Adaptation and Fitness in Animal Populations

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Evolutionary and Breeding Perspectives on Genetic Resource Management

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Emeritus Professor James Stuart Flinton Barker in honour of his contributions to Population Genetics and Animal Breeding

Preface

At the 16th AAABG conference in 2005, a proposal was launched to organise a symposium to examine advances in understanding of "adaptive fitness, both in managed populations being conserved and domestic animal species being utilised for food and agriculture production". After discussion about the term "adaptive fitness" some of us decided we should organise a symposium "Adaptation and Fitness in Animal Populations – Evolutionary and breeding perspectives on genetic resource management" to be held at the 2007 AAABG meeting in Armidale.

The term 'adaptive fitness' in itself is somewhat a tautology, but fitness and adaptation are both relevant concepts when one is concerned with the long term sustainability of animal breeding programs and animal production systems in a broader sense. Fitness of farm animal populations is clearly becoming a concern, especially in programs that have achieved substantial genetic change for 'production traits' or where environmental stressors are intense. Adaptation is required to maintain fitness in new or changing environments. Adaptive mechanisms are very important in animal production systems where genotypes are used globally without being explicitly tested in all the environments where they are kept. Adaptability is also important within animal production systems experiencing large between-year variability in the environment. Understanding such mechanisms and their relationships with the production traits may help to more successfully realise sustainable productivity gains.

Evolutionary geneticists are continuously exploring the genetic mechanisms that surround fitness and adaptation in natural populations. Consequently it seemed appropriate to have a debate among animal geneticists and evolutionary biologists. The chapters from this book are a reflection of the symposium papers that were presented in September 2007 in Armidale. The book is divided into four sections, corresponding to the different sessions that we identified for the symposium. Each of these sessions addressed specific questions that we had posed before the symposium. We asked the chair of each session to summarise the discussion and this discussion summary completes the set of papers for each section of the book.

Prof. Stuart Barker's primary life's work has been directed at understanding fitness and the evolution of natural populations. He begins the book by reviewing developments over time in the definition of fitness and its components. This assisted us in arriving at a uniform set of terms for use in addressing these components.

Animal breeders have clearly been very successful in achieving selection goals. In high-input production systems in particular, dairy cows are now much more productive, sheep produce more and better wool, and cattle, broilers, and pigs grow faster and have improved meat quality. Fitness, in the sense of general health, wellbeing and reproductive ability, is commonly perceived to have declined in farm animal populations particularly of developed country production systems. Why? One straightforward model is to consider fitness as just another quantitative trait. There appears to be a negative genetic correlation with production traits and if fitness as such is not selected for, one would expect a decline. The problem could then be resolved by simply measuring fitness and including it in the selection objective. However, fitness appears to encompass complexes of characters – do these fitness characters behave like other quantitative traits? And why is the correlation with production traits negative? Can we trust that the simple quantitative genetic model will work? At this symposium, Prof. Dick Frankham explores the genetic architecture of fitness characters and shed some light on those questions. Prof. Mike Goddard discusses the same issues in the context of animal breeding programs and makes suggestions for accommodating fitness in breeding objectives.

When considering long term consequences of selection (in the farm animal context more than 10–20 generations), the main question is how genetic variation can be maintained as well as exploited, particularly for fitness traits. We are somewhat surprised by the amount of genetic variation that is still observed for traits after many generations of artificial selection. In natural populations, considerable variation exists for fitness even after millions of years of natural selection. Prof. Bill Hill and Dr. Xu-Sheng Zhang review the mechanisms that determine genetic variation and the implications for maintenance of variation in long term (artificial) selection programs. Prof. Mark Blows and Prof. Bruce Walsh discuss the constraints to selection response and adaptation, and explore whether within a multi-dimensional selection space all points are equally attainable. Dr. Piter Bijma addresses the question of how to best utilise genetic variation for fitness traits in selection programs. A relatively new approach in animal breeding is the use of a genetic model that includes effects of interactions between individuals within populations. Such models can be used to select against aggressiveness in livestock populations, an aspect that is clearly linked to fitness and animal welfare.

