BIO240

Pedigrees and Quantitative Genetics

I. Pedigree Analysis:

A. Symbols and meaning

B. Interpretation

1. Recessive

2. Dominant

3. X-linked

4. Maternal

C. Pedigrees and predictions

1. Inbreeding coefficients

- estimation of identity by descent i.e. probability that two alleles in an individual are descended from the same single copy of the allele in an ancestor.

II. Quantitative Genetics:

A. Types of variation

1. Discontinuous - intermediate classes are not possible (color and plant size in Mendel's peas)

2. Continuous - many intermediate types are theoretically possible (hair color and height in humans)

3. continuous traits usually result of polygenic or quantitative inheritance where two or more gene loci contribute to the phenotype in an additive manner

B. Expression of continuous traits from discrete phenotypes:

1. lack of fit to predicted F2 3:1 ratio

- does not support Mendelian hypothesis

2. William Bateson and Udny Yule proposed that outcome could be explained by Mendelian inheritance of a large number of genetic factors

each of which influences the trait in a cumulative or quantitative way (Multiple-gene hypothesis)

3. Tobacco corolloa length phenotype experiment of Edward East.

C. Workings of Additive Alleles

1. Multiple gene loci influence a single continuous trait in an additive (cumulative, quantitative) way
2. Each locus may be occupied by an additive or a non-additive allele
3. Roughly speaking, phenotype results from the sum total of all of the additive alleles
    a. most extreme positive phenotype where all loci are occupied by additive alternate alleles
    b. most extreme negative phenotype where all loci are occupied by non-additive alternate alleles
4. Presumed that total effect of each additive allele, though small, is roughly equal to any other
5. The more loci influencing the trait, the more potential phenotypic variation for the trait
6. Must be analyzed using large numbers of progeny

D. Calculating # of loci involved in determining a quantitative trait: determine the proportion of F2 progeny that resemble either parental phenotype (separately, not added together)

Proportion = $1/4^n$

III. Statistics of Quantitative Genetics:

A. General usefulness if statistics

1. reduce data to descriptive summaries

2. data from a small sample can be used to better understanding of population on the whole

3. two or more sets of data can be compared to determine whether they represent the same or different populations

B. Frequency distribution

C. Statistics of central tendency

1. mode

2. median

3. mean (arithmatic average)

D. Variance - measure of dispersion from the mean, average squared deviation of the measurements from the mean

(demonstrate that sum of the differences = 0;
explain that intuitively we would divide by "n" to get estimate of average dispersion per case, but use "n - 1" instead)

Variance is useful in quantitative genetics when effect of environment also influences the phenotype

E. S.D. - square root of variance, a measure of the average deviation from the mean in the original unit of measure

F. S.E., an estimate of the expected variation around the standard deviation assuming replicates of similar samples

IV. Heritability: a measure of how genotype (as opposed to the environment) influences expression of quantitative traits

A. Broad-sense heritability - H^2: estimating proportion of observed phenotypic variation attributable to genetic factors as opposed to environmental ones

$$V_P = V_G + V_E + (V_{GE})$$

P = phenotypic; G = Genotypic; E = Environment

Demonstrate control of each on implications of experimental design and interpretation and effects on V_{GE}

$$H^2 = V_G/V_P$$

Broad-sense heritability most useful when applied to specific populations and usually only if trait results from relative inbred condition i.e. genotype must be controllable

B. Narrow-sense Heritability: heritability analysis that can be applied to determine the selective potential of a trait

$$V_G = V_A + V_D + V_I$$

V_A: results from average effects of <u>additive</u> components of genes

V_D: deviation from purely additive effects due to <u>dominance</u> (i.e. the phenotypic expression of the heterozygote is not equal to the mean of the two corresponding homozygotes)

V_I: deviation from purely additive effects due to epistasis between loci (often negligible)

$$h^2 = V_A/V_P \quad \text{or} \quad h^2 = V_A/(V_E + V_A + V_D)$$

C. Artificial selection: creating a subpopulation of an extreme phenotype by isolating and interbreeding extreme members of the parent population

M = parental population;

M1 = extreme subset of parental population;

M2 = offspring from M1 cross

1. Formulas and relationships

2. Limits of narrow-sense heritability: when no more variation for the selected trait exists in the population then heritability equals zero.

3. Traits resistant to artificial selection: