Chapter 9

Heritability: the nature and nurture of phenotypes

The heritability is precisely the quantity that answers the nature-nurture question.
J. Gillespie

9.1 Introduction

We have already emphasized that if we look at a population, different individuals may have different phenotypes. We have also emphasized that most of these phenotypes such as hair color, weight, blood pressure or allocation of resources to offspring are quantitative. So far, however, we have assumed that all this variation is genetic, since we assumed that phenotypes were entirely genetically determined. Full genetic determinism is unlikely in practice, as the environment also contributes to the phenotype of an individual. In fact, a fundamental question in evolutionary biology (and increasingly in the social sciences) involves comparing how much of the variation we observe in a population is due to genes to how much is due to environmental effects. In other words, how much phenotypic variation is due to nature (innate) and how much is due to nurture?

In order to address this question from a quantitative point of view, we will develop a model allowing us to assess the relative contribution of genes and environment to a phenotype. This model will also allow us to quantify the phenotypic resemblance between relatives.

9.2 Broad sense heritability

We denote by x_i the quantitative trait (or phenotype) carried by individual i (assumed diploid), and where i is an index running over all individuals in the population ($i = 1, 2, ..., n$). In general, the trait value $x_i \in \mathbb{R}$ depends on the individual's genotype and its environment. The relation between phenotype and genotype is called the *genotype-phenotype map*. This is generally unknown, likely to be extremely complicated, and so we seek to approximate this map (or function) in a statistical way.

The simplest model is to let the genotype and the environment of an individual determine its phenotype additively as

$$x_i = g_i + e_i. \tag{9.1}$$

Here, $g_i \in \mathbb{R}$ is the *genetic value* of individual i, namely the net contribution of all genetic factors to the phenotype and $e_i \in \mathbb{R}$ represents the net contribution of all environmental factors to the phenotype.

Eq. (9.1) is the standard additive model of quantitative genetics. From this, we will now express the average and variance in phenotype in the population in terms of genetic and environmental values. To that end, we denote the average of any quantity $z_i \in \{x_i, g_i, e_i\}$ in the population as

$$\bar{z} = \frac{1}{n}\sum_{i=1}^{n} z_i \tag{9.2}$$

and the variance as

$$V_{\mathrm{z}} = \frac{1}{n}\sum_{i=1}^{n}(z_i - \bar{z})^2. \tag{9.3}$$

The covariance between two variables, say between genetic (the g_i's) and the environmental values (the e_i's), will be denoted

$$\mathrm{Cov}[g_i, e_i] = \frac{1}{n}\sum_{i=1}^{n}(g_i - \bar{g})(e_i - \bar{e}), \tag{9.4}$$

where the right hand side correspond to the standard expression for the covariance between two variables; namely, the average of the product of their deviations from their expected values.

Taking the average of eq. (9.1) over population members then gives the mean in phenotype in the population as

$$\bar{x} = \bar{g} + \bar{e}, \tag{9.5}$$

where $\bar{g}$ is the mean genetic value and $\bar{e}$ is the mean environmental value. Owing to eq. (9.1) and the fact that the variance of a sum of variables is the sum of the variances plus the sum of all covariances, the variance in phenotype in the population is

$$V_{\mathrm{x}} = V_{\mathrm{g}} + V_{\mathrm{e}} + 2\,\mathrm{Cov}[g_i, e_i]\,, \tag{9.6}$$

which depends on the covariance $\mathrm{Cov}[g_i, e_i]$ between genetic effects and the environment (for instance, the association between genes and the place where an individual foragers may results in a positive covariance). This renders the phenotypic variance complicated and things are simpler if we could assume that the covariance between genetic and environmental values is zero,

$$\mathrm{Cov}[g_i, e_i] = 0. \tag{9.7}$$

Such a zero covariance between genetic and environmental contribution can be reached under two situations.

• For many traits there is no covariance between genetic effects and the environment, since the genes and the environment tend to be transmitted independently from one generation to the next.

• One can interpret the decomposition (eq. 9.1) as the linear regression of phenotype on genetic effects so that e_i is the "error" (or residual) in the prediction of phenotype given genetic values. Under this interpretation, eq. (9.7) necessarily applies as any genetic effect is absorbed into the regression. Hence, in a planned experiment $\mathrm{Cov}[g_i, e_i]$ can be controlled and set at 0.

For these reasons we assume throughout that $\mathrm{Cov}[g_i, e_i] = 0$, so the "environment" has to be understood as any environmental effect independent of the genes. The phenotypic variance (eq. 9.6) allow us to define the *broad sense heritability*

$$H^2 = \frac{V_{\mathrm{g}}}{V_{\mathrm{x}}} = \frac{V_{\mathrm{g}}}{V_{\mathrm{g}} + V_{\mathrm{e}}}. \tag{9.8}$$

This is the proportion of phenotypic variance in the population that is explained by the genetic values. If the heritability is low, then much of the differences between individuals in the population is explained by them having different environments. Conversely if the heritability is high, then the difference between individuals is explained by them having different genetic values, and thus different alleles. The broad sense heritability H^2 can be determined empirically, for instance, by analyzing the phenotypic correlation between monozygotic twins (which share the same genotype) raised in different environments.

If the broad sense heritability is high, say $H^2 = 0.9$, an individual's genetic value is strongly predicted by its phenotypic value, which means its phenotype is likely to be influenced by genes. Broad sense heritability $H^2 = V_{\mathrm{g}}/V_{\mathrm{x}}$ can thus be thought as giving the proportion of "nature" there is in the phenotypic variance in the population, while $1 - H^2 = V_{\mathrm{e}}/V_{\mathrm{x}}$ can be thought as the proportion of "nurture" (when eq. 9.7 holds). This is useful for predicting by how much the phenotype of a representative individual in the population would change if we were to change the environment to which that individual is exposed (say improve the schools). Suppose we change the environment of individual i from e_i to $e_i + \Delta e_i$, where Δe_i denotes the environmental change. Then, the change in the phenotype of individual i, denoted Δx_i, resulting from the environmental change and

holding g_i constant is, according to equation eq. (9.1), given by

$$\Delta x_i = \Delta e_i. \tag{9.9}$$

Suppose we were to change the environment by one unit of standard deviation, namely, $\Delta e_i = V_{\mathrm{e}}^{1/2}$, which is a typical environmental deviation from the mean environment $\bar{e}$ in the population. Then, the resulting phenotypic change is

$$\Delta x_i = V_{\mathrm{e}}^{1/2}, \tag{9.10}$$

where, from eq. (9.6) and assuming that eq. (9.7) holds, we can write

$$\begin{aligned} V_{\mathrm{e}} &= V_{\mathrm{x}} - V_{\mathrm{g}} \\ &= \left(1 - \frac{V_{\mathrm{g}}}{V_{\mathrm{x}}}\right) V_{\mathrm{x}} \\ &= \left(1 - H^2\right) V_{\mathrm{x}} \end{aligned} \tag{9.11}$$

Taking the square root of V_{e} and substituting into eq. (9.10) then produces

$$\Delta x_i = \left(1 - H^2\right)^{1/2} V_{\mathrm{x}}^{1/2}, \tag{9.12}$$

which shows that the phenotypic change resulting from a change in the environment is decreasing with an increase in heritability.

One point worth emphasizing about the broad sense heritability H^2, is that it is really a population concept. As such, a broad sense heritability of 0.6 does not say that a proportion 0.6 of the phenotype of an individual is determined by its genes and 0.4 by the environment, but that a proportion 0.6 of phenotypic variance in the population is explained by individuals have different alleles. Hence, H^2 is not an index of how much the phenotype of any given individual depends on its genotype.

9.3 Narrow sense heritability

9.3.1 Genetic value decomposed into additive and interaction effects

If genes matter in determining phenotype, we expect offspring to resemble their parents. If large parents tend to have large offspring, then the genetic value for "large size" tends to be inherited. One can then seek to predict the phenotypic relationship between parent and offspring. In order to do this, we need to decompose the genetic value g_i of individual i into two terms as

$$g_i = a_i + d_i, \tag{9.13}$$

where a_i is the *additive* effect of genes, which results from the separate contribution to the phenotype of an individual of paternally and maternally inherited genes, and d_i is the *interaction* effect of genes due to interactions between paternally inherited genes, interactions between maternally inherited genes, and interactions between paternally and maternally inherited genes. The additive effects are chosen such that they maximize their contribution to the genetic values (see section for more details 9.3.4) and to ensure they are uncorrelated to the interaction effects

$$\mathrm{Cov}[a_i, d_i] = 0. \tag{9.14}$$

According to this and eq. (9.13), the variance in genetic value V_g in the population can be expressed as

$$V_\mathrm{g} = V_\mathrm{a} + V_\mathrm{d}, \tag{9.15}$$

where V_a is called the additive genetic variance and V_d is the interaction variance. We can now define the ratio

$$h^2 = \frac{V_\mathrm{a}}{V_\mathrm{x}}. \tag{9.16}$$

This is the proportion of phenotypic variance in the population that is explained by the additive genetic values and is called the *narrow sense heritability*.

9.3.2 Resemblance between parent and offspring

The narrow sense heritability allows us to describe the phenotypic relationship between parent and offspring. To understand this, we now assume that the population is not evolving; there is no natural selection, no mutation or genetic drift. We also introduce the *midparent* phenotypic value

$$x_{\mathrm{mp}(i)} = \frac{x_{\mathrm{f}(i)} + x_{\mathrm{m}(i)}}{2}, \tag{9.17}$$

where $x_{\mathrm{f}(i)}$ is the phenotype of the mother (female) of i and $x_{\mathrm{m}(i)}$ is the phenotype of the father (male) of i. The narrow sense heritability h^2 turns out to be precisely the regression of offspring phenotype on midparent phenotype $x_{\mathrm{mp}(i)}$ when the environment of parents and offspring (during the development of the phenotype) is uncorrelated; namely the heritability is equal to

$$h^2 = \frac{\mathrm{Cov}\left[x_i, x_{\mathrm{mp}(i)}\right]}{\mathrm{Var}\left[x_{\mathrm{mp}(i)}\right]} \tag{9.18}$$

(see Derivation 1), where $\mathrm{Cov}\left[x_i, x_{\mathrm{mp}(i)}\right]$ is the covariance between the phenotype of an individual and the midparent phenotype, and $\mathrm{Var}\left[x_{\mathrm{mp}(i)}\right]$ is the variance in midparent phenotype. The expression for narrow sense heritability given by eq. (9.18) is useful because the phenotype of an individual can then be written as the linear regression on the midparent phenotype as

$$x_i = \bar{x} + h^2 \left(x_{\mathrm{mp}(i)} - \bar{x}\right) + \epsilon_{\mathrm{x},i}, \tag{9.19}$$

where $\epsilon_{\mathrm{x},i}$ is the least square residual error and $\bar{x}$ is the mean in parents and in offspring after reproduction. Hence, if one knows the phenotype of the parents of an individual, then

narrow sense heritability predicts the phenotype of offspring. If the heritability is high the resemblance between parent and offspring will be high. Since this resemblance is due to inherited genes, a high h^2 is another way of saying (in addition to high H^2) that genes affect phenotype.

Now, by substituting eq. (9.17) into eq. (9.19), we can write

$$x_i = \bar{x} + \frac{h^2}{2}\left(x_{\mathrm{f}(i)} - \bar{x}\right) + \frac{h^2}{2}\left(x_{\mathrm{m}(i)} - \bar{x}\right) + \epsilon_{\mathrm{x},i}, \tag{9.20}$$

which shows that the regression of the phenotype of an individual on that of a single parent (either mother or father) is $h^2/2$. Eq. (9.20) shows that the regression of the phenotype of an individual on the phenotype of a single parent, say the mother, is $h^2/2$. This is exactly the narrow sense heritability h^2 times the relatedness $1/2$ between the parent and the offspring (see eq. 8.1). Hence, the phenotypic resemblance between an offspring and any of its parents is $h^2/2$; namely, the heritability times the relatedness.

9.3.3 Resemblance between relatives

More generally, we may be interested in characterizing the resemblance between different pairs of individuals. To work this out, we denote by $x_{s(i)}$ the phenotype of an individual in role s in relation to individual i. A *role* is a possible situation an individual can occupy in relation to individual i. For instance, for interactions among families members, a role might include "mother", "sib", or "cousin". We can then write the regression of the phenotype of an individual in role s on the phenotype of individual i as

$$x_{s(i)} = \bar{x} + \beta_s\left(x_i - \bar{x}\right) + \epsilon_{\mathrm{r},i}, \tag{9.21}$$

where $\epsilon_{\mathrm{r},i}$ is the least square residual error in the prediction of phenotype and

$$\beta_s = \frac{\mathrm{Cov}\left[x_i, x_{s(i)}\right]}{\mathrm{Var}[x_i]} \tag{9.22}$$

is the regression coefficient of the phenotype of individual in role s on that of a focal individual.

If the environment and genetic interaction effects (the d_i's) between the pairs of relatives are independent, then the regression coefficient between an individual and another in s is

$$\beta_s = h^2 r_s, \tag{9.23}$$

where r_s is the relatedness between the two individuals. This thus generalizes the parent-offspring relation $h^2/2$ to other type of relatives. Importantly, offspring cannot inherit the genotype of a parent (they only inherit one gene) so that in a random mating population parent and offspring cannot have a genotype identical-by-descent, the can only have a single gene identical-by-descent. But other type of relatives, like full sibling may share a genotype identical-by-descent, and in this case additional terms need to take into account in

the phenotypic regression β_s as individuals may share interactions effects (due to common ancestry of full genotypes), so that eq. (9.23) is only an approximation for the phenotypic resemblance between non parent-offspring pairs of relatives.

9.3.4 Evaluating the additive genetic effects

We said that the additive effects in eq. (9.1) are chosen so as to maximize their contributions to the genetic values and ensuring they are uncorrelated to the interaction effects. This is, maybe, not a very explicit description. In order to better understand what this means and implies, we provide an example of this maximization procedure when only one gene with two alleles contribute to the phenotype of an individual in a diploid panmictic population. This section is not easy and presented for completeness.

We consider, as usual, a A allele with frequency p and a B allele with frequency $1-p$. The genetic values of the three genotypes AA, AB, and BB, as, respectively, denoted by g_{AA}, g_{AB} and g_{BB}. With this the phenotype of an individual, say a homozygote AA, is determined according to its genetic value g_{AA}, and some environmental value, and this may be different for each individual carrying the AA genotype. We assume that this environmental value is uncorrelated to genetic effects ($\mathrm{Cov}[g_i, e_i] = 0$, eq. 9.7) and has zero mean. Hence, the average phenotype x_{AA} of a homozygote AA can be taken to be directly given by the genotypic values g_{AA} so that

Genotype:	AA	AB	BB
Average phenotype:	$x_{\mathrm{AA}} = g_{\mathrm{AA}}$	$x_{\mathrm{AB}} = g_{\mathrm{AB}}$	$x_{\mathrm{BB}} = g_{\mathrm{BB}}$

Following eq. (9.13), we now decompose the genetic values of each type of individual in terms of additive and interaction effect as

$$g_{\mathrm{AA}} = 2a_{\mathrm{A}} + d_{\mathrm{AA}}, \qquad g_{\mathrm{AB}} = a_{\mathrm{A}} + a_{\mathrm{B}} + d_{\mathrm{AB}} \quad \text{and} \quad g_{\mathrm{BB}} = 2a_{\mathrm{B}} + d_{\mathrm{BB}}, \tag{9.24}$$

where a_{A} and a_{B} are the additive genetic values stemming from carrying one copy of allele A and B, respectively and the d_j's (for $j \in \{\mathrm{AA}, \mathrm{AB}, \mathrm{BB}\}$) are the interaction effects.

Because we have only one gene determining the phenotype, the interaction effects actually measure dominance effects stemming from the interaction of the paternally and maternally inherited alleles in an individual (recall eqs. 7.1–7.3 for the concept of dominance). Note that because the additive values a_{A} and a_{B} are ascribed to genes, they are transmitted from parent to offspring, while the d_j's are not, since they depend on exactly which allele an individual inherits from its two parents. The various components of the genetic values are summarized in the following table along with the frequencies of the various genotypes.

Genotype:	AA	AB	BB
Probability:	p^2	$2p(1-p)$	$(1-p)^2$
Genetic value:	$g_{\rm AA}$	$g_{\rm AB}$	$g_{\rm BB}$
Additive genetic value:	$2a_{\rm A}$	$a_{\rm A}+a_{\rm B}$	$2a_{\rm B}$
Dominance value:	$d_{\rm AA}$	$d_{\rm AB}$	$d_{\rm BB}$

From the elements of this table the mean genetic and additive values are given by

$$\begin{aligned}\bar{g} &= g_{\rm AA}p^2 + g_{\rm AB}2p(1-p) + g_{\rm BB}(1-p)^2 \\ \bar{a} &= 2a_{\rm A}p^2 + (a_{\rm A}+a_{\rm B})2p(1-p) + 2a_{\rm B}(1-p)^2 \end{aligned} \tag{9.25}$$

Likewise we can compute the genetic variance $V_{\rm g}$, the additive genetic variance $V_{\rm a}$, and the genetic variance due to interactions $V_{\rm d}$. For instance, the additive genetic variance is

$$V_{\rm a} = \left(2a_{\rm A} - \bar{a}\right)^2 p^2 + \left(a_{\rm A} + a_{\rm B} - \bar{a}\right)^2 2p(1-p) + \left(2a_{\rm B} - \bar{a}\right)^2 (1-p)^2. \tag{9.26}$$

In order for the additive values $a_{\rm A}$ and $a_{\rm B}$ to contribute the most to the genetic values and for them to be uncorrelated to the dominance effects, two features should hold. First, that the average additive value should be equal to the average genetic value

$$\bar{g} = \bar{a}, \tag{9.27}$$

which implies that the mean dominance value is zero $\bar{d} = 0$ (from eq. 9.13), otherwise the average additive values would represent a biased description of the average genetic value. Second, that the additive variance $V_{\rm a}$ contributes the most to the $V_{\rm g}$ variance. In other words, we want to minimize the contribution of the dominance variance $V_{\rm d}$ by fitting the $a_{\rm A}$ and $a_{\rm B}$ values.

When $\bar{d} = 0$, the dominance variance is given by

$$V_{\rm d} = d_{\rm AA}^2 p^2 + d_{\rm AB}^2 2p(1-p) + d_{\rm BB}^2 (1-p)^2, \tag{9.28}$$

and on substituting eq. (9.24), we have

$$V_{\rm d} = (g_{\rm AA} - 2a_{\rm A})^2 p^2 + (g_{\rm AB} - (a_{\rm A}+a_{\rm B}))^2 2p(1-p) + (g_{\rm BB} - 2a_{\rm B})^2 (1-p)^2. \tag{9.29}$$

We can now minimize this expression to derive the additive effects, whereby

$$\begin{aligned}\frac{\partial V_{\rm d}}{\partial a_{\rm A}} &= -4\left[(g_{\rm AA} - 2a_{\rm A})p^2 + (g_{\rm AB} - (a_{\rm A}+a_{\rm B}))p(1-p)\right] = 0 \\ \frac{\partial V_{\rm d}}{\partial a_{\rm B}} &= -4\left[(g_{\rm BB} - 2a_{\rm B}))(1-p)^2 + (g_{\rm AB} - (a_{\rm A}+a_{\rm B}))p(1-p)\right] = 0. \end{aligned} \tag{9.30}$$

Solving for $a_{\rm A}$ and $a_{\rm B}$ then produces the additive effects

$$\begin{aligned} a_{\rm A} &= \frac{1}{2}\left(g_{\rm AA} - 2(1-p)^2\left(\frac{g_{\rm AA}+g_{\rm BB}}{2} - g_{\rm AB}\right)\right) \\ a_{\rm B} &= \frac{1}{2}\left(g_{\rm BB} - 2p^2\left(\frac{g_{\rm AA}+g_{\rm BB}}{2} - g_{\rm AB}\right)\right), \end{aligned} \tag{9.31}$$

and on substitution into eq. (9.26) and eq. (9.28) produces the variances

$$\begin{aligned} V_{\mathrm{a}} &= 2p(1-p)\left[g_{\mathrm{AB}} - g_{\mathrm{AA}} + 2p\left(\frac{g_{\mathrm{AA}} + g_{\mathrm{BB}}}{2} - g_{\mathrm{AB}}\right)\right]^2 \\ V_{\mathrm{d}} &= p^2(1-p)^2 4\left(\frac{g_{\mathrm{AA}} + g_{\mathrm{BB}}}{2} - g_{\mathrm{AB}}\right)^2. \end{aligned} \tag{9.32}$$

These are expressed in terms of genetic values and allele frequency and allow us to compute heritability h^2 explicitly.

The so obtained additive values thus entails a sum of square minimization (eq. 9.30). This shows that the narrow sense heritability follows from a subtle computation. From a biological point of view the idea is to explain as much as is possible of the genetic variance in terms of additive genetic effects, which can be ascribed to genes, since it is genes and not genotypes that are transmitted from parent to offspring. Hence, it is the narrow sense heritability h^2 (and not the broad sense heritability H^2) that (i) describes the resemblance between parent and offspring and (ii) plays a fundamental role in describing how quantitative traits, that depend on many underlying genes and possibly environmental effects, change under the action of natural selection, which is the topic of the next and last section.

9.4 Selection on quantitative phenotypes

We now consider selection on a quantitative trait that is influenced by many genes. Our aim is to find an expression for the change $\Delta\bar{x} = \bar{x}' - \bar{x}$ of the average phenotype and prove the equation $\Delta\bar{x} = V_{\mathrm{a}}S(\bar{x})$ given in the lectures slides.

To evaluate $\Delta\bar{x} = \bar{x}' - \bar{x}$, we need a expression for the average phenotype $\bar{x}'$ in the offspring generation, and this can be written as

$$x' = \sum_{i=1}^{n} \frac{w_i}{\bar{w}n} x_{\mathrm{o}(i)}. \tag{9.33}$$

Here, w_i is the fitness of individual i per gene number (or ploidy, like in section 6.3.2) and $w_i/(\bar{w}n)$ is the fraction of individuals in the descendant generation that are offspring of individual i, and $x_{\mathrm{o}(i)}$ is the average phenotype among all offspring of individual i. Denoting by $\Delta x_i = x_{\mathrm{o}(i)} - x_i$ the difference in the phenotype between individual i and its average offspring, we have from eq. (9.33) that

$$\begin{aligned} \Delta\bar{x} &= \sum_{i=1}^{n} \frac{w_i}{\bar{w}n} x_i + \sum_{i=1}^{n} \frac{w_i}{\bar{w}n} \Delta x_i - \bar{x} \\ &= \frac{1}{n\bar{w}} \sum_{i=1}^{n} w_i x_i - \frac{\bar{w}}{\bar{w}}\bar{x} + \frac{1}{n\bar{w}} \sum_{i=1}^{n} w_i \Delta x_i \\ &= \frac{1}{n\bar{w}} \sum_{i=1}^{n} x_i \left(w_i - \bar{w}\right) + \frac{1}{n\bar{w}} \sum_{i=1}^{n} w_i \Delta x_i. \end{aligned} \tag{9.34}$$

Using the definitions of covariance (recall eqs. 9.2–9.4) and writing the average of the change in phenotype between parent and offspring as

$$\mathrm{E}[w_i \Delta x_i] = \frac{1}{n} \sum_{i=1}^{n} w_i \Delta x_i, \tag{9.35}$$

we have

$$\Delta \bar{x} = \frac{\mathrm{Cov}(w_i, x_i)}{\bar{w}} + \frac{\mathrm{E}[w_i \Delta x_i]}{\bar{w}}. \tag{9.36}$$

This equation partitions the change in phenotype into two terms.

• The change in phenotype due solely to natural selection, which depends on the covariance between fitness and trait value.

• The change due to any other factor, like mutation or recombination. This is sometimes called the change due to transmission.

This equation for the change in phenotype (eq. 9.36) is called the *Price equation*, as it was originally derived by George R. Price. It describes the total evolutionary change; namely, the dynamics of any phenotype can be put into this simple form, regardless of the biological complexity of the situation. It also shows that the change in phenotype depends only on natural selection if a phenotype is faithfully transmitted from a parent to its offspring (i.e., if $\mathrm{E}[w_i \Delta x_i] = 0$).

The Price equation does in itself not allow to predict long-term evolutionary dynamics (e.g., which strategy will be uninvadable) as it only describes the change of phenotype over a single generation. To reach more definite results we need to make more specific assumptions. In particular, we now write the phenotype of individual i by following the decomposition of the last section as

$$x_i = a_i + d_i \tag{9.37}$$

were a_i is the additive genetic effect (the best linear fit of genetic values when alleles within individual i are used as predictors of phenotypes, see section 8.3.4), d_i is the genetic deviation from additivity, and, without loss of generality, we have set the environmental effect e_i to zero (any environmental effects can be thought to be absorbed into the deviation d_i from genetic additivity). This entails that $\mathrm{Cov}(a_i, d_i) = 0$ and $\bar{d} = 0$ (e.g, eq. 9.27). Thus, the average phenotype in the population can be written as

$$\bar{x} = \bar{a}. \tag{9.38}$$

With this, eq. (9.36) implies that the change in the average phenotype in the population can be written as

$$\Delta \bar{x} = \frac{\mathrm{Cov}(w_i, a_i)}{\bar{w}} + \frac{\mathrm{E}[w_i \Delta a_i]}{\bar{w}}. \tag{9.39}$$

This shows that what really matters for genetic evolution by natural selection is the association between the heritable part of the phenotype and fitness. Now, $\mathrm{E}[w_i \Delta a_i]$ is the change in the additive effects when genetic transmission occurs between a parent and its

offspring. Since on average offspring inherit gametes that have the same allele frequencies as in parents, offspring inherit (on average and in the absence of mutation) the additive gene effects of their parents. Hence, on average, we have

$$\mathrm{E}[w_i \Delta a_i] = 0 \tag{9.40}$$

(see Derivation 2 for a more formal proof).

We now further assume that selection is weak so that the fitness of individual i can be expressed in the linear form

$$w_i = 1 + S(\bar{x})(x_i - \bar{x}) + \text{higher order terms}, \tag{9.41}$$

where $S(\bar{x})$ is the selection gradient evaluated at the mean population value (this is the same function as in the chapters on adaptive dynamics, eq. 6.6, and kin selection, eq. 8.9, with the difference that now each individual may have a different phenotype, while in the previous chapters we considered the special case where we considered only a monomorphic resident population, whereby we had $\bar{x} = x$).

Substituting eqs. (9.41) into eq (9.39) and using $\mathrm{Cov}(a_i, d_i) = 0$, then, on average, the average change in phenotype in a population evolving at a demographic equilibrium ($\bar{w} \approx 1$) is

$$\Delta \bar{x} = V_{\mathrm{a}} S(\bar{x}) + \text{higher order terms}, \tag{9.42}$$

This is a key result: the change in a complex phenotype (being morphological, physiological or behavioral), possibly depending on an untold number of interacting genes, is to the first order proportional to the additive genetic variance in the phenotype and the selection gradient.

9.5 Appendix

Derivation 1. We here prove eq. (9.18). Using the expressions for the decomposition of phenotypic and genotypic value (eq. 9.1 and eq. 9.13), we can write the phenotype of individual i as

$$x_i = \underbrace{a_i + d_i}_{g_i} + e_i. \tag{9.43}$$

This can also be written as

$$x_i = \frac{1}{2} a_{\mathrm{f}(i)} + \frac{1}{2} a_{\mathrm{m}(i)} + d_i + e_i, \tag{9.44}$$

where $a_{\mathrm{f}(i)}$ is the additive genetic value in the mother of individual i and $a_{\mathrm{f}(i)}$ is the additive genetic value in the father of i. The phenotypes of the mother and the father are, respectively,

$$\begin{aligned} x_{\mathrm{f}(i)} &= a_{\mathrm{f}(i)} + d_{\mathrm{f}(i)} + e_{\mathrm{f}(i)} \\ x_{\mathrm{m}(i)} &= a_{\mathrm{m}(i)} + d_{\mathrm{m}(i)} + e_{\mathrm{m}(i)}, \end{aligned} \tag{9.45}$$

whereby the mid-parent phenotypic value (eq. 9.17) is

$$x_{\mathrm{mp}(i)} = \underbrace{\frac{a_{\mathrm{f}(i)} + d_{\mathrm{f}(i)} + a_{\mathrm{m}(i)} + d_{\mathrm{m}(i)}}{2}}_{g_{\mathrm{mp}(i)}} + \frac{e_{\mathrm{f}(i)} + e_{\mathrm{m}(i)}}{2}, \tag{9.46}$$

where $g_{\mathrm{mp}(i)}$ is the mid-parent genetic value.

When there are no covariances between genetic and environmental values within individuals, and the environmental values of a parent and its offspring ($\mathrm{Cov}[g_i, e_i] = \mathrm{Cov}\left[e_i, e_{\mathrm{f}(i)}\right] = \mathrm{Cov}\left[e_i, e_{\mathrm{m}(i)}\right] = 0$), we have

$$\mathrm{Cov}\left[x_i, x_{\mathrm{mp}(i)}\right] = \mathrm{Cov}\left[g_i, g_{\mathrm{mp}(i)}\right]. \tag{9.47}$$

Further, there is no covariance between additive and interaction effects ($\mathrm{Cov}_{\mathrm{I}}[a_i, d_i] = 0$, eq. 9.14), and no covariance between the interaction effects of parent and offspring

$$\mathrm{Cov}\left[d_i, d_{\mathrm{f}(i)}\right] = \mathrm{Cov}\left[d_i, d_{\mathrm{m}(i)}\right] = 0 \tag{9.48}$$

This stems from the fact that under random random mating offffspring cannot inherit the interaction effect from their parents. Hence, using eq. (9.46) we have

$$\begin{aligned} \mathrm{Cov}\left[g_i, g_{\mathrm{mp}(i)}\right] &= \mathrm{Cov}\left[\frac{a_{\mathrm{f}(i)} + a_{\mathrm{m}(i)}}{2} + d_i, \frac{a_{\mathrm{f}(i)} + a_{\mathrm{m}(i)}}{2} + \frac{d_{\mathrm{f}(i)} + d_{\mathrm{m}(i)}}{2}\right] \\ &= \frac{1}{4}\mathrm{Var}\left[a_{\mathrm{f}(i)}\right] + \frac{1}{4}\mathrm{Var}\left[a_{\mathrm{m}(i)}\right], \end{aligned} \tag{9.49}$$

since $\mathrm{Cov}\left[a_{\mathrm{f}(i)}, a_{\mathrm{f}(i)}\right] = \mathrm{Var}\left[a_{\mathrm{f}(i)}\right]$, $\mathrm{Cov}\left[a_{\mathrm{m}(i)}, a_{\mathrm{m}(i)}\right] = \mathrm{Var}\left[a_{\mathrm{m}(i)}\right]$, the other covariances are zero.

Finally, owing to the fact that the phenotype of the mother and father of i are independent, we have, by using eq. (9.17), that

$$\mathrm{Var}\left[x_{\mathrm{mp}(i)}\right] = \frac{1}{4}\mathrm{Var}\left[x_{\mathrm{f}(i)}\right] + \frac{1}{4}\mathrm{Var}\left[x_{\mathrm{m}(i)}\right]. \tag{9.50}$$

In a panmictic population, the variances in males and females are the same and assuming they don't change over time (not natural selection, drift, or mutation), we can write

$$\begin{aligned} 2V_{\mathrm{a}} &= \mathrm{Var}\left[a_{\mathrm{f}(i)}\right] = \mathrm{Var}\left[a_{\mathrm{m}(i)}\right] \\ 2V_{\mathrm{x}} &= \mathrm{Var}\left[x_{\mathrm{f}(i)}\right] = \mathrm{Var}\left[x_{\mathrm{m}(i)}\right]. \end{aligned} \tag{9.51}$$

The reason for the factor 2 is that, when we evaluate $\mathrm{Var}\left[a_{\mathrm{f}(i)}\right]$, we are taking the variance (average over the squared deviation) over all individuals in the offspring generation, thus counting twice the contribution to the variance of any parent (since each offspring has two parents so that on average every parent has two offspring). Hence, both the additive genetic variance of mothers and fathers of all offspring is equal to twice the additive variance ($\mathrm{Var}\left[a_{\mathrm{f}(i)}\right] = 2V_{\mathrm{a}}$, $\mathrm{Var}\left[a_{\mathrm{m}(i)}\right] = 2V_{\mathrm{a}}$), and the same argument applies for the phenotypic variance. With this and using eqs. (9.47)-(9.51) we can write

$$\frac{\mathrm{Cov}\left[x_i, x_{\mathrm{mp}(i)}\right]}{\mathrm{Var}\left[x_{\mathrm{mp}(i)}\right]} = \frac{\mathrm{Cov}\left[g_i, g_{\mathrm{mp}(i)}\right]}{\mathrm{Var}\left[x_{\mathrm{mp}(i)}\right]} = \frac{V_{\mathrm{a}}}{V_{\mathrm{x}}} = h^2, \tag{9.52}$$

which is precisely the heritability in the population.

Derivation 2. We here prove that $\mathrm{E}[w_i \Delta a_i] = 0$ holds on average, where $\Delta a_i = a_{\mathrm{o}(i)} - a_i$. To that end, we need an explicit expression for the average additive genetic effect $a_{\mathrm{o}(i)}$ of all offspring of individual i. This can be written as

$$a_{\mathrm{o}(i)} = \frac{1}{2w_i} \sum_{j \in I'} \sum_{k \in I} A_{j,ik} a_{j,ik}, \tag{9.53}$$

Here, $A_{j,ik} \in \{0, 1\}$ takes the value one when individual $j \in I'$ from the descendant generation has as its genetic ancestors individual $i \in I$ and individual $k \in I$ of the parental generation, zero otherwise, and $a_{j,ik}$ is the additive genetic value of that individual (here I' denotes the set of individuals in the offspring generation and I denotes the set of individuals in the parental generation). With this notation, the total number of offspring that individual i is an ancestor of is

$$\sum_{j \in I'} \sum_{k \in I} A_{j,ik} = 2w_i, \tag{9.54}$$

where w_i is the expected number of successful offspring *per ploidy* of individual i.

Following the three way decomposition of phenotype (eq. 9.13), we write the phenotype of individual j produced by parent i and k as

$$a_{j,ik} = \frac{1}{2}(a_i + s_{ji}) + \frac{1}{2}(a_k + s_{jk}), \tag{9.55}$$

where a_h is the breeding value of individual h, s_{hj} is a deviation due to segregation during the formation of the gamete inherited by j so that $a_h + s_{hj}$ gives the transmitted additive genetic effect of individual i to j. Substituting eq. (9.55) into eq. (9.53), we can write the average change in additive genetic value due to transmission as

$$\begin{aligned}
\mathrm{E}[w_i \Delta a_i] &= \frac{1}{n} \sum_{i \in I} w_i \Delta a_i \\
&= \frac{1}{n} \sum_{i \in I} w_i \left(z_{\mathrm{o}(i)} - a_i \right) \\
&= \frac{1}{n} \left(\frac{1}{2} \sum_{j \in I'} \sum_{i \in I} \sum_{k \in I} A_{j,ik} a_{j,ik} - \sum_{i \in I} w_i a_i \right) \\
&= \frac{1}{n} \left(\frac{1}{2} \sum_{i \in I} w_i a_i + \frac{1}{2} \sum_{k \in I} w_k a_k - \sum_{i \in I} w_i a_i \right) + \Lambda_{\mathrm{a}} \\
&= \Lambda_{\mathrm{a}}
\end{aligned} \tag{9.56}$$

where

$$\Lambda_{\mathrm{a}} = \frac{1}{2n} \sum_{i \in I} \sum_{j \in I'} \sum_{k \in I} A_{j,ik} \left(\frac{1}{2} s_{ji} + \frac{1}{2} s_{jk} \right). \tag{9.57}$$

is the average deviation of the additive genetic value in the offspring generation due to the segregation of parental alleles (meiosis). If we take the expectation over the (stochastic) segregation process we have $\mathrm{E}[s_{ji}] = 0$ and so $\mathrm{E}[\Lambda_{\mathrm{a}}] = 0$. This thus

holds on average in a finite population. In a very large population it implies that we actually have $\Lambda_a = 0$, as the deviations will cancel out, owing to the law of large numbers.