

Lecture 14. Broad sense heritability. Narrow sense heritability.

5.1 Broad sense heritability

As a rule a QT is influenced by many genes (polygenes)
each gene exerting relatively small effect
considerable environmental variation

Ex 1: bristle number

Fig 9.1, p. 399: *D.melanogaster*

X = number of bristles on 5th abdominal sternite
histogram with $N(\bar{X}, s^2)$ curve, $\bar{X} = 18.7$, $s = 2.1$
phenotypic variance $\sigma_p^2 = \text{Var}(X) \approx (2.1)^2$

Phenotypic value of an individual: $P = M + E$

genotype value $M = \mu + G$ (mean μ)

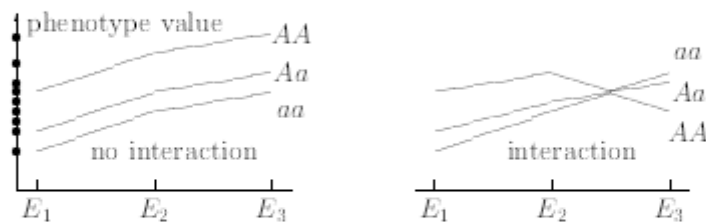
environmental deviation E (mean 0)

Assumption: independent deviations G and E

$P = \mu + G + E$, variance decomposition $\sigma_p^2 = \sigma_g^2 + \sigma_e^2$

$$\boxed{\text{Broad sense heritability } H^2 = \sigma_g^2 / \sigma_p^2}$$

Absence of genotype-environment interaction:



Examples of environmental effects:

nutritional and climatic factors, maternal effects
cultural environment

Ex 2: estimation of variance components

- 1) estimate σ_e^2 observing inbred populations
- 2) find $\sigma_g^2 + \sigma_e^2$ from random-bred populations

Thorax length in *D.melanogaster* (in 0.01 mm)

inbred populations $\sigma_e^2 = 0.186$

random-bred populations $\sigma_p^2 = 0.366 (= \sigma_g^2 + \sigma_e^2)$

$\sigma_g^2 = \sigma_p^2 - \sigma_e^2 = 0.180$, $H^2 = 49.2\%$

Breeding value of a genotype

A = twice the average G across possible offspring
factor 2: only half of genes come from one parent
Dominance effect $D = G - A$, within locus interaction

$$P = \mu + A + D + E$$

5.2 Narrow sense heritability

One locus model

two alleles A_1 and A_2 with frequencies p and q
assuming random mating and HWE

genotype	A_1A_1	A_1A_2	A_2A_2
frequency	p^2	$2pq$	q^2
M	$\mu^* + a$	$\mu^* + d$	$\mu^* - a$
G	$2q\alpha - 2q^2d$	$(q-p)\alpha + 2pqd$	$-2p\alpha - 2p^2d$
A	$2q\alpha$	$(q-p)\alpha$	$-2p\alpha$
D	$-2q^2d$	$2pqd$	$-2p^2d$

Genotypic values M given around $\mu^* = \frac{M_{11} + M_{22}}{2}$

do not depend on the allele frequency p

Genotypic deviation $G = M - \mu$ depends on p

since population mean $\mu = \mu^* + (p - q)a + 2pqd$

Average effect of gene substitution

when a randomly chosen A_1 is replaced by A_2

$$\alpha = p(a - d) + q(a + d)$$

Breeding value of A_1A_1 genotype

$$A_{11} = 2(pG_{11} + qG_{12}) = 2q\alpha$$

random mate contributes allele A_1 with probability p

A and D values are uncorrelated

additive variance $\sigma_a^2 = 2pq\alpha^2$, additive gene effects

within locus interaction component $\sigma_d^2 = (2pqd)^2$

$$\sigma_g^2 = \sigma_a^2 + \sigma_d^2, \text{ so that } \sigma_p^2 = \sigma_a^2 + \sigma_d^2 + \sigma_e^2$$

$$\text{Narrow sense heritability } h^2 = \sigma_a^2 / \sigma_p^2$$

Narrow heritability h^2 is more important than broad H^2

since parents pass their genes not genotypes

Ex 3: LDL-cholesterol level

Narrow sense heritability

$$554.2 = 500.0 + 39.2 + 15.0, h^2 = 0.071$$

Ex 4: inheritance of rare diseases

autosomal disease allele A_2 of low frequency $q \approx 0$

assuming $\sigma_e^2 = 0$

Rare recessive disease: $d = a$

$$\alpha = 2qa, \sigma_a^2 = 8pq^3a^2, \sigma_d^2 = 4p^2q^2a^2$$

Low inheritance $h^2 = \frac{\sigma_a^2}{\sigma_a^2 + \sigma_d^2} = \frac{q}{1+q} \approx 0$

affected offspring come usually from unaffected parents

Rare dominant disease: $d = -a$

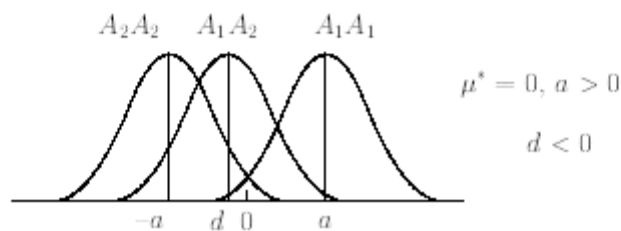
$$\alpha = 2pa, \sigma_a^2 = 8p^3qa^2, \sigma_d^2 = 4p^2q^2a^2$$

High inheritance $h^2 = 2\frac{p}{1+p} \approx 1$

affected offspring have one affected parent

Dependence on allele frequency

Phenotypic value distribution is a $(p^2, 2pq, q^2)$ mixture of three distributions for three genotypes



Draw two pdf curves for phenotypic value

- 1) at $p = \frac{1}{2}$ with negative $\mu = \frac{d}{2}$
- 2) at $p = \frac{1}{4}$ with positive $\mu = \frac{a}{2} + \frac{3d}{8}$

Fundamental theorem of natural selection:
the increase in average fitness at any time is the
additive genetic variance in fitness at that time

Literature:

1. D.L.Hartl, A.G.Clarc. Principle of population genetics. Sinauer Associates, 2007.
2. R.Nielson, M. Statkin. An introduction to population genetics: theory and applications, Sinauer Associates. 2013.