

**Day 3a**  
**BASIC QUANTITATIVE GENETICS**

**Objective**

Review some basic quantitative genetics important for subsequent material

1. **Single-locus quantitative genetic theory**
  - a. Average allele effects, allele substitution effects
  - b. Single-locus breeding values
  - c. Single-locus genetic variances
2. Extension to multiple loci
3. Model for breeding values of progeny
  - a. Parental average and Mendelian sampling terms
4. Environmental effects

1

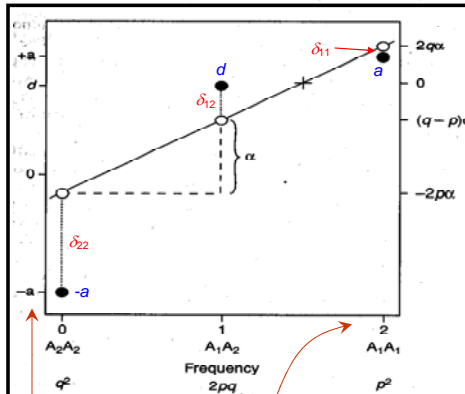
**1. SINGLE-LOCUS QUANTITATIVE GENETIC THEORY**

Genotype	$A_2A_2$	$A_1A_2$	$A_1A_1$	
Genotypic value	$\mu - a$	$\mu$	$\mu + d$	$\mu + a$
		$\mu$ usually = 0		

**Single locus allelic effects model for individual with genotype  $T = A_iA_j$ :**

Genotypic value =  $G_{ij} = M + \alpha_i + \alpha_j + \delta_{ij}$   
 $M$  = population mean (see previous page)  
 $\alpha_i$  = average effect of allele  $i$   
 $\alpha_j$  = average effect of allele  $j$   
 $\delta_{ij}$  = dominance deviation effect of the interaction of alleles  $i$  and  $j$

**Average effect  $\alpha_i$**  = Average deviation from the population mean of individuals receiving **allele  $i$**  from one parent with the **other allele** having come at random from the population



**Allele substitution effect =  $\alpha = \alpha_1 - \alpha_2 = a + (q - p)d$**

= coefficient of regression of genotypic value on number of  $A_1$  alleles

2

## SINGLE LOCUS BREEDING VALUES

Geno- type $T$	Fre- quen- cy	Genotypic value	Average allele effects		Dominance deviations	Breeding value
		$G_T$	$\alpha_i$	$\alpha_j$	$\delta_{ij}$	$A_{ij} = \alpha_i + \alpha_j$
$A_1A_1$	$p^2$	a	$\alpha_1 = q\alpha$	$\alpha_1 = q\alpha$	$-2q^2d$	$2\alpha_1 = 2q\alpha$
$A_1A_2$	$2pq$	d	$\alpha_1 = q\alpha$	$\alpha_2 = -p\alpha$	$2pqd$	$\alpha_1 + \alpha_2 = (q-p)\alpha$
$A_2A_2$	$q^2$	-a	$\alpha_2 = -p\alpha$	$\alpha_2 = -p\alpha$	$-2p^2d$	$2\alpha_2 = -2p\alpha$

Breeding value (BV) = 2 x expected deviation of the individual's progeny mean from the population mean when the individual is mated at random.

BV for individual with genotype  $A_iA_j = A_{ij} = 2 E(P_{\text{progeny}} - M)$

$$= 2 * \left( \frac{1}{2}\alpha_i + \frac{1}{2}\alpha_j \right) = \alpha_i + \alpha_j$$

Prob( $A_i$  passed on)     Ave.effect of  $A_i$   
Ave.effect of  $A_i$      Prob( $A_i$  passed on)

3

## SINGLE LOCUS GENETIC VARIANCES

Geno- type $T$	Fre- quen- cy	Genotypic value	Average allele effects		Dominance deviations	Breeding value
		$G_T$	$\alpha_i$	$\alpha_j$	$\delta_{ij}$	$A_{ij} = \alpha_i + \alpha_j$
$A_1A_1$	$p^2$	a	$\alpha_1 = q\alpha$	$\alpha_1 = q\alpha$	$-2q^2d$	$2\alpha_1 = 2q\alpha$
$A_1A_2$	$2pq$	d	$\alpha_1 = q\alpha$	$\alpha_2 = -p\alpha$	$2pqd$	$\alpha_1 + \alpha_2 = (q-p)\alpha$
$A_2A_2$	$q^2$	-a	$\alpha_2 = -p\alpha$	$\alpha_2 = -p\alpha$	$-2p^2d$	$2\alpha_2 = -2p\alpha$

