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CONCEPT: MATHMATICAL MEASRUMENTS

● Common statistical measurements are used in genetics to ______________________ phenotypes

□ The **mean** is an average of values

- A **population** is all individuals within the group you're measuring
- A **sample** is a representative subset of individuals in a population

EXAMPLE: Mean calculation

$$M = \frac{\Sigma(X)}{N}$$

Where Σ = Sum of
X = Individual data points
N = Sample size (number of data points)

□ The **variance** measures how far a set of values is from the mean

- **Covariance** measures how much variation is common to 2+ traits

EXAMPLE: Variance calculation

$$S^2 = \frac{\Sigma(X-M)^2}{n - 1}$$

Where Σ = Sum of
X = Individual score
M = Mean of all scores
N = Sample size (number of scores)

□ The **standard deviation** measures the amount of variation that exists within a set of data

- **Standard error** measures the accuracy of the sample mean

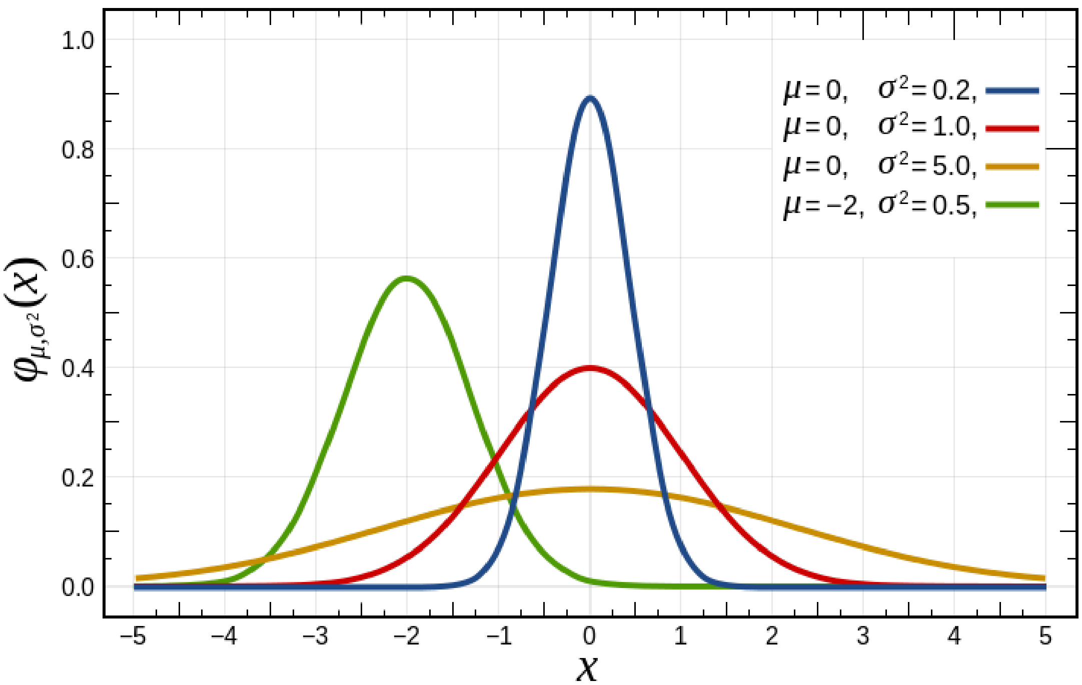
EXAMPLE: Standard deviation calculation

$$s = \sqrt{\frac{\sum(x-\bar{x})^2}{n-1}}$$

□ A **normal distribution** is the "bell curve" and visualizes the range of variation of a phenotype

- Sometimes called a "frequency histogram" as it measures frequency of the trait on y axis

EXAMPLE:



PRACTICE:

Bristle Number	Number of Individuals
1	2,
2	3,
3	9
4	29,
5	55,
6	18,
7	4,

1. The table shows a distribution of bristle numbers in a *Drosophila* population. What is the mean bristle number?
   a. 4.7
   b. 80
   c. 562
   d. 5.0

Bristle Number	Number of Individuals	$(X\text{-}M)^2$	Sum $(X\text{-}M)^2$
1	2	13.69	27.38
2	3	7.29	21.87
3	9	2.89	26.01
4	29	0.49	14.21
5	55	0.09	4.95
6	18	1.69	30.42
7	4	5.29	21.16

2. The table shows a distribution of bristle numbers in a *Drosophila* population. What is the variance?
   a. 1.0
   b. 1.2
   c. 5.5
   d. 3.0

3. Using the variance calculated in problem #2, what is the standard deviation?
    a. 1.0
    b. 1.1
    c. 2.3
    d. 1.7

