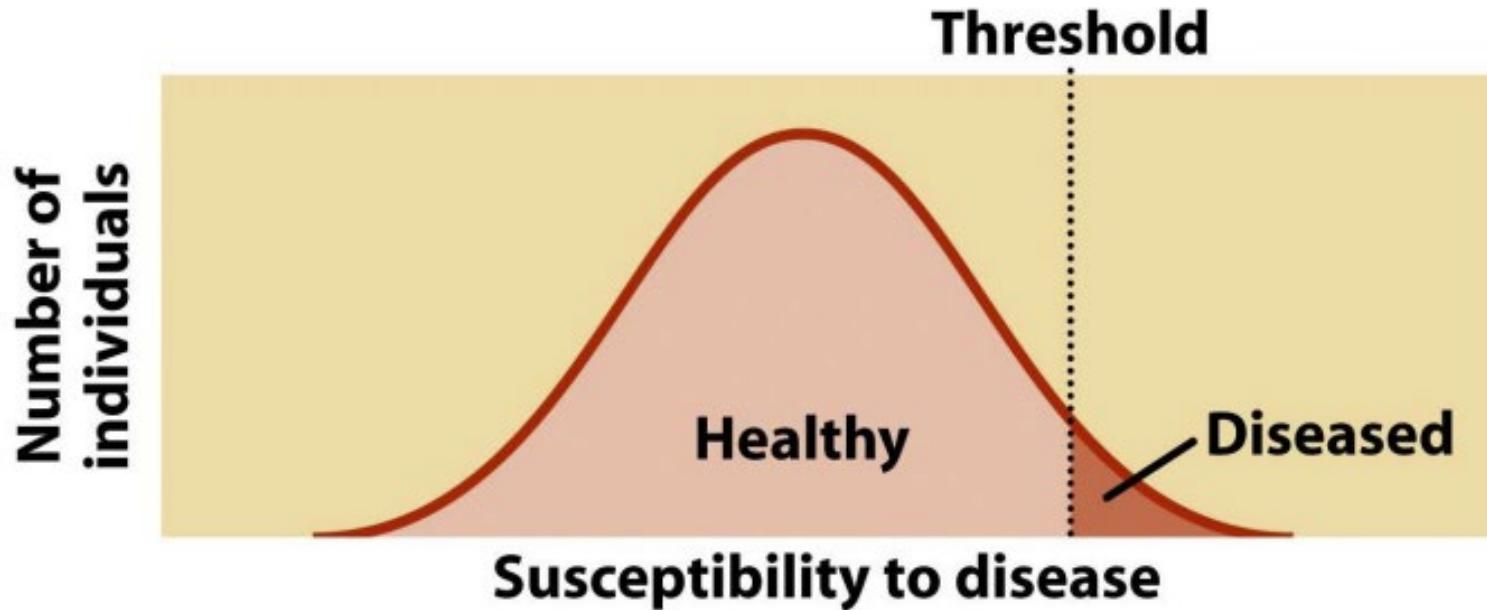
Qualitative vs Quantitative



Meristic Traits: quantitative traits depending on polygenes but organised in closed categories.



Threshold Traits: quantitative traits that are discretely expressed in a limited number of phenotypes (usually two).

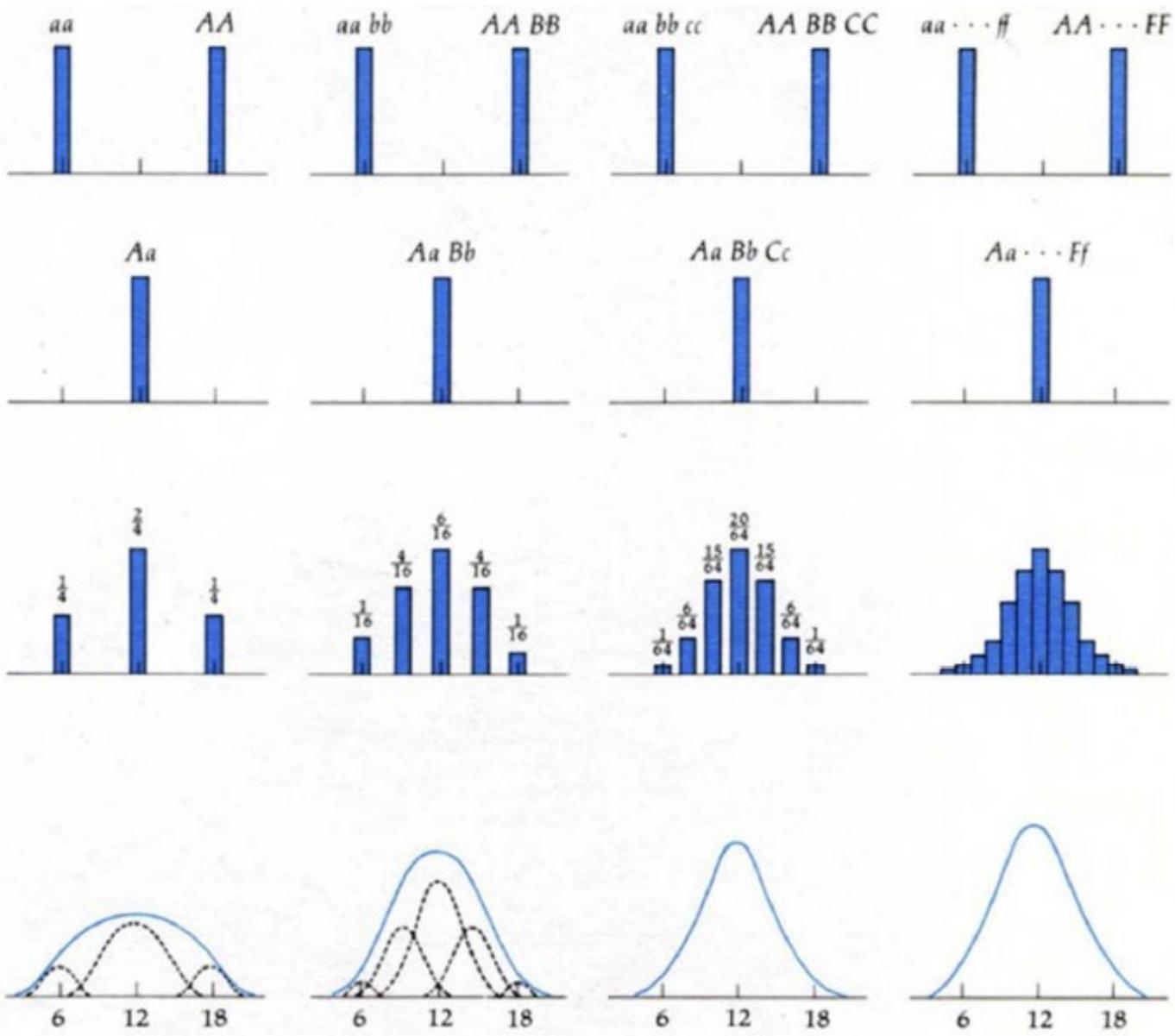


Questions of Quantitative Genetics

- **Can we make predictions** on how many individuals with a particular phenotype we might expect?
- **How many genes** are involved in a particular trait?
- What portion of observed variability is due to a **genetic differences** between individuals? What portion is due to **environmental factors**?
- Where are these genes **located in the genome**?

Questions of Quantitative Genetics

- **Can we make predictions** on how many individuals with a particular phenotype we might expect?



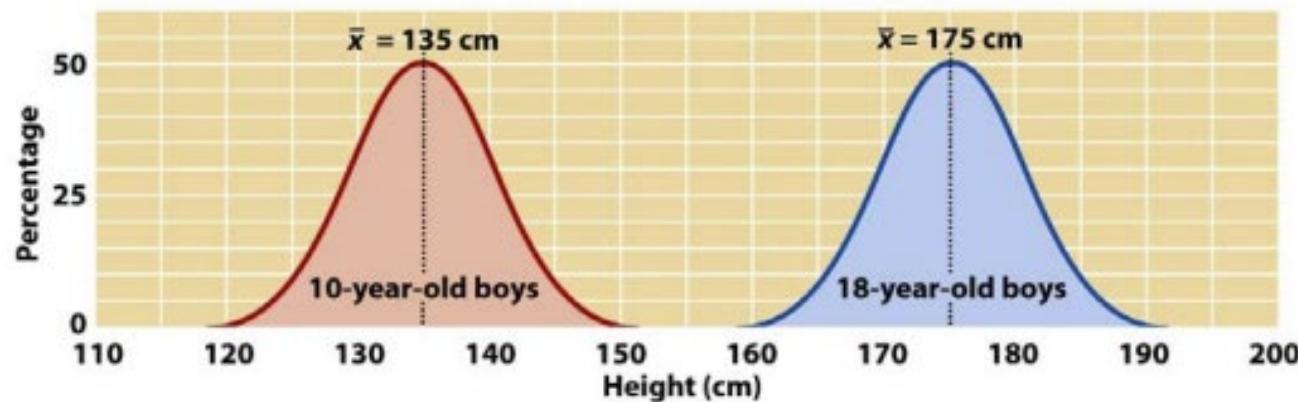


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Mean: sum of a collection of numbers divided by the count of numbers in the collection