**Variance of genotypic values** = (Total) **Genetic variance** =  $\sigma_G^2$

$$\sigma_G^2 = \text{var}(G_T) = p^2 a^2 + 2pqd^2 + q^2 a^2 - M^2 = 2pq[a + (q-p)d]^2 + (2pqd)^2 =$$

with:  $\alpha = a + (q-p)d$       $= 2pq\alpha^2$       $+ (2pqd)^2$

**Additive genetic variance** = variance of additive genetic values =  $\sigma_A^2$

$$\sigma_A^2 = \text{var}(A_T) = p^2(2q\alpha)^2 + 2pq[(q-p)\alpha]^2 + q^2(-2p\alpha)^2 - O^2 = 2pq\alpha^2 \quad (\text{Note that } E(A_T)=0)$$

Also:  $\sigma_A^2 = \text{var}(A_T) = \text{var}(\alpha_i + \alpha_j) = \text{var}(\alpha_i) + \text{var}(\alpha_j) = pq\alpha^2 + pq\alpha^2 = 2pq\alpha^2$

→ Variance associated with each of the 2 alleles that an individual carries =  $\frac{1}{2}\sigma_A^2$

**Dominance variance** = variance of Dominance deviations =  $V_D$

$$\sigma_D^2 = \text{var}(D_T) = \text{var}(\delta_{ij}) = p^2(-2q^2d)^2 + 2pq(2pqd)^2 + q^2(-2p^2d)^2 - O^2 = (2pqd)^2 \quad (\text{Note: } E(D_T)=0)$$

4

**Example: the pygmy gene in mice** Allele frequency  $\Pr(A_1) = p = 0.6$   $q = 0.4$

	Genotype		
	$A_1A_1$	$A_1A_2$	$A_2A_2$
Weight (g)	14	12	6
$\mu = 10$	$a = 4$	$d = 2$	$-a = -4$
Frequency (HWE)	$p^2=0.36$	$2pq=0.48$	$q^2=0.16$

$$V_G = 2 \cdot .6 \cdot .4 \cdot [4 + (.4 - .6) \cdot 2]^2 + (2 \cdot .6 \cdot .4 \cdot 2)^2 = .48 \cdot [3.6]^2 + (.96)^2 = 6.22 + .92 = 7.14$$

$$V_A = 6.22 \quad V_D = .92 \quad \text{Genetic standard deviation} = \sigma_G = \sqrt{V_G} = \sqrt{7.14} = 2.67$$

$$\text{Additive genetic s.d.} = \sigma_A = \sqrt{V_A} = \sqrt{6.22} = 2.49$$

$$\text{Dominance genetic s.d.} = \sigma_D = \sqrt{V_D} = \sqrt{0.92} = 0.96$$

### Special cases

No dominance:  $V_A = 2pq a^2$

$$V_D = 0$$

$p = q = 0.5$ :  $V_A = \frac{1}{2} a^2$

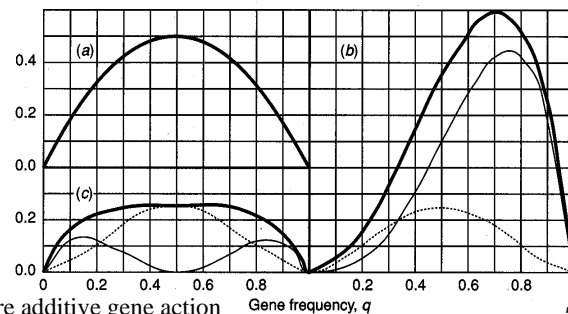
$$V_D = \frac{1}{4} d^2$$

(F & M, p. 128)

