

Conifer Translational Genomics Network Coordinated Agricultural Project



CAP

Genomics in Tree Breeding and
Forest Ecosystem Management

Module 4 – Quantitative Genetics

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Quantitative genetics

- “Quantitative genetics is concerned with the inheritance of those differences between individuals that are of degree rather than of kind, quantitative rather than qualitative.” Falconer and MacKay, 1996
- Addresses traits such as
 - *Growth, survival, reproductive ability*
 - *Cold hardiness, drought hardiness*
 - *Wood quality, disease resistance*
 - *Economic traits! Adaptive traits! Applied and evolutionary*
- Genetic principles
 - *Builds upon both Mendelian and population genetics*
 - *Not limited to traits influenced by only one or a few genes*
 - *Analysis encompasses traits affected by many genes*

Height in humans is a quantitative trait



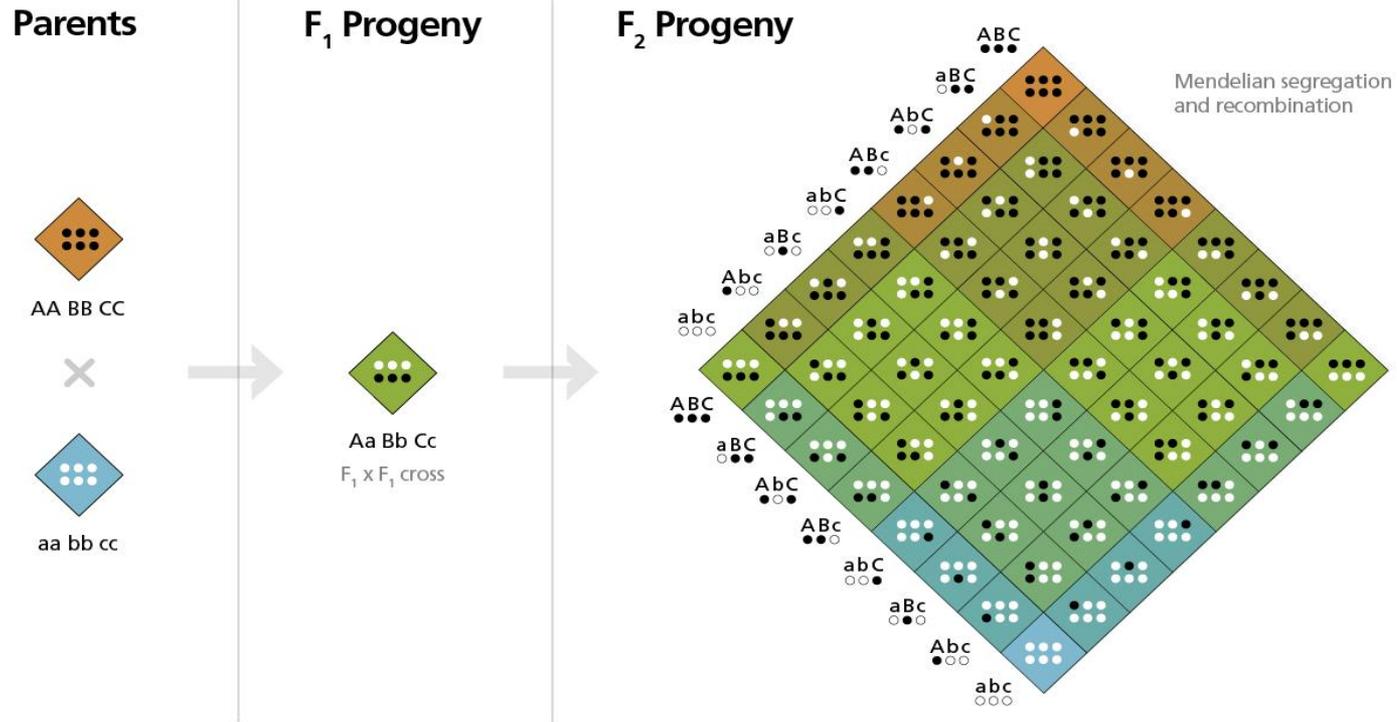
Students from the University of Connecticut line up by height: 5'0" to 6'5" in 1" increments. Women are in white, men are in blue

Figure Credit: Reproduced with permission of the Genetics Society of America, from [Birth defects, jimsonweed and bell curves, J.C. Crow, Genetics 147, 1997]; permission conveyed through Copyright Clearance Center, Inc.

Quantitative genetics

- Describes genetic variation based on phenotypic resemblance among relatives
- Is usually the primary genetic tool for plant and animal breeding
- Provides the basis for evaluating the relative genetic merit of potential parents
- Provides tools for predicting response to selection (genetic gain)
- How can we explain the continuous variation of metrical traits in terms of the discontinuous categories of Mendelian inheritance?
 - *Simultaneous segregation of many genes*
 - *Non-genetic or environmental variation (truly continuous effects)*

Consider a trait influenced by three loci



The number of 'upper-case' alleles (black dots) behave as unit doses. Genotypes with comparable doses are grouped together in colored boxes. In this example, gene effects are additive

Figure Credit: Modified from Hartl and Jones. 2001. Fig. 18.4.

Phenotypes

- Phenotypic categories from the previous slide are represented here in the histogram

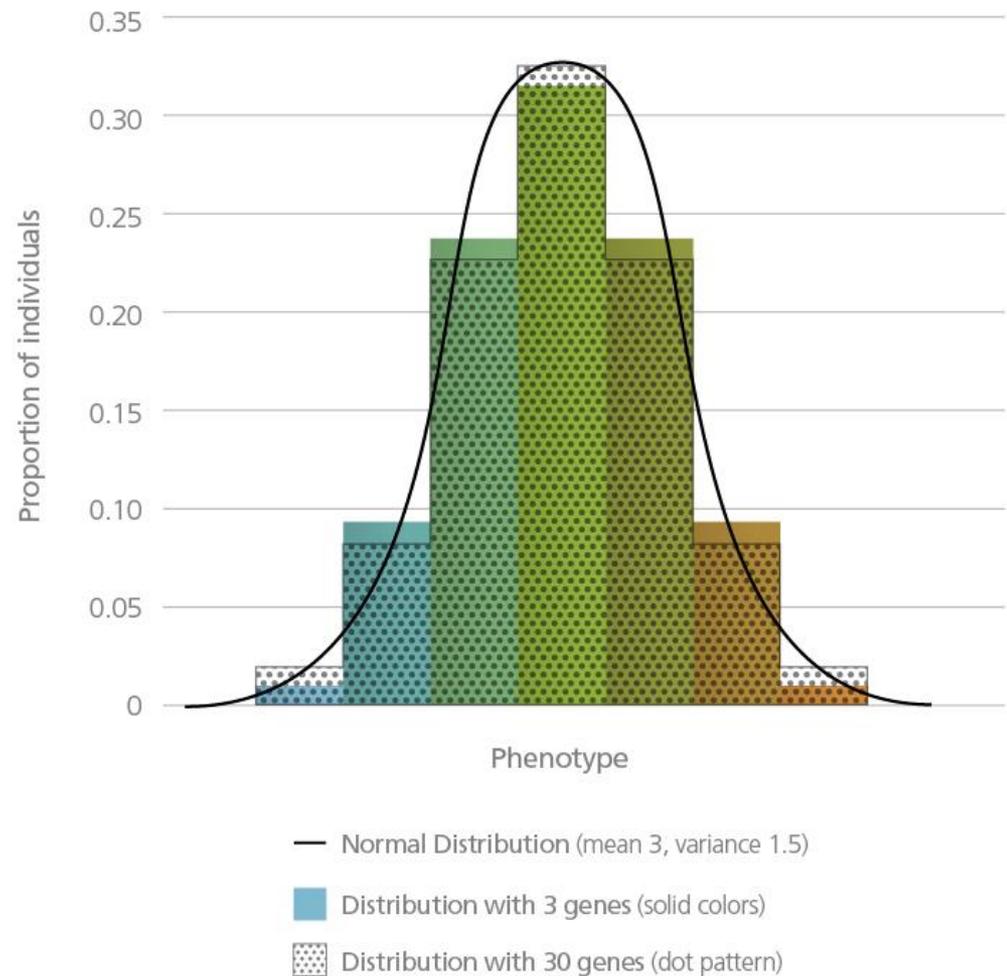
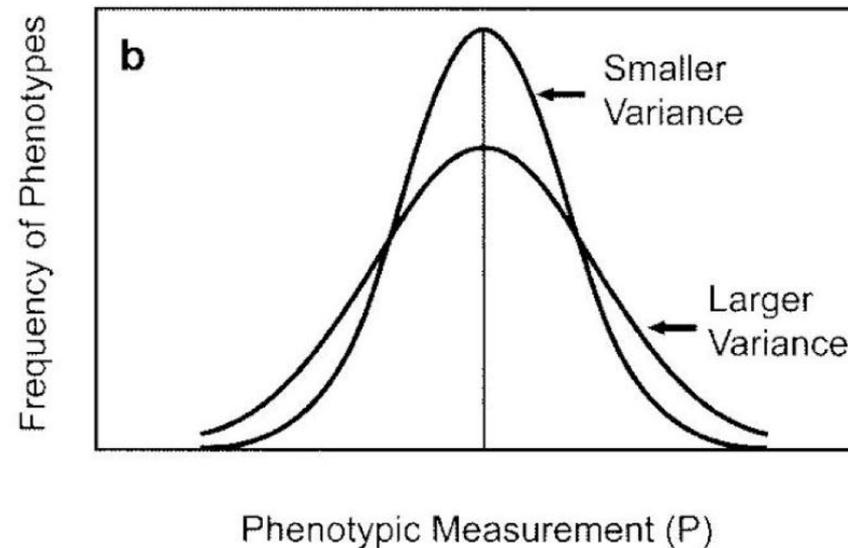


Figure Credit: Modified from Hartl and Jones. 2001. Fig. 18

How to describe a population?

- Mean \approx average
- Variance is dispersion around the mean
 - *Individual observations (usually) differ from the mean*
 - *Deviation is distance from mean*
 - *Variance is average squared deviation*



Population properties for metric traits

Means, variances, covariances

- Measuring variation within and among families allows estimation of genetic and environmental variance components
- Phenotypic resemblance among relatives allows estimation of trait heritability, parental breeding values, genetic correlations among traits, and so forth

Properties of genes

- Gene action (dominance, additive) – Allelic interactions at a locus
- Epistasis – Non-allelic interactions
- Pleiotrophy – Allelic effects on multiple traits
- Linkage

Phenotypic expression of a metrical trait

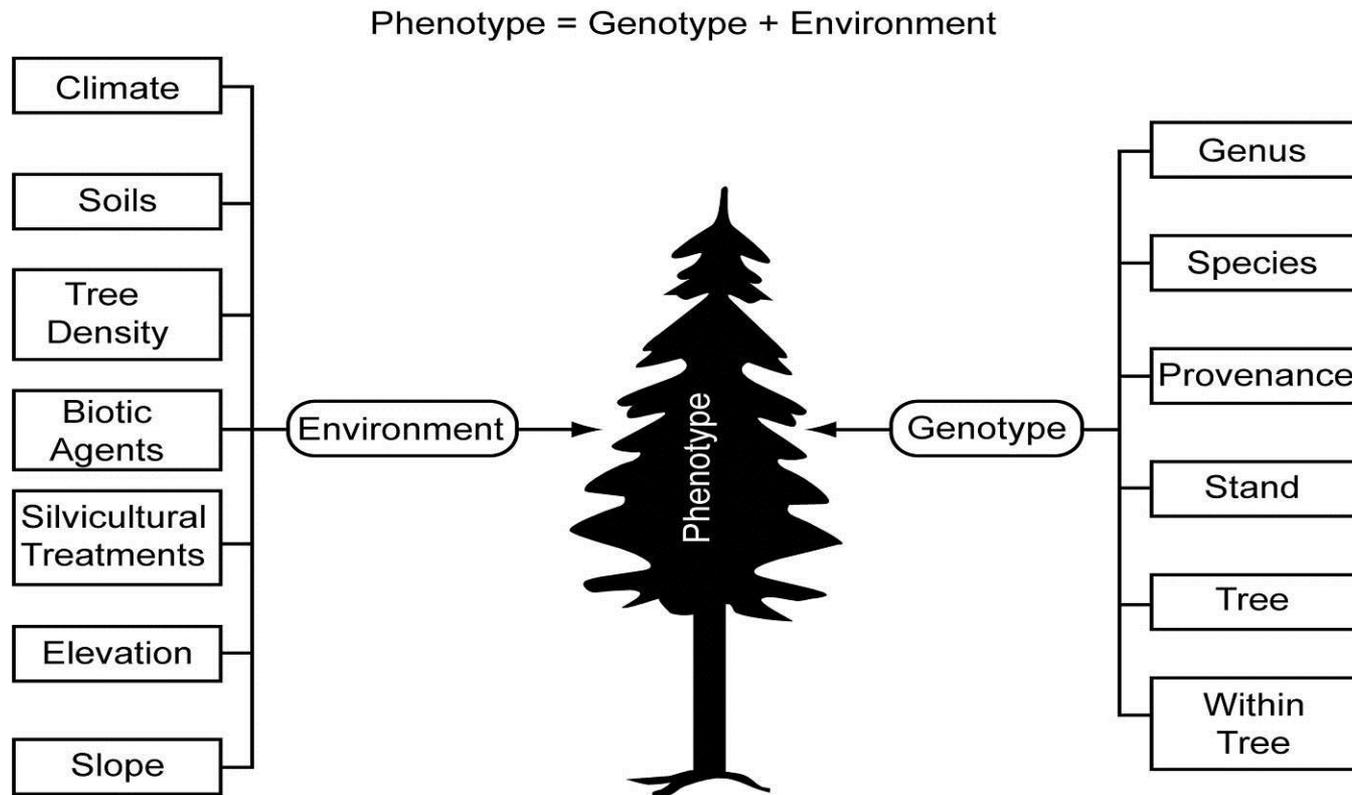


Figure Credit: White, T. L, W. T. Adams, and D. B. Neale. 2007. Forest genetics. CAB International, Wallingford, United Kingdom. Used with permission.

Partitioning phenotypic variance

- The phenotypic variance among individual trees in a reference population for a given trait, σ^2_p , is derived as

$$\text{Var (P)} = \text{Var } (\mu) + \text{Var (A)} + \text{Var (I)} + \text{Var (E)}$$

Or

$$\sigma^2_p = \sigma^2_A + \sigma^2_I + \sigma^2_E$$

- Where
 - μ = Pop mean = constant, with 0 variance
 - A = Additive genetic variance (breeding value)
 - I = Non-additive genetic variance
 - E = Environmental variance

Non-additive genetic variance

$$\sigma^2_I = \sigma^2_D + \sigma^2_\epsilon$$

- Dominance variance
 - *Genetic variance at a single locus attributable to dominance of one allele over another*
- Epistatic variance
 - *The masking of the phenotypic effect of alleles at one gene by alleles of another gene. A gene is said to be epistatic when its presence suppresses or obscures the effect of a gene at another locus*

Breeding value (additive genetic value)

- Breeding value is a concept associated with parents in a sexually breeding population. The sum of all average allelic effects at all loci influencing the trait of interest
 - *Alleles, not genotypes, are passed on to the next generation*
- Historically, average allelic effects could not be measured. With the ability to identify allelic states at the molecular level, we can now obtain estimates of allelic effects in controlled experiments
 - *The relevance of this extends beyond tree improvement to management of natural populations*

Heritability

A measure of the degree to which the variance in the distribution of a phenotype is due to genetic causes

- In the narrow sense, it is measured by the genetic variance due to additive effects divided by the total phenotypic variance
- In the broad sense, heritability is measured by the total genetic variance divided by the total phenotypic variance
- Heritability is mathematically defined in terms of population variance components. It can only be estimated from experiments that have a genetic structure: Sexually produced offspring in this case

More h^2

- Thus, narrow sense heritability can be written as

$$h^2 = \sigma^2_A / (\sigma^2_A + \sigma^2_I + \sigma^2_E)$$

- Where
 - σ^2_A is the additive genetic variance (variance among breeding values in a reference population)
 - σ^2_I is the interaction or non-additive genetic variance (which includes both dominance variance and epistatic variance)
 - σ^2_E is the variance associated with environment

Broad sense heritability (H^2 , or h^2_b)

- Broad sense heritability is used when we deal with clones! Clones can capture all of genetic variance due to both the additive breeding value and the non-additive interaction effects. Thus,

$$H^2 = (\sigma^2_A + \sigma^2_I) / (\sigma^2_A + \sigma^2_I + \sigma^2_E)$$

- Consequently, broad sense heritability is typically larger than narrow sense heritability and progress in achieving genetic gain can be faster when clonal selection is possible. What might be a drawback to clonal based programs?

Calculating genetic gain

$$G = i h^2 \sigma_p$$

- Selection Intensity (i)
 - *Difference between the mean selection criterion of those individuals selected to be parents and the average selection criterion of all potential parents, expressed in standard deviation units*
 - *The proportion of trees selected from the population of trees measured for the trait*
- Heritability (h^2 or H^2)
 - *Measure of the degree to which the variance in the distribution of a phenotype is due to genetic causes*
- Phenotypic standard deviation of a trait (σ_p)

A little more on selection intensity

- The factor most under breeder's control
- i increases as the fraction of trees selected decreases

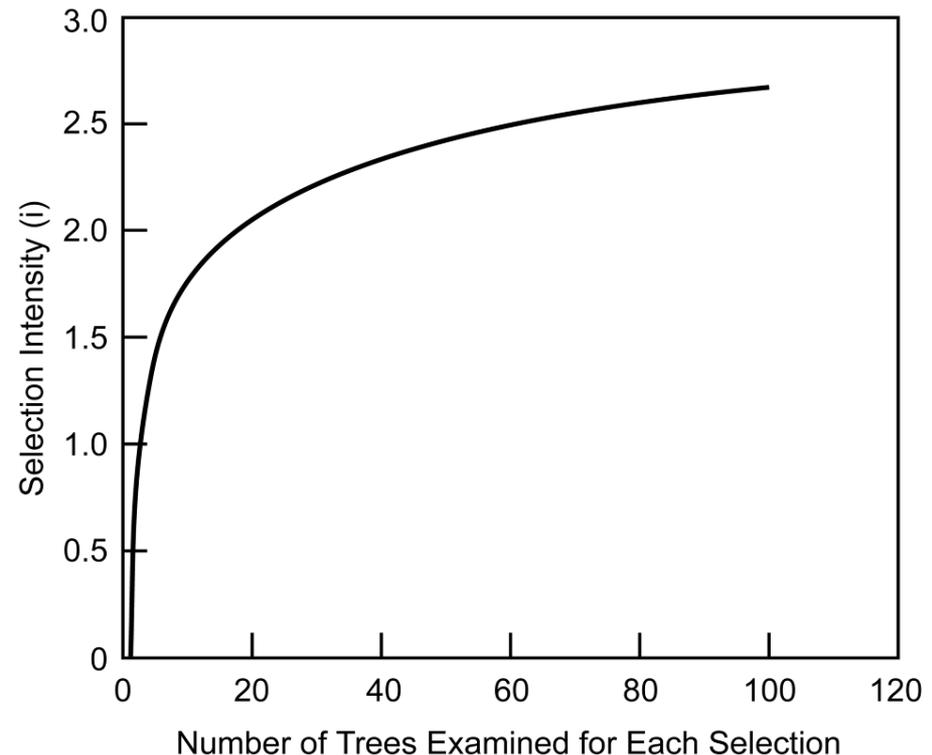


Figure Credit: White, T. L, W. T. Adams, and D. B. Neale. 2007. Forest genetics. CAB International, Wallingford, United Kingdom. Used with permission.

Predicting genetic gain

$$\text{Gain} = h^2 \times (\text{selection differential})$$

selection differential = $i \times \sigma_p$

$$\text{Gain} = h^2 \times i \times \sigma_p$$

Get more gain by controlling the environmental variation and increasing h^2

Get more gain by selecting a smaller proportion of the population (increased i)

Including more traits

- How do the models change as we examine more traits?
- Additional consideration must be paid to
 - *Genetic correlations*

Genetic correlations

- Correlations in phenotype
 - *May be due to genetic or environmental causes*
 - *May be positive or negative*
- Genetic causes may be due to
 - *Pleiotropy*
 - *Linkage*
 - *Gametic phase disequilibrium*
- The **additive genetic correlation** (correlation of breeding values) is of greatest interest to plant breeders
 - **Genetic correlation** usually refers to the additive genetic correlation (r_G is usually r_A)

Selection

- Using genetic markers (marker informed breeding) to facilitate selection of the best individuals requires a working knowledge of other concepts
 - *Indirect selection and correlated response to selection*
 - *Multi-trait selection*

Indirect selection

- Indirect selection occurs when individuals are selected on the measurements of one trait (Y) and gain is predicted for a second, or target, trait (X). Gain from indirect selection is estimated as

$$G_x = i_y * r_{g,xy} * h_x h_y * \sigma_{px}$$

- Where
 - i_y = selection intensity of the measured trait
 - $r_{g,xy}$ = the genetic correlation between measured and target traits
 - h = square root of the heritability of traits x and y
 - σ_{px} = phenotypic standard deviation of the target trait
- All terms are unitless except the last, so predicted gain is given in terms of the target trait

Indirect selection is better when...

- To compare the relative effectiveness of indirect and direct selection we compare the ratio of gains from the two approaches

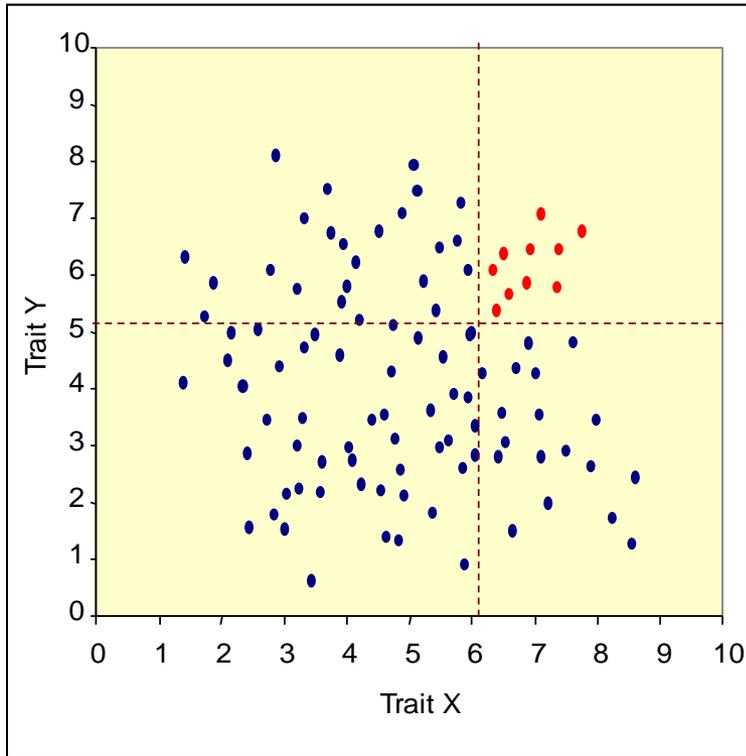
$$\begin{aligned} &= (i_y * r_{g,xy} * h_x h_y * \sigma_{px}) / (i_x * h_x^2 * \sigma_{px}) \\ &= r_{g,xy} (i_y / i_x) (h_y / h_x), \text{ therefore} \end{aligned}$$

- Dependent on size and sign of genetic correlation (r)
- When selection intensity is greater for measured trait (i)
- When heritability of measured trait is higher (h_y)
- Cost/Time considerations

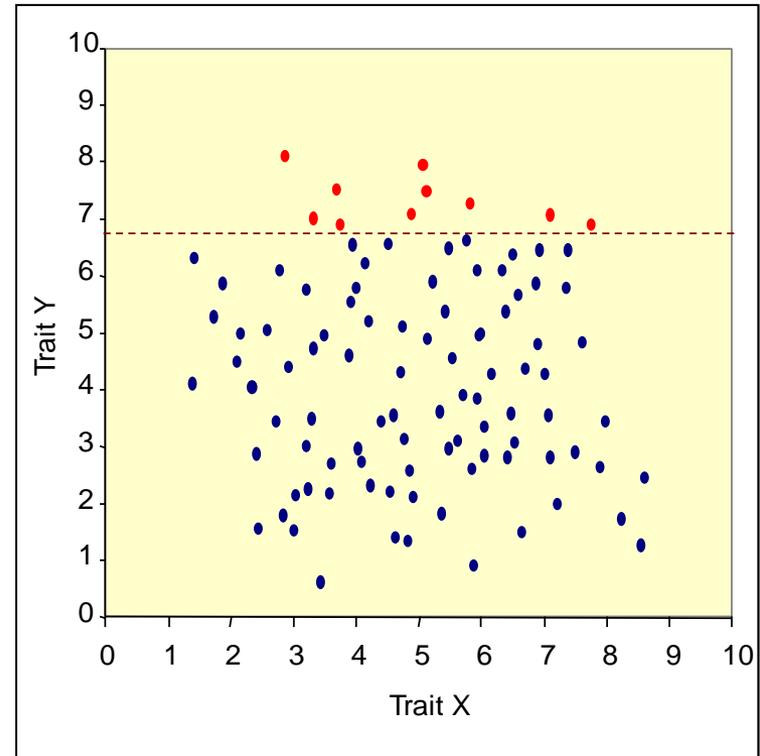
Strategies for multiple trait selection

- We often wish to improve more than one trait at a time
- Traits may be correlated or independent from each other
- Options...
 - *Independent culling*
 - *Tandem selection*
 - *Index selection*