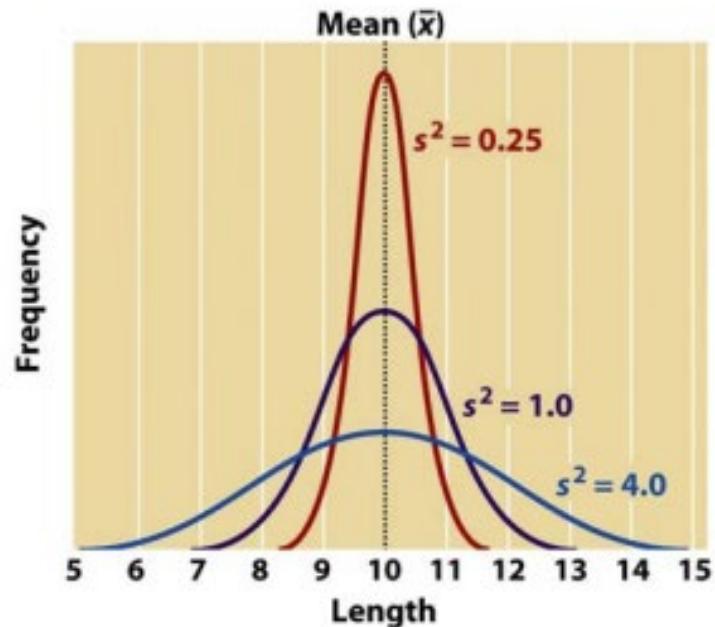


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Variance: measure of data dispersion (how far a set of numbers is spread out from their average value).

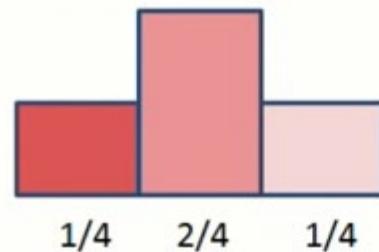
Number of gametes produced in the F₁ polyhybrid

| <i>1 locus</i> | <i>2 loci</i> | <i>3 loci</i> | <i>n loci</i> |
|-------------------|---|---|---------------|
| 2 (A,a) | 4 (AB, ab, Ab, aB) | 8 (ABC, ABC, Abc, abc, aBC, abC, AbC, aBc) | 2^n |
| 3 (AA, Aa, aa) | 9 (AABB, AaBB, aaBB, AABb, AaBb, Aabb, aaBB, AAbb, aabb) | 27 (AABBCC, ..., aaabbbcccc) | 3^n |
| 3 | 5 | 7 | $2n+1$ |
| 1/4 (AA/aa) | 1/16 (AABB/aabb) | 1/64 (AABBCC/aabbcc) | $1/4^n$ |

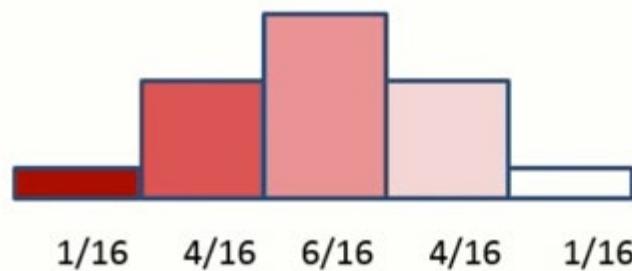
Number of phenotypes expected in F₂

Number of individuals with parental phenotype expected in F₂

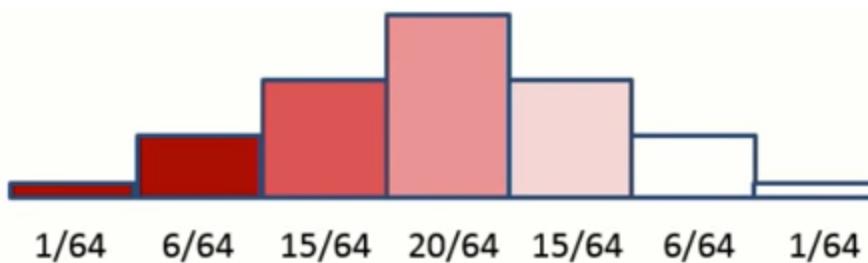
Calculating the number of genes involved in a quantitative trait



1 gene



2 genes



3 genes

AABBCCDDEE x aabbccdde

25 cm 15 cm



AaBbCcDdEe

20 cm

In a F_2 of 512, how many of them will have 19 cm?

