TWO TYPES OF TRAITS

<u>Discrete or discontinuous traits:</u> traits occur in distinct Categories: Trait is there or it is not (examples: albinism, cystic fibrosis, Huntington's disease) <u>Mendelian inheritance</u>, single genes, dominance, recessiveness

Continuous traits: Distribution of phenotypes in the population varies along a continuum. Individuals differ by small degrees. (examples include height, blood pressure, reaction time, learning ability) Polygenic quantitative or multifactorial inheritance. Genes act additively.

INHERITANCE OF CONTINUOUS TRAITS

A simple case with two loci, two alleles each

A

 A^1

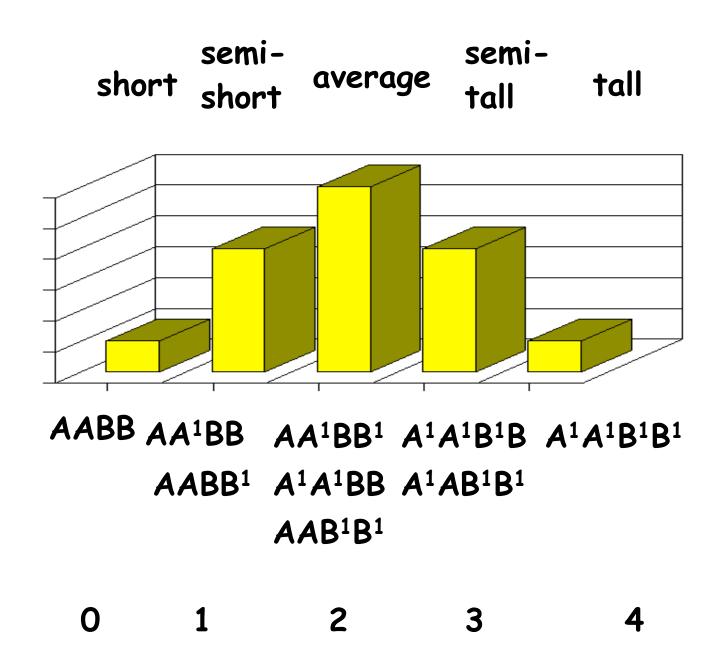
B

B¹

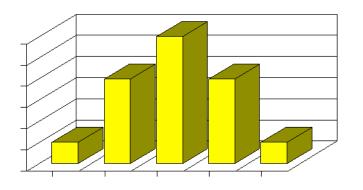
An allele with a prime ¹, adds one unit of measure to the phenotype

If a trait, say height, is controlled by two loci, A and B, and each locus has two alleles, one regular and one prime allele, what are the possible genotypes and phenotypes?

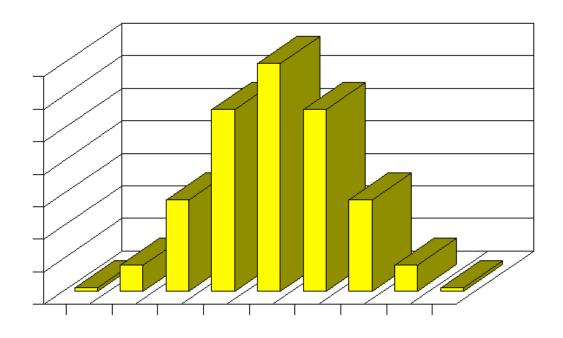
AABB	0short
AA ¹ BB	1semi-short
AABB ¹	1semi-short
AA ¹ BB ¹	2average
A^1A^1BB	2average
AAB ¹ B ¹	2average
$A^1A^1B^1B$	3semi-tall
$A^1AB^1B^1$	3semi-tall
$A^1A^1B^1B^1$	4tall



Two loci, Two alleles



Three loci, Two alleles



Characteristics of polygenic inheritance:

- 1. A substitution at one locus usually produces the same effect on the phenotype as a substitution at another.
- 2. Many loci with small, additive effects.
- 3. Tall parents can produce a short child, etc
- 4. Average parents can produce a tall or short child

Why are the traits continuous? Because of the environment.

In height, for example, nutritional differences can play a major role in variation

For skin color, exposure to sun can modify the phenotype

How do we know how much of the variability we see among people is due to genetic differences between them as opposed to environmental differences?

What are the components of variability in a group of people?

$$Vt = Ve + Vg$$

Vt = total variability

Ve = environmental variability

Vg = genotypic variability

Heritability: the proportion of the variability among Individuals in a population that is due to genotypic differences among them or Vg/Vt

Heritability can range from 0.0 to 1.0

If H = 0.0 it means that all of the variability observed in a trait in a given population is due to environmental differences experienced by the individuals

If H = 1.0 it means that all of the differences are due to genetic differences among the people

For most continuous traits, heritability is somewhere in between.

TWIN STUDIES TO MEASURE HERITABILITY

Dizygotic (DZ) twins: two egg twins, same degree of genetic relatedness as normal siblings (25% genes in common)

Monozygotic (MZ) twins: one egg twins genetically identical (100% genes in common)

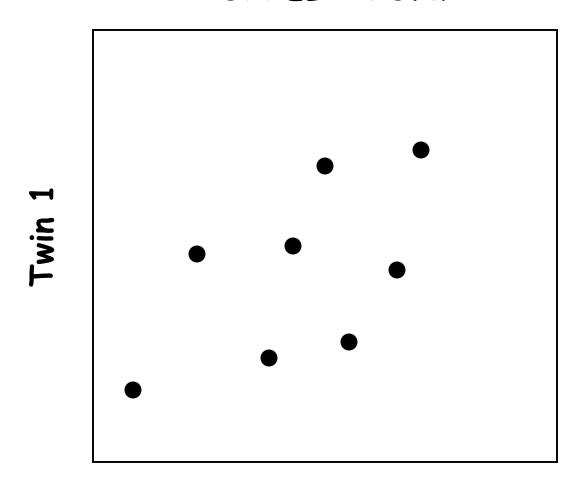
Twin Studies in Humans

- Monozygotic Twins:
 - phenotypic variance should be due to environmental variance
- Dizygotic Twins:
 - genetically different individuals who share similar environment
- Concordant traits vs. Discordant traits

Concordant: have the same trait

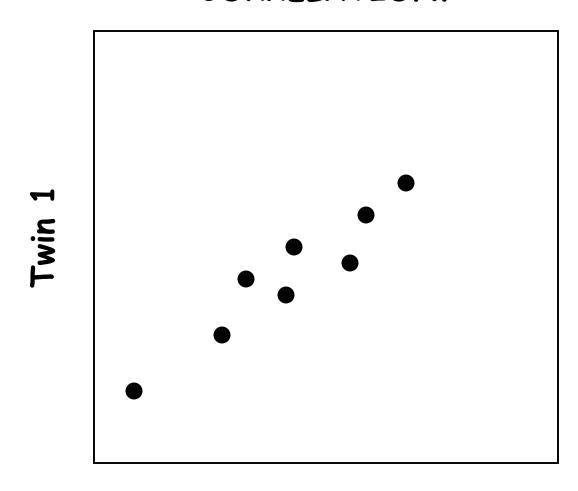
Discordant: do not have the same trait

CORRELATION?



Twin 1
DIZYGOTIC TWINS

CORRELATION?

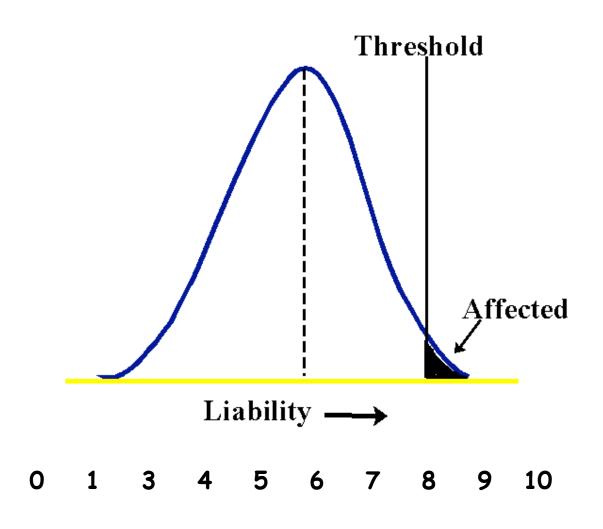


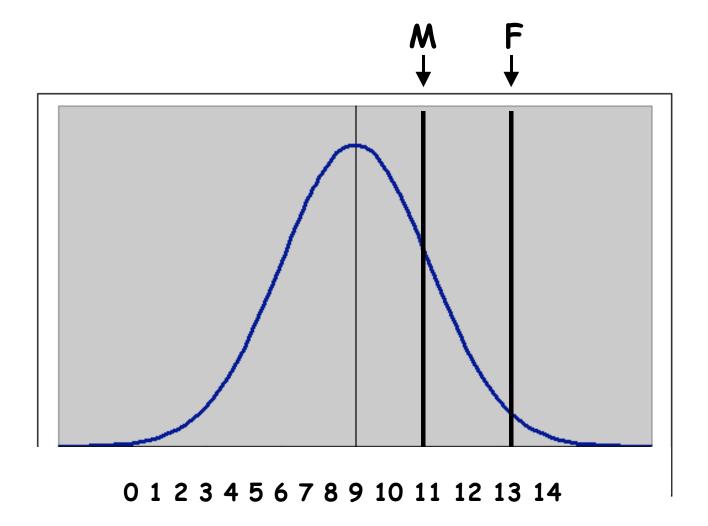
Twin 1

MONOZYGOTIC TWINS

Trait	Heritability
Height	0.60-0.81
Weight	0.42-0.63
Body Mass Index (BMI)	0.50-0.90
Systolic Bood Pressure	0.17-0.39
Triglyceride level	0.68-0.72
Plasma cholesterol	0.37-0.50

Polygenic threshold traits: a certain number of alleles for the trait must be present before it appears phenotypically





Who has the greater probability of having a child with cleft lip and palate? A person with unilteral or with bilateral expression of the trait?

Terms to know:

Continuous trait

Threshold trait

Multifactorial inheritance

Additive genetic variants

Heritability

Concordance-discordance