Basics of Quantitative Genetics

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Background

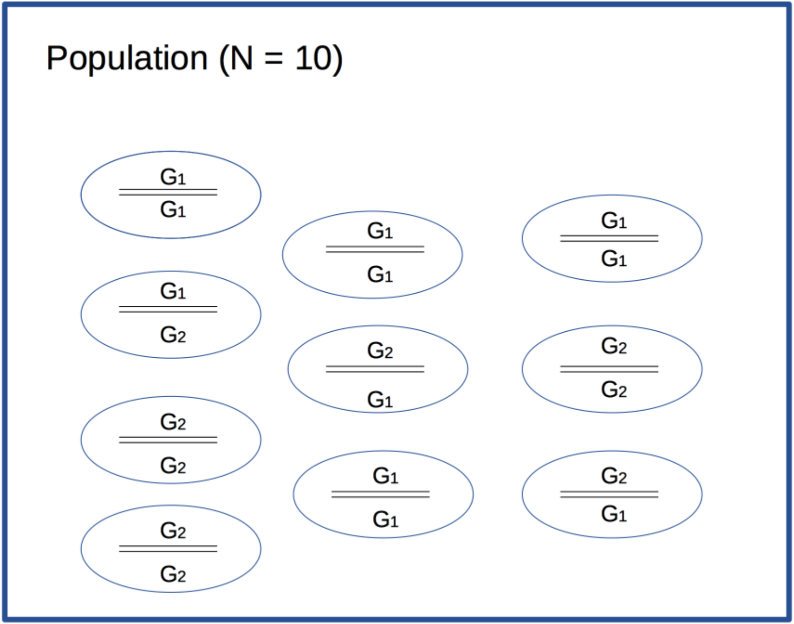
- Central Dogma of Molecular Biology

$\rightarrow$ Genotypes are the basis for phenotypic expression

- Start with simple model

$\rightarrow$ one locus that affects quantitative trait

Population


Population (N = 10)
G1 / G1
G1 / G2
G2 / G2
G2 / G2
G1 / G1
G2 / G1
G1 / G1
G1 / G1
G2 / G2
G2 / G1

Terminology

- **alleles**: variants occuring at a given genetic Locus
- **bi-allelic**: only two alleles, e.g., G_1 and G_2 at a given locus G in population
- **genotype**: combination of two alleles at locus G in an individual
- **homozygous**: genotypes G_1G_1 and G_2G_2 where both alleles identical
- **heterozygous**: genotype G_1G_2 different alleles

Frequencies in Example Population

- **genotype frequencies**

$$f(G_1G_1) = \frac{4}{10} = 0.4$$
$$f(G_1G_2) = \frac{3}{10} = 0.3$$
$$f(G_2G_2) = \frac{3}{10} = 0.3$$

- **allele frequencies**

$$f(G_1) = f(G_1G_1) + \frac{1}{2} * f(G_1G_2) = 0.55$$
$$f(G_2) = f(G_2G_2) + \frac{1}{2} * f(G_1G_2) = 0.45$$

Hardy-Weinberg Equilibrium

- **allele frequencies**

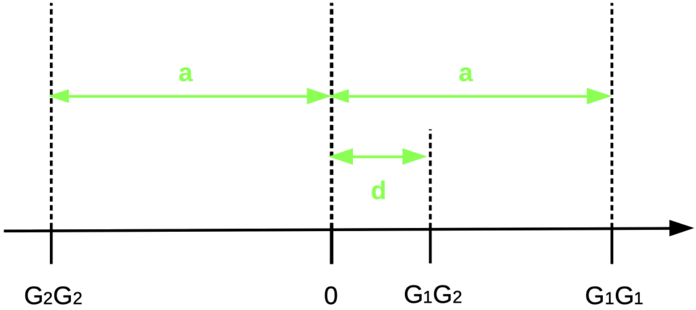
$$f(G_1) = p,\ f(G_2) = q = 1 - p$$

- **genotype frequencies**

Alleles	G_1	G_2
G_1	$f(G_1G_1) = p^2$	$f(G_1G_2) = p * q$
G_2	$f(G_1G_2) = p * q$	$f(G_2G_2) = q^2$

$$f(G_1G_1) = p^2,\ f(G_1G_2) = 2pq,\ f(G_2G_2) = q^2$$

Genotypic Values



Population Mean

- Expected value of genotypic value V as discrete random variable

$$
\begin{aligned}
\mu &= V_{11} * f(G_1G_1) + V_{12} * f(G_1G_2) + V_{22} * f(G_2G_2) \\
&= a * p^2 + d * 2pq + (-a) * q^2 \\
&= (p - q)a + 2pqd
\end{aligned}
$$

Breeding Values Definition

The breeding value of an animal i is defined as two times the difference between the mean value of offsprings of animal i and the population mean.

Derivation of Breeding value for G_1G_1

	Mates of S	
	$f(G_1) = p$	$f(G_2) = q$
Parent S		
$f(G_1) = 1$	$f(G_1G_1) = p$	$f(G_1G_2) = q$

Computation of Breeding value for G_1G_1

$$\mu_{11} = p * a + q * d$$

The breeding value BV_{11} corresponds to

$$\begin{aligned}
BV_{11} &= 2 * (\mu_{11} - \mu) \\
&= 2\left(pa + qd - [(p-q)a + 2pqd]\right) \\
&= 2\left(pa + qd - (p-q)a - 2pqd\right) \\
&= 2\left(qd + qa - 2pqd\right) \\
&= 2\left(qa + qd(1-2p)\right) \\
&= 2q\left(a + d(1-2p)\right) \\
&= 2q\left(a + (q-p)d\right)
\end{aligned}$$

Computation of Breeding value for G_2G_2

$$\mu_{22} = pd - qa$$

The breeding value BV_{22} corresponds to

$$\begin{aligned}
BV_{22} &= 2 * (\mu_{22} - \mu) \\
&= 2\,(pd - qa - [(p-q)a + 2pqd]) \\
&= 2\,(pd - qa - (p-q)a - 2pqd) \\
&= 2\,(pd - pa - 2pqd) \\
&= 2\,(-pa + p(1-2q)d) \\
&= -2p\,(a + (q-p)d)
\end{aligned}$$

Computation of Breeding value for G_1G_2

$$\mu_{12} = 0.5pa + 0.5d - 0.5qa = 0.5\left[(p-q)a + d\right]$$

The breeding value BV_{12} corresponds to

BV12

$$\begin{aligned}
ZW_{12} &= 2 * (\mu_{12} - \mu) \\
&= 2\left(0.5(p-q)a + 0.5d - \left[(p-q)a + 2pqd\right]\right) \\
&= 2\left(0.5pa - 0.5qa + 0.5d - pa + qa - 2pqd\right) \\
&= 2\left(0.5(q-p)a + (0.5 - 2pq)d\right) \\
&= (q-p)a + (1-4pq)d \\
&= (q-p)a + (p^2 + 2pq + q^2 - 4pq)d \\
&= (q-p)a + (p^2 - 2pq + q^2)d \\
&= (q-p)a + (q-p)^2 d \\
&= (q-p)\left[a + (q-p)d\right]
\end{aligned}$$