Adaptation is a key principle for individuals to survive in the environment they are managed in. With the possibility of environmental changes, e.g. due to global warming and perhaps increased variability of climate events, the ability to adapt to such changes has become a key issue. Dr. Kathleen Donohue discusses the ability of organisms to affect, or rather determine their environment, and how this can form a basis for genotype by environment interaction and how it can determine response to selection for phenotypic performance in different environments. Prof. John James presents principles and examples that are used in animal breeding literature to model genotype by environment interaction and interprets the meaning of different modelling approaches. An important question is how to assess fitness in different environments In *Drosophila*, fitness has been measured extensively under laboratory conditions, and Prof. Ary Hoffman explores to what extent such assessments can be extrapolated to natural conditions.

Maintaining enough variation and diversity in genetic resources is an essential component of animal breeding programs as it is a key for maintaining fitness. Prof. Brian Kinghorn et al. give an overview of the concepts and proposed strategies for maintaining genetic diversity in breeding programs. The objective of directional selection for increased merit needs to be balanced with the objective to maintain diversity. International bodies such as FAO have made considerable investments in developing strategies to assist the countries of the world to better manage their farm animal genetic resources. Dr. Louis Ollivier and Dr. Jean-Louis Foulley review measures of between and within population diversity, demonstrating also the strengths and weaknesses of this information when trying to establish conservation tactics and population adaptability. Prof. John Gibson gives an outline on the possible roles for the different institutional entities that have a responsibility to implement sustainable utilisation and conservation policies for farm animal genetic resources.

Finally, to celebrate the contributions over 55 years by Professor Stuart Barker to the research, development and teaching of population and quantitative genetics, we dedicated the symposium to him. Stuart's passion is "genetics". Fitness and adaptation have been important keywords in his research career. He has been instrumental in translating these fundamental concepts to applications in animal breeding and to the characterisation and conservation of genetic resources. This symposium brought together scientists in evolutionary genetics and animal breeding in the hope that lessons can be learned about the relevance of fitness in domesticated breeding populations and how fundamental questions about fitness and adaptation can be addressed through experimental results and observations in natural populations. Stuart has always been a strong advocate of such interactions and this book creates a unique opportunity for mutual benefits for both groups.

We thank all authors of this book for their contributions and for reviewing the contributions of their colleagues. We thank the chairpersons of the symposium for summarising the session discussions for this book. We thank Dr. Keith Hammond for raising the original idea of the symposium and the book, and his tireless efforts in keeping us on track in many ways. In relation to the program, we thank Professor Stuart Barker for his succinct and effective suggestions, Professor Frank Nicholas for his cheerful and thoughtful contributions and Professor Bill Hill for his advice on topics and speakers. We are very grateful to Kathryn Dobos who did a tremendous and meticulous job in fixing up errors in the 'final version' of the manuscript. Finally we acknowledge the financial support of the Federal Department of Agriculture, Fisheries and Forestry (DAFF) and Meat and Livestock Australia (MLA).

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Part I
Modelling Fitness

Defining Fitness in Natural and Domesticated Populations

James S.F. (Stuart) Barker

Abstract The term 'fitness' has been applied differently and with different definitions throughout the history of population genetics. Five concepts and definitions are presented – distinguishing (phenotypic) fitness, (genotypic) fitness, adaptedness, adaptability and durability. While the heritabilities of fitness components are low, substantial genetic change is achievable, and breeding programs should include in the breeding objectives genotypic fitness (for known QTLs) and fitness traits such as fertility and longevity, as well as production traits.

Keywords Fitness · net fitness · adaptedness · adaptability · durability

1 Introduction

Some 47 years ago, I had the temerity in the introduction to my PhD thesis (Barker 1960) to discuss the meaning of fitness. Now I am asked to come back to this question, and to consider if the concept is or should be applied differently in natural and domesticated populations.

To go back to the beginning, the concept of fitness derives from Darwin's adoption of Herbert Spencer's (1864) term 'survival of the fittest' as an alternative to 'natural selection'. Dawkins (1982, p. 179) states that Darwin adopted 'survival of the fittest' in a letter to Wallace in 1866. However, the term first appears in the various editions of 'The Origin of Species' in 1869 (5th edition), in the first paragraph of Chapter 4:

> ...Let it also be borne in mind how infinitely complex and close-fitting are the mutual relations of all organic beings to each other and to their physical conditions of life; and consequently what infinitely varied diversities of structure may be of use to each being under changing conditions of life. Can it, then, be thought improbable, seeing that variations useful to man have undoubtedly occurred, that other variations useful in some way to each being

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> in the great and complex battle of life, should sometimes occur in the course of thousands of generations? If such do occur, can we doubt (remembering that many more individuals are born than can possibly survive) that individuals having any advantage, however slight, over others, would have the best chance of surviving and of procreating their kind? On the other hand, we may feel sure that any variation in the least degree injurious would be rigidly destroyed. This preservation of favourable variations, and the destruction of injurious variations, I call Natural Selection, or the Survival of the Fittest.

But it is appropriate in this Symposium to note that the prior and first use by Darwin was in the first edition of 'The Variation of Animals and Plants under Domestication' (Darwin 1868).

Fitness may refer to a genotype, an individual, a population or a species, and defining fitness has plagued evolutionary biologists for many years, partly because the term has been applied differently and with different definitions depending on which level in the hierarchy is being considered.

In the 1920s and 1930s, the theoretical foundations of population genetics were laid by Fisher, Haldane and Wright, primarily considering selection within populations and thus the relative fitness of genotypes. In his book *The Causes of Evolution*, Haldane (1932), entitled one chapter 'What is fitness?', but provided no answer (perhaps consciously and wisely for that time!). Twenty years later, Dobzhansky (1955) pointed out that 'the work in this field (*population genetics*) is severely handicapped by lack of reliable methods of comparing the fitness of populations and, of course, by the lack of clarity in the concept of fitness itself.' (italics added).

As noted here by Dobzhansky, there are two interrelated problems – defining fitness and measuring fitness.

Broadly speaking, fitness relates to the ability of organisms to pass on the genes they carry. But to illustrate the problems of definition of fitness, consider a sample:

- *The relative ability of an organism to survive and transmit its genes to the next generation (King and Stansfield 1990)*,
- *The relative capacity of carriers of a given genotype to transmit their genes to the gene pool of the following generations constitutes the adaptive value, or the Darwinian fitness, of that genotype. The adaptive value is, then, a statistical concept which epitomizes the reproductive efficiency of a genotype in a certain environment (Dobzhansky 1951, p. 78)*,
- *...relative fitness (is) the relative ability of different genotypes to pass on their alleles to future generations (Hedrick 1983, p. 120)*,
- *Relative fitness: the fitness of a genotype, compared to another genotype, usually at the same locus (Frankham et al. 2002, p. 543)*,
- *Reproductive fitness: the number of fertile offspring surviving to reproductive age contributed by an individual. Characters that contribute to fitness include male fertility, female fecundity, parental care, offspring survival and offspring fertility. Often referred to as fitness (Frankham et al. 2002, p. 543)*,
- *(A numerical measure of) ability to survive and reproduce in a particular environment (Brown 1993)*,

- *The probability that a unit of evolution (population) will survive for a given long period of time, such as 10^8 years, that is to say will leave descendants after the lapse of that time, is the fitness of the unit (Thoday 1953, 1958).*

Many more examples could be quoted, but these show the main variations – genotype, individual or population; one, a few or many generations; recognition of the environment or not. Dawkins (1982), Hedrick and Murray (1983) and Endler (1986) provide (from different perspectives) excellent discussions of the various ways in which the term fitness has been used, and their context (genes, genotypes, individuals, groups). Nevertheless, some of the examples above show that more than 20 years on, there are still varying definitions.