(a):  $a > 0, d = 0$

(b):  $a > 0, d = a$

(c):  $a = 0, d > 0$



→ Additive variance does not require additive gene action

## EXTENSION TO MULTIPLE LOCI – without epistasis

For individual with alleles  $i$  and  $j$  at locus A and  $k$  and  $l$  at locus B

$$\begin{aligned} \text{Genotypic value} = G_T &= G_A + G_B & G_i &= \text{genotypic value locus } i \\ &= A_A + \delta_A + A_B + \delta_B = (A_A + A_B) + (\delta_A + \delta_B) \\ &= A_T + D_T \end{aligned}$$

$A_T =$  breeding value:  $A_{ijkl} = \alpha_{A_i} + \alpha_{A_j} + \alpha_{B_k} + \alpha_{B_l}$  with each  $\alpha_{ni}$  as for 1-locus case

$D_T =$  dominance deviation:  $D_{ijkl} = \delta_{A_{ij}} + \delta_{B_{kl}}$  with each  $\delta_{nij}$  as for 1-locus case

### Genetic variance:

$$\begin{aligned} \sigma_G^2 = \text{var}(G_T) &= \text{var}(G_A + G_B) = \text{var}(G_A) + \text{var}(G_B) + 0 \quad \text{if loci are in LE} \\ &= \{\sigma_{A_A}^2 + \sigma_{D_A}^2\} + \{\sigma_{A_B}^2 + \sigma_{D_B}^2\} \\ &= \{\sigma_{A_A}^2 + \sigma_{A_B}^2\} + \{\sigma_{D_A}^2 + \sigma_{D_B}^2\} \\ &= \sigma_A^2 + \sigma_D^2 \end{aligned}$$

### With many loci:

Breeding value = sum of average effect of paternal and maternal

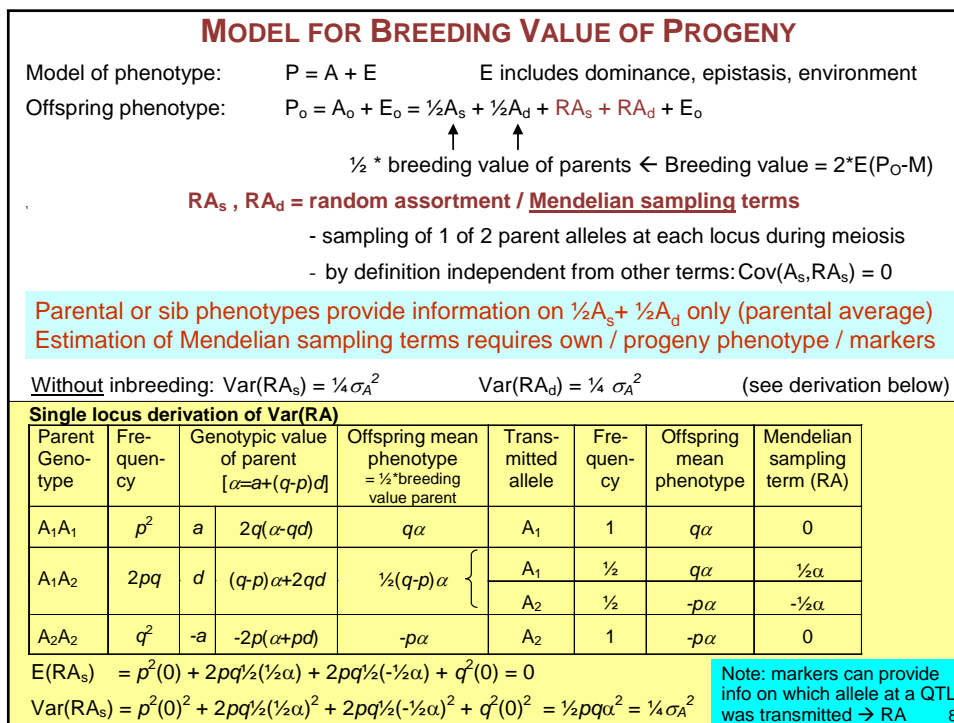
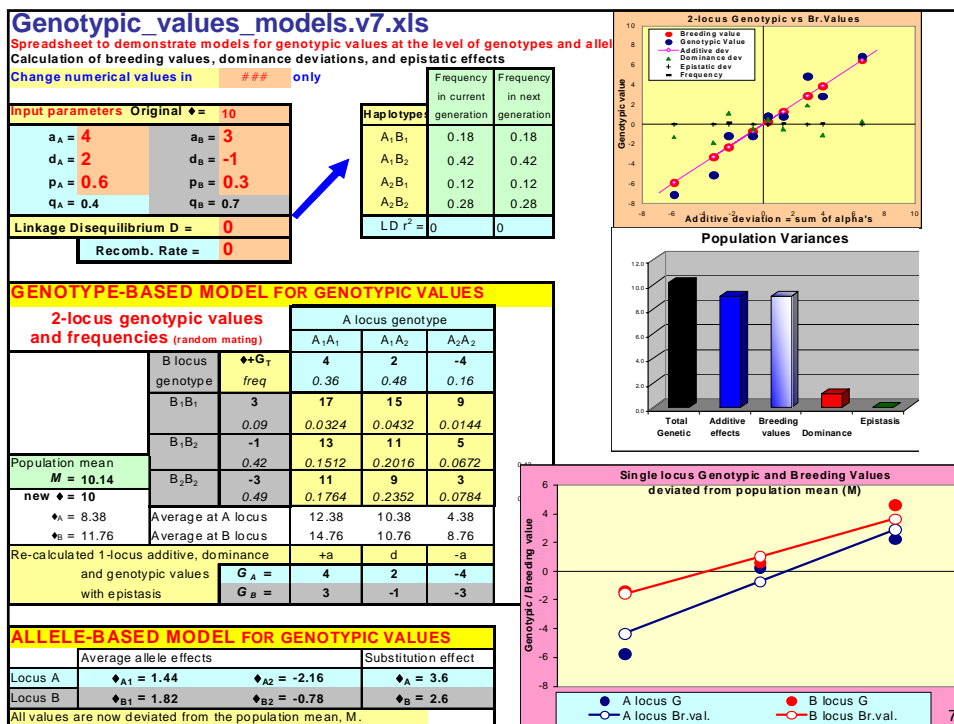
$$\text{alleles at all QTL} = A = \sum (\alpha_i^{pat} + \alpha_i^{mat})$$