AABBCCDDEE x aabbccdde

25 cm 15 cm



AaBbCcDdEe

20 cm

| NUMBER OF ADDITIVE ALLELES | PHENOTYPE |
|----------------------------|-----------|
| 0 | 15 |
| 1 | 16 |
| 2 | 17 |
| 3 | 18 |
| 4 | 19 |
| 5 | 20 |
| 6 | 21 |
| 7 | 22 |
| 8 | 23 |
| 9 | 24 |
| 10 | 25 |

AABBCCDDEE x aabbccdde

25 cm 15 cm



AaBbCcDdEe

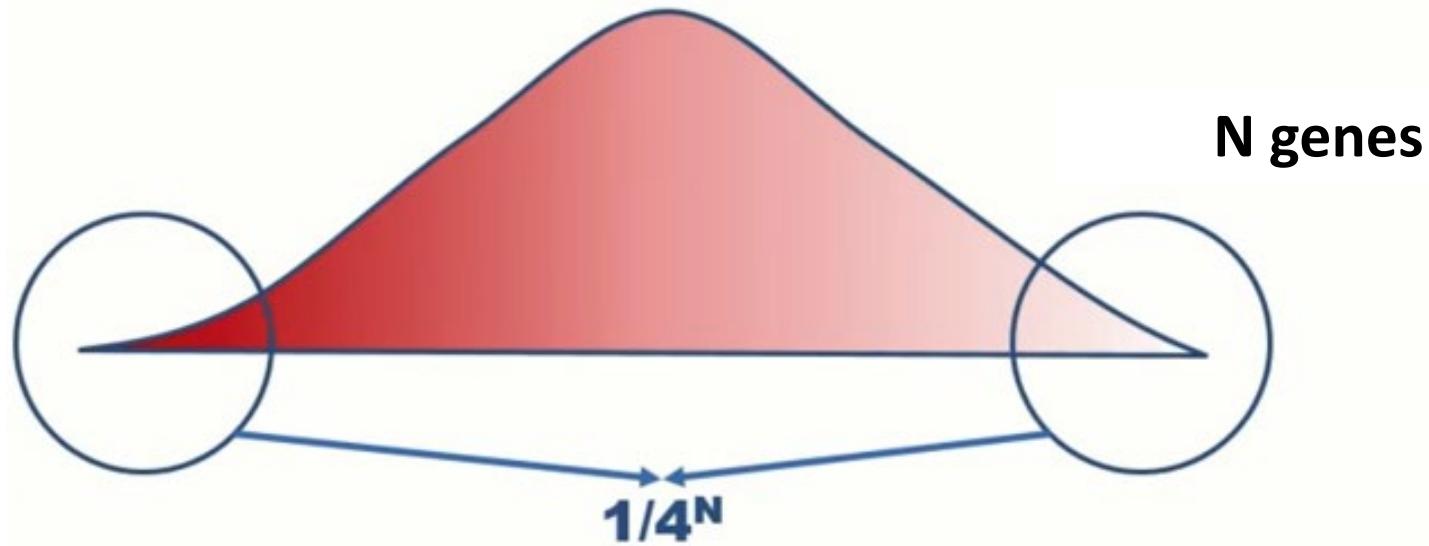
20 cm

$$\frac{10!}{4! 6!} \times \frac{(1/2)^4 \times (1/2)^8}{1024} = \frac{210}{1024} \times 512 = 105$$

Questions of Quantitative Genetics

- **How many genes** are involved in a particular trait?

Calculating the number of genes involved in a quantitative trait



$$p = 1/4^N$$

$$\rightarrow \text{Log } (p) = \log(1/4^N) \rightarrow \text{Log } (p) = N \log(1/4) \rightarrow$$
$$\rightarrow N = \text{Log } (p) / \log(1/4)$$

Calculating the number of genes involved in a quantitative trait

We have two purebreds plants producing fruits of 50g and 100g each.