2 Fitness and Related Concepts

Fitness differences among individuals (phenotypes) are necessary for selection to lead to change in a population – whether this be selection in natural populations, or artificial selection in the laboratory or animal breeding, where some individuals are culled on phenotypic criteria and thus have a fitness of zero. Such selection will cause a change in the distribution of phenotypes *within* a generation, but will cause a changed distribution of phenotypes in the next generation only if the fitness differences are, at least partly, genetic. Failure to distinguish these two phases has been one factor contributing to confusion in defining fitness. In fact, the problem is deeper than that because, depending on context, we operate at the genetic level or at the phenotypic level, and we do not have the knowledge to transform from one to the other (Lewontin 1974). This deficiency in our understanding of the transformation from genotype to phenotype needs to be clearly recognised in relation to much of the current work attempting to identify quantitative trait loci (QTLs). A QTL found in one population in one environment may not have the same phenotypic expression in a different genetic background or in a different environment.

At the genetic level (population genetics), generation transitions are expressed in terms of change in allele frequency (Δq) of an allele at a locus after one generation:

$$\Delta q = \frac{q(1-q)}{2}\frac{\mathrm{d}\ln \bar{w}}{\mathrm{d}q} \tag{1}$$

where q is the current allele frequency and $\bar{w}$ is the mean fitness of the genotypes at this locus in the population.

At the phenotypic level (quantitative genetics), generation transitions (assuming directional selection) are expressed in terms of changes in mean phenotype:

$$\Delta P = Sh^2 \tag{2}$$

where ΔP is the change in mean phenotype in one generation, S is the selection differential (difference in mean phenotypes of the selected individuals and the whole population) and h^2 the heritability of the trait.

Given that this Symposium is titled ‘Adaptation and Fitness in Animal Populations’, we must recognise another aspect of the problem – that adaptation and fitness are often confused, and although related concepts, they are not identical (Endler 1986).

3 Meanings of Fitness

Endler’s (1986) five concepts and definitions (Table 1) provide a basis for discussion and much of the following is adapted from his presentation.

Table 1 Concepts that have been called ‘fitness’ by various authors (after Endler 1986)

1. Fitness: The degree to which there is a consistent relationship between the trait and survival. Measured by the average contribution to the breeding population by a phenotype or by a class of phenotypes, relative to the contribution of other phenotypes.
2. Rate coefficient: The rate at which the process of selection proceeds. Measured by the average contribution to the gene pool of the following generation, by the carriers of a genotype or by a class of genotypes, relative to the contributions of other genotypes. May be replaced by ‘net fitness’ or ‘genotypic fitness’ to allow prediction of the dynamics of gene frequency change.
3. Adaptedness: The degree to which an organism is able to live and reproduce in a given environment or set of environments. Measured by the average absolute contribution to the next generation by a phenotype or class of phenotypes (R_0), or by the intrinsic rate of natural increase (r_m).
4. Adaptability: The degree to which an organism, population or species can remain or become adapted to a wider range of environments by physiological or genetic means.
5. Durability: Probability that an allele or genotype, a class of genotypes, a population or a species will leave descendants after a given long period of time.

3.1 Fitness (Concept 1)

Fitness is a measure of the degree of relationship between a trait and survival. Obviously, no relationship means that different phenotypes for a given trait will not differ in fitness, while a strong relationship means substantial fitness differences among different phenotypes for that trait. As a relative measure (i.e. relative to other phenotypes), fitness may apply to individuals, known genotypes that differ in phenotype or to populations, provided the comparison is made in the same environment. Fitness rankings when the same set of phenotypes are compared in some other environment may be quite different – phenotype x environment interaction for fitness. Although fitness is here defined in terms of a trait, that trait may simply be the overall phenotype (e.g. comparing individuals), or the overall average phenotype (e.g. comparing genotypes or comparing populations such as breeds of domesticated animals). For quantitative traits, the ‘selection differential’, and for polymorphic traits, the ‘selection coefficient’ are algebraically related measures. Robertson (1966), in a companion paper to two others on dairy cattle genetics (Barker and Robertson 1966, Robertson and Barker 1966) first noted that the selection differential is equal to the covariance of phenotype and fitness (survival – as defined here), and this relationship was extended and generalised by Price (1970, 1972).