CONCEPT: TRAITS AND VARIANCE

● There are many different types of inherited ______________________________

□ **Continuous traits** can take a potentially infinite number of states within a range (Ex: height)

□ **Categorical traits** are traits that can be sorted into discrete categories (Ex: purple or white flowers)

- **Threshold traits** are expressed when people reach a threshold of genetic and environmental factors

  - Ex: Type 2 diabetes

- **Meristic traits (counting traits)** are traits that can be divided into a range of discrete values

  - Ex: Birds can lay 1, 2, 3, or 4 eggs, but cannot lay 2.58 eggs

EXAMPLE:

Example	**Trait Type**
Number of spots on a Dalmatian	
Human weight	
Foot size	
Cat Litter Sizes	

● Traits can be inherited in _________________________ ways

□ **Complex inheritance** is inheritance involving multiple genes and environmental factors

□ **Simple inheritance** is observed when progeny have standard Mendelian ratios (3:1, or 9:3:3:1)

● Most traits for complex organisms are controlled via polygenic inheritance (_________________________ inheritance)

□ Many genes are each behaving in a Mendelian fashion, and contribute to phenotype

□ Two types of alleles exist in polygenic inheritance

- **Additive allele** is an allele that contributes and is added to the phenotype

- **Non-additive allele** is an allele that does not contribute to the phenotype

EXAMPLE:

P — Plants with white kernals (aabb) x Plants with purple kernals (AABB)

↓

F_1 — Plants with red kernals (AaBb)

↓

F_2 —
1/16 plants with purple kernals (AABB)
4/16 plants with dark-red kernals (AaBB or AABb)
6/16 plants with red kernals (AaBb)
4/16 plants with light-red kernals (Aabb or aaBb)
1/16 plants with white kernals (aabb)

- There is a formula to predict how many genes ___________________________ to a trait
  - □ The formula $(1/4)^n$ calculates the F2 ratio of individuals expressing the parental phenotype (grandparents)
    - \- n = number of polygenes involved
  - □ The formula 2n+1 calculates the number of phenotypic categories observed

PRACTICE:

1. A trait controlled through polygenic inheritance was observed in a series of experiments. A brown eyed rabbit was mated with a blue eyed rabbit. 130 F_2 offspring were produced. 2 offspring had brown eyes and 2 offspring had blue eyes. How many polygenes control eye color in rabbits?
    a. 1
    b. 2
    c. 3
    d. 4

2. If a trait is controlled by 5 polygenes, how many phenotypic categories will be observed in the F_2 generation?
    a. 2
    b. 5
    c. 10
    d. 11

3. Polygenic inheritance is what type of inheritance?
    a. Simple
    b. Complex
    c. Additive
    d. Non-additive

CONCEPT: ANALYZING TRAIT VARIATION

● Trait variation can be caused by genetic and environmental ___________________________

□ The formula used to calculate phenotypic variation is: $V_P = V_G + V_E$

- Phenotypic variance = V_P
- Genetic variance = V_G
- Environmental variance = V_E

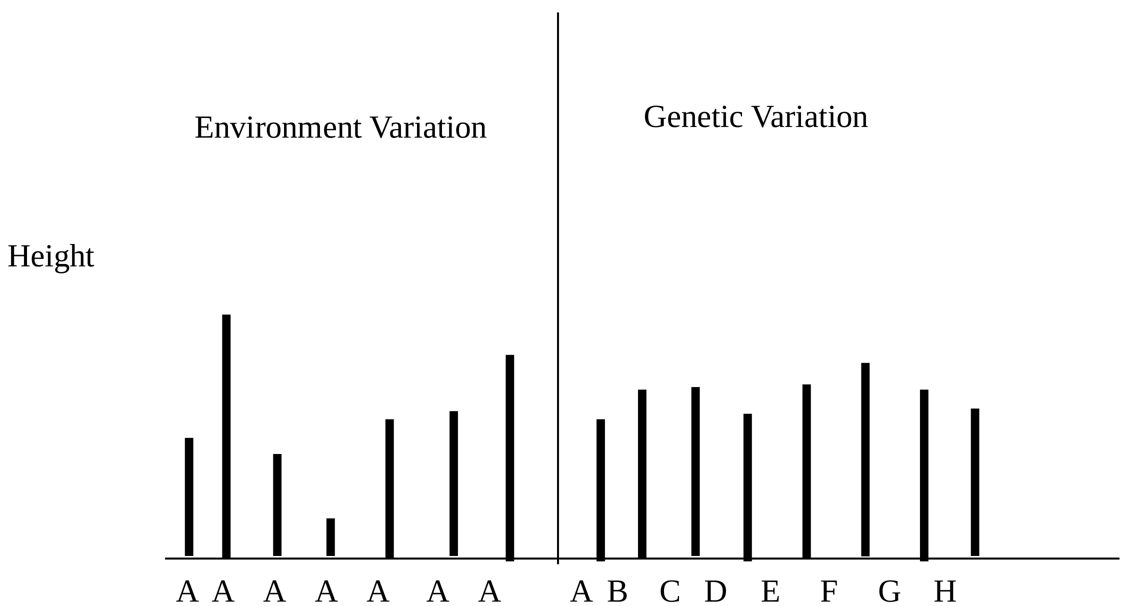
□ To determine the variation attributed to genetics you must control for _______________________