$$\text{Genetic variance} = \sigma_G^2 = \sum \sigma_{G_i}^2 = \sum \sigma_{A_i}^2 + \sum \sigma_{D_i}^2 = \sigma_A^2 + \sigma_D^2$$

$$\text{Additive variance} = \sigma_A^2 = \sum \sigma_{A_i}^2 = \sum 2p_i q_i \alpha_i^2$$

$$\text{Dominance variance} = \sigma_D^2 = \sum \sigma_{D_i}^2$$

6



## ENVIRONMENTAL EFFECTS

Individual's phenotype is determined by genetic and environmental factors:

$$P = \mu + G + E$$

$\mu$  includes mean and *systematic (environmental) effects*

- Factors that can be identified and, therefore, be removed by statistical analysis by fitting them as effects in the model
  - E.g. herd, plot, year, season
  - Also: age, sex, parity

$G$  = genotypic value

$E$  = *Random environmental effects*

- effects of non-identifiable non-genetic factors that create differences in phenotype between individuals that are exposed to the same systematic effects
  - e.g. cows in same herd, plants in same field
  - e.g. micro-environmental differences in nutrition, climate, soil, housing, management
- effects of sources of external variation that are not under experimental control and that can, therefore, not be adjusted for by statistical analysis
- Also includes measurement error

**Phenotypic variance** = variance of phenotypes after removal/adjustment for systematic effects

$$= \sigma_p^2 = \text{var}(P-\mu) = \text{var}(G+E) = \text{var}(G) + \text{var}(E)$$

$$\text{Broad sense heritability } H^2 = \frac{\sigma_G^2}{\sigma_P^2} \quad \text{Narrow sense heritability } h^2 = \frac{\sigma_A^2}{\sigma_P^2}$$

9

## Day 3a

### BASIC QUANTITATIVE GENETICS

#### Objective

Review some basic quantitative genetics important for subsequent material

1. **Single-locus quantitative genetic theory**
  - a. Average allele effects, allele substitution effects
  - b. Single-locus breeding values
  - c. Single-locus genetic variances
2. Extension to multiple loci
3. Model for breeding values of progeny
  - a. Parental average and Mendelian sampling terms
4. Environmental effects

10