F_1 is uniform and produces in average fruits of 75g.

F_2 is highly variable. 1 out of 1030 plants produces fruits of 50g.

$$\mathbf{N = Log (p) / log(1/4)} \rightarrow \mathbf{N = Log (1/1030) / log(1/4)} \rightarrow$$

↓

$$\rightarrow \mathbf{N = -3,013 / -0,602} \rightarrow \mathbf{N = 5,005}$$

Questions of Quantitative Genetics

- What portion of observed variability is due to a **genetic differences** between individuals? What portion is due to **environmental factors**?

GENOTYPE (G) + ENVIRONMENT (E) → PHENOTYPE (P)

Heritability: estimates the degree of variation in a phenotypic trait in a population that is due to genetic variation between individuals in that population.

GENOTYPE (G) + ENVIRONMENT (E) → PHENOTYPE (P)

$$\text{VAR}(P) = \text{VAR}(G) + \text{VAR}(E) + \text{VAR}(G/E)$$



Additive variance

Dominance variance

Gene-interaction variance

VAR(G/E): the expression of some genes relies on environmental factors.

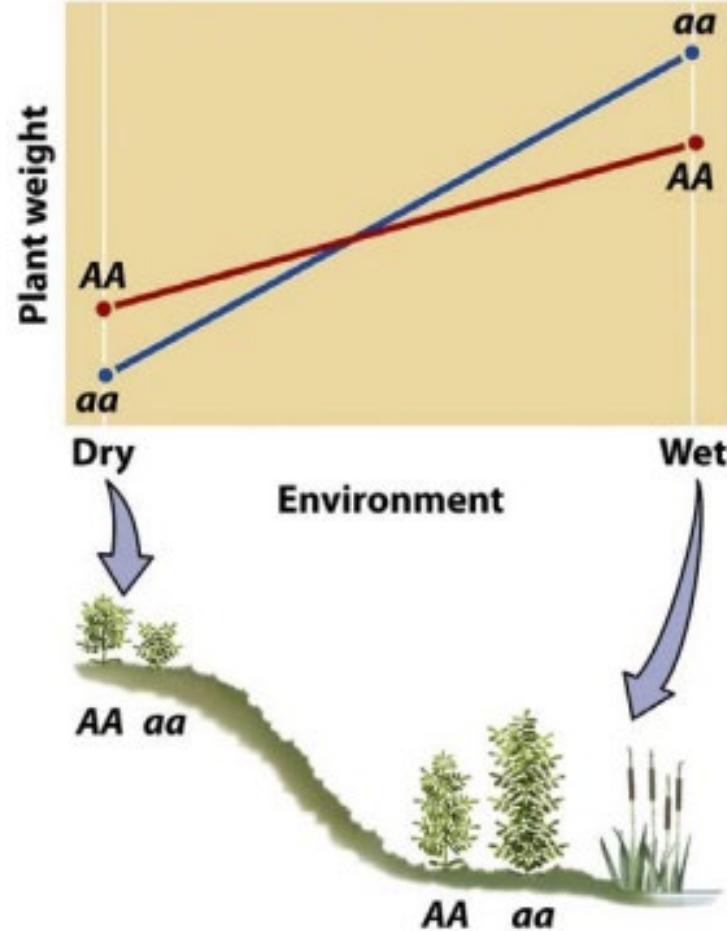


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Broad-sense heritability (H^2)

Reflects all the genetic contributions to a population's phenotypic variance including additive, dominant, and the one due to gene interactions.

$$H^2 = \frac{\text{Var}(G)}{\text{Var}(P)}$$

Calculating broad-sense heritability (H^2)

by elimination of some components of the equation.

$$\mathbf{VAR(P) = VAR(G) + VAR(E)}$$

If all individuals are bred in an identical and controlled environment

$$\mathbf{VAR(E) = 0}$$

$$\mathbf{VAR(P) = VAR(G)}$$

$$H^2 = \frac{\text{Var}(G)}{\text{Var}(P)}$$

Calculating broad-sense heritability (H^2)

by elimination of some components of the equation.

$$\mathbf{VAR(P) = VAR(G) + VAR(E)}$$

If all individuals are clones (genetically identical)

$$\mathbf{VAR(G) = 0}$$

$$\mathbf{VAR(P) = VAR(E)}$$

$$H^2 = \frac{\text{Var}(G)}{\text{Var}(P)}$$

Calculating broad-sense heritability (H^2)

by elimination of some components of the equation.

Experiment 1

We cultivate a type of plant and observe the size of the fruits. All variance will be considered VAR(P)

$$\text{VAR}(P) = \text{VAR}(G) + \text{VAR}(E) = 573$$

Experiment 2

We reproduce an individual asexually and observe the size of the fruits. All variance will be considered VAR(E)

$$\text{VAR}(P) = \text{VAR}(E) = 340$$

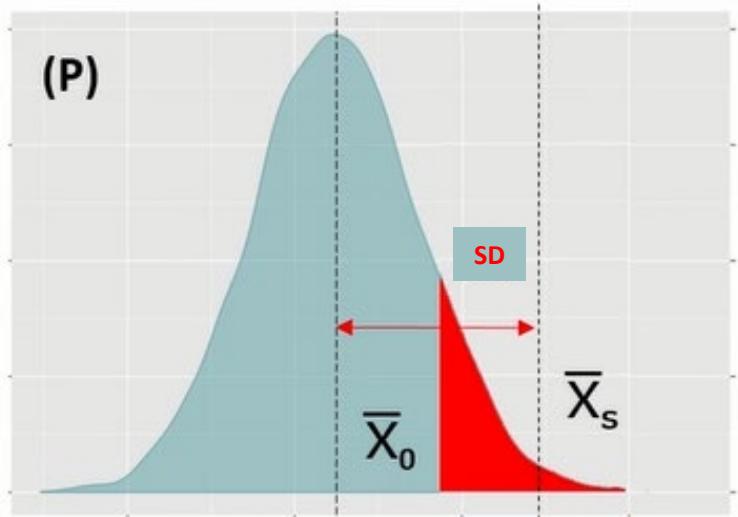
$$\text{VAR}(G) = \text{VAR}(P) - \text{VAR}(E) = 573 - 340 = 233$$

$$H^2 = \frac{\text{Var}(G)}{\text{Var}(P)} = \frac{233}{573} = 0.41$$

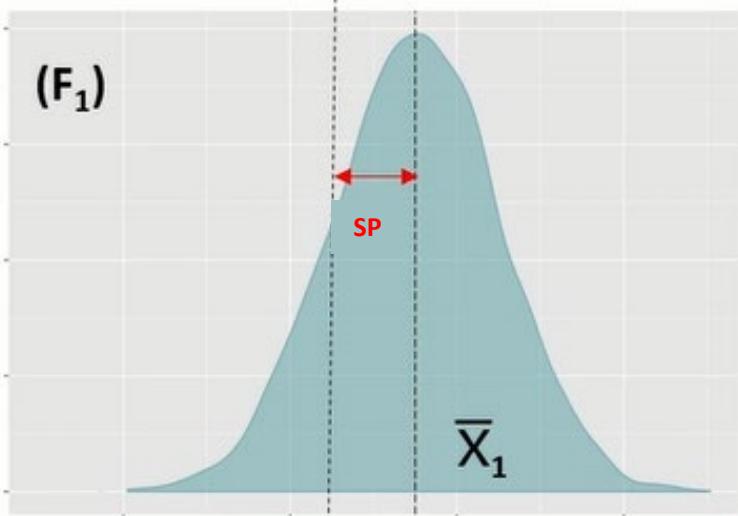
Narrow-sense heritability (h^2)