- If you are looking for how much genetic variation contributes to stem height in one species of flowers then:
  - Plant multiple seeds from one species in a carefully controlled greenhouse ($V_E = 0$)

□ To determine the variation attributed to environment you must control for genetics

- How much environmental variation contributes to stem height in one species of flowers?
  - Plant multiple genetically identical seeds in many different environmental conditions ($V_G = 0$)

EXAMPLE:



PRACTICE:

1. Which of the following represent trait variation caused from genetic variation?
    a. V_P
    b. V_G
    c. V_E
    d. V_V

2. If you wanted to identify what proportion of trait variation is due to the environment, you would do what?
    a. Control for environmental variation
    b. Control for overall variation
    c. Control for genetic variation
    d. Control of phenotypic variation

3. If you wanted to identify what proportion of trait variation is due to genetics, you would do what?
    a. Control for environmental variation
    b. Control for overall variation
    c. Control for genetic variation
    d. Control of phenotypic variation

CONCEPT: HERITABILITY

● **Heritability** is the proportion of variation in a population that's due to genetic factors

□ It is a very ___________________ measurement that is only true for a certain population in a certain environment

- It measures from 0 to 1, and the larger the value, the more variation is explained by genetic differences
- Ex: h=0.65 means 65% of the overall population variation is explained by genetic differences in individuals

□ **Broad-sense heritability** measures the contribution of genotypic variance to total phenotypic variance

- $H^2 = V_G/V_P$
- A H^2 close to 1 = environmental conditions had little impact on variation
- A H^2 close to 0 = environmental conditions had a major impact on variation

EXAMPLE: Calculate broad sense heritability for each trait

Trait	V_P	V_G	V_A
Body Fat	40.5	16.9	7.66
Body Length	43.6	17.9	5.12

□ **Narrow-sense heritability** measures the proportion of phenotypic variation due to additive genotypic variance

- **Additive variation (V_A)** is genetic variance caused by average differences between allelic characteristics
    - Dominant and recessive alleles have different characteristics
- **Dominance variance (V_D)** is gene variance from heterozygotes not being intermediates of homozygotes
    - Heterozygotes are different than an intermediate between dominant and recessive homozygotes
- The ________________________________ to know are:
    - $h_2 = V_A/V_P$
    - $V_G = V_A + V_D$

EXAMPLE: Calculate narrow-sense heritability for each trait

Trait	**V_P**	**V_G**	**V_A**
Body Fat	40.5	16.9	7.66
Body Length	43.6	21.7	5.12

Artificial Selection

● **Artificial selection** is the process of choosing specific individuals for phenotypic breeding purposes

□ Breeders use narrow-sense heritability to predict the impact of ____________________________

- The higher the h_2 value the more likely the breeder will observe a change in offspring

□ h_2 = R/S

- R = Mean of the offspring – overall mean – called **selection response**
- S = Mean of the parents – overall mean - called **selection differential**

EXAMPLE: Which of the following traits will respond best to selection by a breeder?

Trait	V_P	V_G	V_A
Body Fat	40.5	16.9	7.66
Body Length	43.6	21.7	5.12

Twin Studies

● Humans cannot be bred to determine heritability, so __________________ studies are used

□ **Monozygotic twins** arise from a single zygote that mitotically divides and splits into two cells

- Have same genetics, and therefore only exhibit environmental variation
- But some genetic changes can occur in early development (Ex: copy-number variations)

□ **Dizygotic twins** are from two separate fertilization events

- Are genetically as close as any other sibling set, but often share similar environment

□ Twin expression of a trait can be classified in two ways

- **Concordant** is when both or neither twins express a trait
- **Discordant** is when one twin expresses a trait but not the other

PRACTICE:

1. A chicken breeder has a population of chickens where the average number of eggs laid per hen per month is 34. The narrow-sense heritability is 0.75. With this information is it likely that a breeder could select for an increase in eggs per hen laid each month?
   a. No, breeders never know whether they can select for a trait
   b. No, the breeder will need to know the broad-sense heritability to determine whether selection could cause an increase in eggs?
   c. Yes, because the narrow-sense heritability is 0.75, this means selection is likely to occur

2. The narrow-sense heritability of the number of seeds per flower is 0.9. The mean of the population is 6.0 seeds per flower. A flower breeder crosses one flower with 7 seeds to another plant with 9 seeds. What is the expected number of seeds per flower in the offspring of this cross?
   a. 5
   b. 6
   c. 7
   d. 8

3. Heritability calculations were calculated for a variety of different traits. Which of the following traits would respond best to selection?
   a. $H_2 = 0.8$, $h_2 = 0.3$
   b. $H_2 = 0.3$, $h_2 = 0.3$
   c. $H_2 = 0.9$, $h_2 = 0.8$
   d. $H_2 = 0.5$, $h_2 = 0.9$

CONCEPT: QTL MAPPING

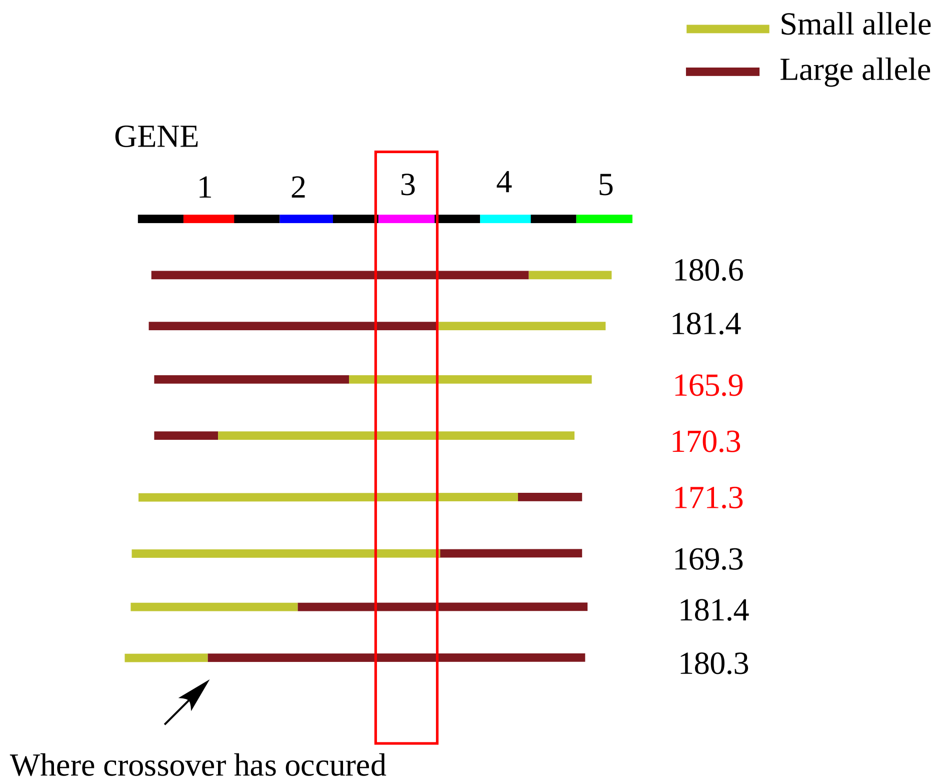
- **Quantitative trait loci (QTL)** are locations of genes that control variation in complex (quantitative) ______________
  - □ **Quantitative traits** are any traits that can be measured (usually continuous)
  - □ **QTL Mapping** is the method for determining QTLs in the genome
  - □ The method of QTL ___________________ is as follows:
    1. Mate two inbred lines with different traits (Ex: Tomato weight of 230g x tomato weight of 10g)
       - Produces intermediate F_1 generation
    2. Backcross F_1 to the large tomato parents (230g)
       - Produces **back-cross 1** generation (BC_1)
    3. Take DNA samples and determine genotype of BC_1 and Parental strains
       - Divide the genome into SNP markers
    4. Calculate weight for each BC_1 tomato
       - Calculate mean for all BC tomatoes
       - Calculate mean for all BC tomatoes with the same markers
    5. Determine if QTL is affecting fruit weight
       - If no QTL is affecting fruit weight then the overall mean will equal the "marker mean"
       - If QTL is affecting fruit weight then overall mean will not equal the "marker mean"
    6. Use **lod scores** to statistically confirm your hypothesis