Strategies for multiple trait selection



Independent culling



Tandem selection

Figure credits: Jennifer Kling, Oregon State University

Selection indices

- Values for multiple traits are incorporated into a single index value for selection

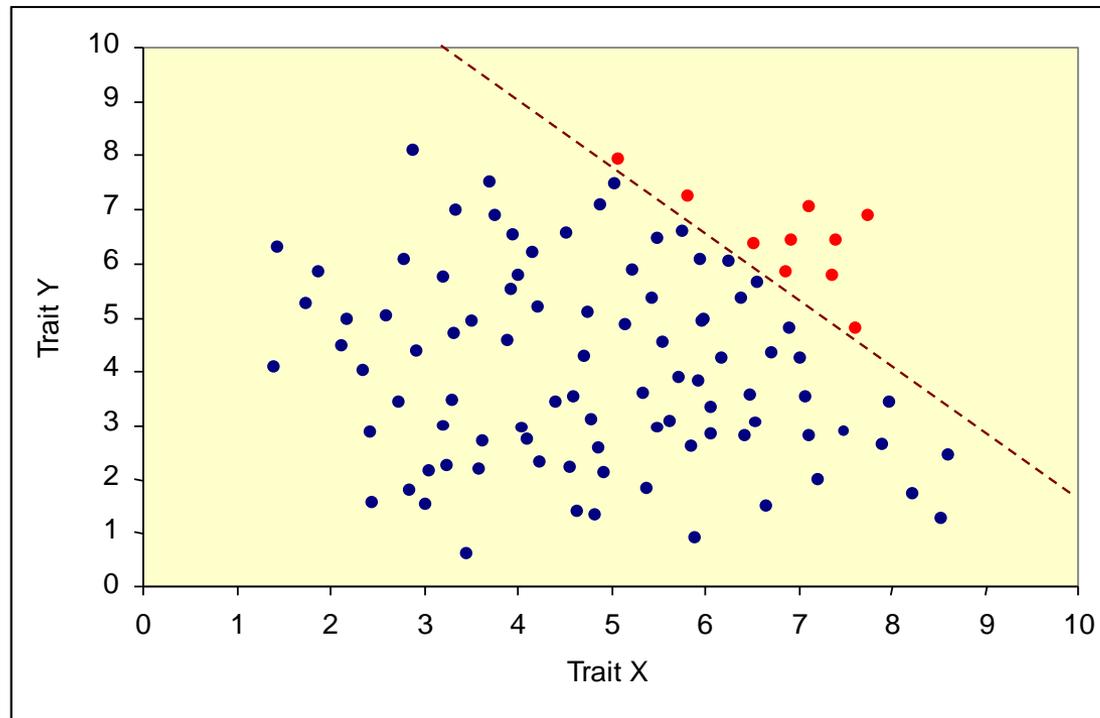


Figure credit: Jennifer Kling, Oregon State University

Estimating variance components, genetic parameters, and breeding values

- Mixed models – genetic effects considered random
- GLS – (Generalized Least Squares) for estimating fixed effects (called BLUE)
- REML (Restricted Maximum Likelihood) for estimating variance components of random effects
- Additive genetic relationship matrix
- BLUP (Best Linear Unbiased Prediction) for estimating breeding values. Selection Indices are a special case of BLUP

Estimating a tree's genotype

- Historically through evaluation trials of phenotypic traits
- As genomics tools and platforms have developed, we are more seriously evaluating the potential of genetic markers to augment phenotypic assessments
 - *QTL mapping in pedigreed populations*
 - *Association genetics*
- How might marker data be incorporated in breeding?
 - *BLUP – selection index*
 - *Additive genetic relationship matrix*
 - *Program management applications*
 - *Genomic selection*

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Thank You.

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