Introduction to Quantitative Genetics

In principle:

We can extend single-locus -> multilocus -> quantitative models

1AA : 2Aa : 1 aa \rightarrow (1A_nA_n : 2A_na_n : 1a_na_n)ⁿ \rightarrow normal distribution

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Genotype / Phenotype correlation → Heritability
Genotypic expression depends on environment
Heritability (h<sup>2</sup>) estimates proportion of phenotypic variation due to genetic variation
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"Is It Genetic?" : <u>Genotype / Environmental interaction</u> is variable (& unpredictable) The <u>Norm of Reaction</u> describes this

Heritability is *not* inevitability; Genetics is *not* destiny

Variation can be **<u>quantified</u>** (review)

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mean \pm standard deviation: \overline{x} \pm \sigma variance: \sigma^2
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Variation follows "<u>normal distribution</u>" (bell-curve) iff Multiple loci are involved (quantitative) Each locus has about equal effect (additive) Each locus acts independently [interaction variance (see below) is minimal] *Ex.*: Suppose a trait is influenced by 5 loci, each with two alleles A & a A contributes 2 units to phenotype, a contributes 1 unit Range of contributions = (2u aa : 3u Aa: 4u AA)⁵ mean = 15 units, range 10 ~ 20 units (Aa Bb Cc Dd Ee) vs (aa bb cc dd ee ff ~ AA BB CC DD EE FF) 3⁵ = 243 genotype classes in 11 phenotype classes → variation continuous

Variation has two sources: genetic (σ^2_{G}) & environmental (σ^2_{E}) variance

<u>Variance is additive</u>: $\sigma^2_{A+B} = \sigma^2 A + \sigma^2 B$

phenotypic variance $\equiv \sigma^2 \mathbf{P} = \sigma^2 \mathbf{G} + \sigma^2 \mathbf{E} + \sigma^2 \mathbf{GxE}$

where σ^2_{GxE} is the interaction variance, if σ^2_G and σ^2_E are not independent

If $\sigma^2 G$ and $\sigma^2 E$ are assumed to be additive, then the interaction variance $\sigma^2 G_{XE} \sim 0$

additive variance $\equiv \sigma^2_A = \sigma^2_G + \sigma^2_E$

Heritability

 \equiv h² = $\sigma^2 \mathbf{G} / \sigma^2 \mathbf{A} = \sigma^2 \mathbf{G} / (\sigma^2 \mathbf{G} + \sigma^2 \mathbf{F})$

"heritability in the narrow sense" ≡ genetic component of the additive variance heritability h² is the fraction of the <u>(additive) phenotypic variance</u> due to genotypic variance

assuming genotype / phenotype relationship is independent of environment

ignoring interaction variance σ^2_{GXE}

genotype / phenotype relationship *differs* in different environments. *Ex.*: same strain of corn produces different yields in different fields

Artificial breeding indicates that phenotypic variation is highly heritable <u>Artificial selection</u> on agricultural species Commercially useful traits can be improved by selective breeding <u>Common Garden</u> experiments

Correlation shows apposition between variables (of regression analysis)

Mid-parent value

Offspring / midparent correlation estimates heritability

Limits of prediction from correlation & regression

For many quantitative traits in many organisms: $h^2 = 0.5 \sim 0.9$

Heritability is typically calculated in a single environment.

The <u>Norm of Reaction</u> mediates genotype through environment to produce phenotype <u>Variation within groups</u>: Is variation 'genetic' ? <u>Variation among groups</u>: Are differences 'genetic'?

Genetics, Heritability, & Society

"Is it Genetic?"

Myth 1: That which is *heritable* is purely *genetic* phenotypic variance $\equiv \sigma^2 \mathbf{p} = \sigma^2 \mathbf{G}$ [ignore $\sigma^2 \mathbf{E} \& \sigma^2 \mathbf{GxE}$]

Myth 2: That which is genetic is fixed & unchangeable

Ex.: <u>Heritability, IQ, & Education</u> <u>IQ test scores in *Homo*</u>: $h^2 = 0.7 \sim 0.8$ within groups Highly heritable traits can be <u>modified by environment</u> if σ^2_{GYE} is large

See also:

<u>Gray & Thompson (2004) [Fig. 3]</u>. Neurobiology of intelligence: ethics and science, <u>Nature</u> <u>Revs Neurosci 5: 471-482</u>. <u>Foster (2006)</u>. Science & Ethics in the Human Genome Project. <u>American Psychological Association (1995)</u> Intelligence: Knowns and Unknowns <u>American Eugenics Archive</u>

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