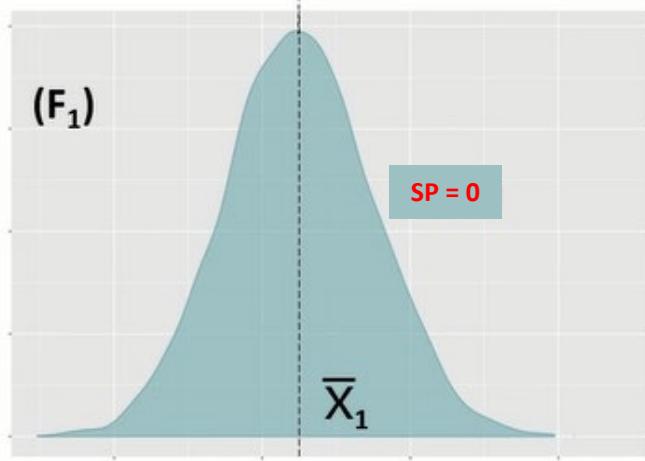
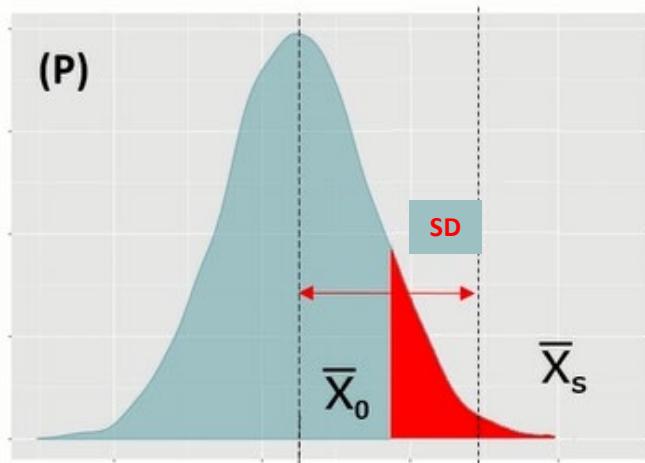
Reflects the genetic contribution of additive alleles to a population's phenotypic variance.

$$h^2 = \frac{\text{Var}(A)}{\text{Var}(P)}$$



$$h^2 = \frac{\text{selection progress}}{\text{selection differential}} = \frac{|x_1 - x_0|}{|x_s - x_0|}$$

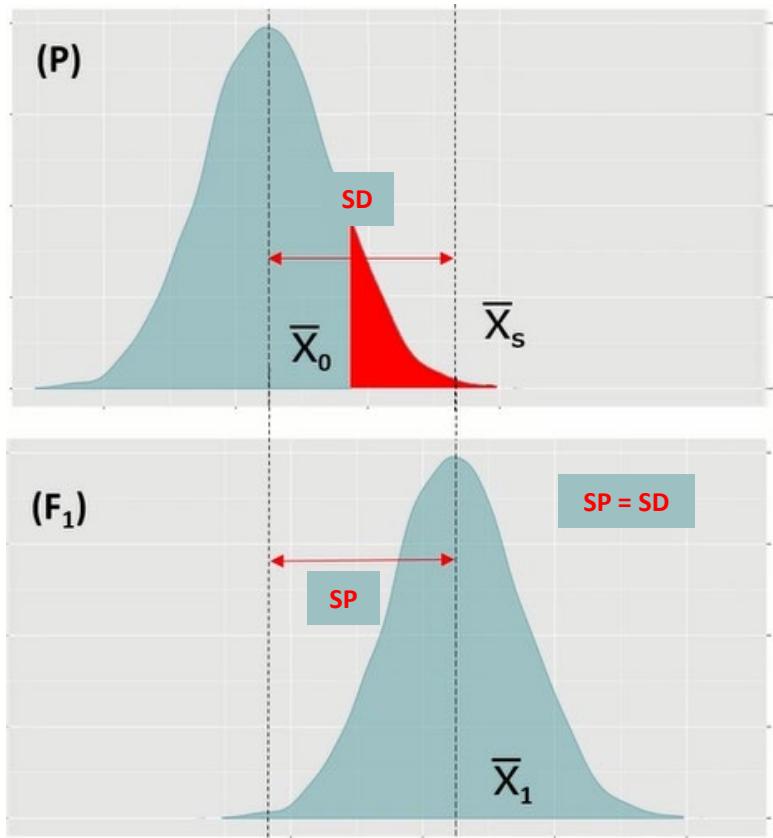




$$h^2 = \frac{\text{selection progress}}{\text{selection differential}} = \frac{|x_1 - x_0|}{|x_s - x_0|}$$

$$\frac{\bar{X}_1 - \bar{X}_0}{\bar{X}_s - \bar{X}_0} = 0$$

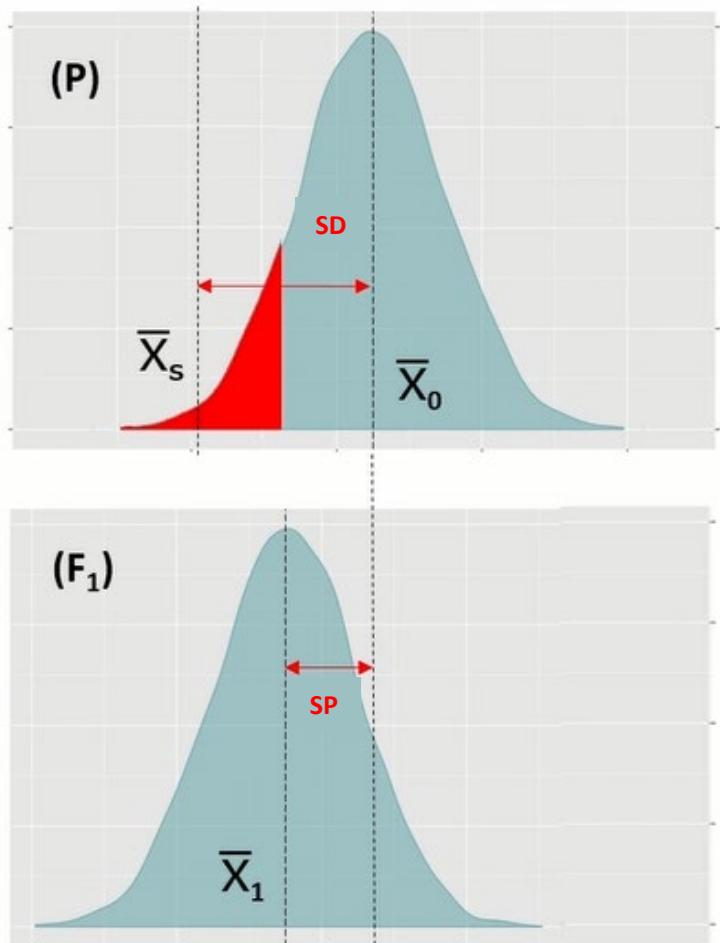
$(\bar{X}_1 = \bar{X}_0)$



$$h^2 = \frac{\text{selection progress}}{\text{selection differential}} = \frac{|x_1 - x_0|}{|x_s - x_0|}$$

$$(\bar{X}_1 = \bar{X}_S)$$

$$\frac{\bar{X}_1 - \bar{X}_0}{\bar{X}_S - \bar{X}_0} = 1$$



$$h^2 = \frac{\text{selection progress}}{\text{selection differential}} = \frac{|x_1 - x_0|}{|x_s - x_0|}$$

$(\bar{X}_1 < \bar{X}_0)$

$$\frac{\bar{X}_1 - \bar{X}_0}{\bar{X}_s - \bar{X}_0}$$

$(\bar{X}_s < \bar{X}_0)$

Heritability

$0 < h^2 > 0.25$

Low

$0.25 < h^2 > 0.5$

Medium

$0.5 < h^2 > 1$

High

In traits with **high heritability**, additive genetic component is predominant. They are easily improved.

In traits with **low heritability**, non-genetic components are more important in the expression of the character (environment). They are not easily improved.

Genetic Improvement

Can we improve a trait indefinitely?

- After many generations of selection, selection response stabilizes and the trait do not longer respond to selection.
- Genetic variability decreases in the population. Additive alleles get fixed. Heritability tends to 0.

Limitations of the heritability

- It is not individual but it refers to a specific population.
- There is no universal value of heritability for a particular trait.
- Even though heritability is high, influence of environment can still be important.

Questions of Quantitative Genetics

- Where are these genes **located in the genome?**

QTLs (Quantitative Trait Loci)

Chromosome regions involved in the expression of a polygenic trait.

If we find a molecular marker systematically inherited with a trait, this is most probably associated to a QTL.

Table 24.2 Quantitative characteristics for which QTLs have been detected

| Organism | Quantitative Characteristic | Number of QTLs Detected |
|-------------|-----------------------------|-------------------------|
| Tomato | Soluble solids | 7 |
| | Fruit mass | 13 |
| | Fruit pH | 9 |
| | Growth | 5 |
| | Leaflet shape | 9 |
| | Height | 9 |
| Corn | Height | 11 |
| | Leaf length | 7 |
| | Tiller number | 1 |
| | Glume hardness | 5 |
| | Grain yield | 18 |
| | Number of ears | 9 |
| | Thermotolerance | 6 |
| Common bean | Number of nodules | 4 |
| Mung bean | Seed weight | 4 |
| Cow pea | Seed weight | 2 |
| Wheat | Preharvest sprout | 4 |
| Pig | Growth | 2 |
| | Length of small intestine | 1 |
| | Average back fat | 1 |
| | Abdominal fat | 1 |
| Mouse | Epilepsy | 2 |
| Rat | Hypertension | 2 |