EXAMPLE: Example data from QTL Mapping

Plant	Fruit Weight	Marker 1	Marker 2	Marker 3	Marker 4
Overall mean weight	176.3	-	-	-	-
Weight of L/L		176.5	178.6	182.1	175.9
Weight of L/S		174.5	173.4	168.4	172.3

- Once the QTL is identified, the gene that causes the variation will need to be ________________________
    - □ There can be 100+ genes in between two genomic markers used for sequencing
    - □ **Fine-mapping** is the method used to determine the gene from the QTL
        - □ Use **congenic stocks (nearly-isogenic):** are identical, but contain crossovers near QTLs

EXAMPLE:



QTL Mapping in Random-Mating Populations

● **Association mapping** can identify QTLs in genomes based on *linkage disequilibrium* between marker and QTL

□ **Linkage disequilibrium** is the nonrandom association of alleles at two loci (so alleles are not independent)

□ This method can be done in ___________________, as it tests many alleles at once & does not need crosses

- It also does not require fine-mapping as it directly identifies the responsible gene at the QTL

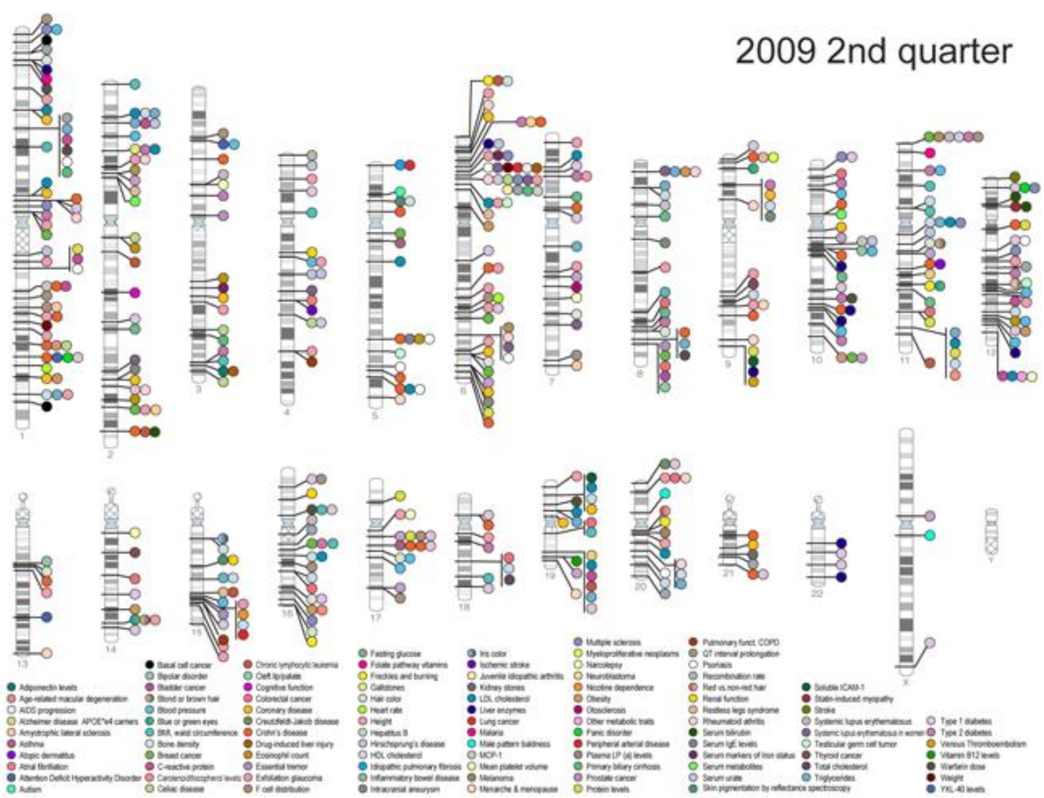
□ The method of mapping using **genome-wide association studies** is as follows:

1. Sequence genome of 2000 individuals with a disease and 2000 without a disease

   - Identify all SNPs in the genomes (HUGE amount of data)

2. Statisticians determine if one SNP is more frequently associated with disease than other

EXAMPLE:



PRACTICE:

1. Both QTL mapping and association (GWA) mapping are used to locate genes responsible for a phenotype. Which of the two techniques does NOT require crosses to produce a mapping population
    a. QTL mapping
    b. Association mapping

2. Both QTL mapping and association mapping are used to locate genes responsible for a phenotype. Which of the following typically tests two differing alleles between the parents of a mapping population?
    a. QTL mapping
    b. Association mapping

3. True or False: Association (GWA) mapping definitively proves that the gene identified is responsible for the trait variation or phenotype?
    a. True
    b. False