Transforming through the genetic system gives the next concept – the selection rate coefficient.

3.2 Selection Rate Coefficient (Concept 2)

This coefficient determines the characteristic rate at which selection proceeds. It is measured by the average contribution to the gene pool of the following generation by the carriers of a genotype, or by a class of genotypes, relative to the contributions of other genotypes, and should be measured at the same life stage in two successive generations.

Consider the simplest case of a single locus with two alleles, sex-independent selection and random mating (the classical model – Prout 1969), and counting at the zygote stage in each generation. The rate coefficients are estimated by expressing the zygote frequencies in the progeny generation in terms of the parental generation allele frequencies and the selection coefficients, dividing each of these by the parental zygote frequencies, and finally expressing these ratios relative to the heterozygote genotype by dividing each homozygote ratio by the heterozygote ratio (see Endler 1986, pp. 41–42). The resulting rate coefficients for the two homozygotes are reciprocal, and they are frequency dependent, even though the selection coefficients are constant. That is, they will change in value as the allele frequencies change over successive generations within a population, or will be different in populations at different initial allele frequencies. Thus while the selection rate coefficient is of theoretical interest, it is not sufficient for application.

When considering genotypes, and possible selective differences among them, the aim is to understand the process of evolution, or in a more limited way, to predict the dynamics of gene frequency change in the short term. To do this, we need to measure the magnitude of any selective differences among the genotypes, i.e. ‘net fitness’ (Prout 1965). I hesitate to introduce further terminology, but this may be visualised as ‘genotypic fitness’ to distinguish it from fitness as in concept 1 above, i.e. ‘phenotypic fitness’.

Analogous to measuring rate coefficients, estimation of net fitness involves comparing the frequencies of genotypes in one generation with the frequencies of these genotypes in the next generation. However, there are complications: Prout (1965) showed for the classical model that the estimates obtained will show spurious frequency dependence, and will not be estimates of the true net fitness *unless* the selection process has been completed at the time when the genotypes are counted in each of the two successive generations. In practice, this means counting the numbers of each genotype in each generation when all surviving individuals have reached post-reproductive age. Alternatively, mature adult numbers may be used, if it were known from separate experiments that there were no differences among genotypes in adult survival.

The four basic components of selection that contribute to net fitness are (Christiansen and Prout 2000):

- Zygotic selection: differential survival of zygotes to maturity.
- Sexual selection: differential recruitment of mature individuals to parents.
- Fecundity selection: variation in fecundity of parental pairs.
- Gametic selection: differential survival of gametes.

Prout (1969) developed a general model incorporating these components, and then reducing this to the classical model, showed that:

(i) if genotype frequencies are determined in each generation after all selection is completed, then one transition (two generations) is sufficient for the estimation of net fitness (the case discussed above),
(ii) if genotype frequencies are determined in each generation before selection has started, then two transitions are necessary and sufficient,
(iii) if genotype frequencies are determined in each generation at a partially selected stage, then both pre- and post-observational components of net fitness must be estimated, and three transitions are both necessary and sufficient.

I am not aware that net fitnesses of genotypes have been estimated for any case in domestic animal populations. However, with increasing emphasis on the identification of quantitative trait loci (QTLs) affecting the expression of economically important production traits, this should become an issue. When a QTL is identified in a particular population, the breeder may assume that the breeding objective is to bring the most favourable allele to fixation. The unstated assumption then is that the genotypes at this locus do not differ in net fitness. Perhaps more likely, there will be net fitness differences among the genotypes, and the net fitness of each genotype will need to be determined and included in the overall breeding objective. How this should best be done, I leave as an open question, but in addition, the breeder needs to recognise:

(i) the difficulties of net fitness estimation, even more so for the more complex models than the classical one,
(ii) the statistical (sampling) problems of estimation in finite populations (the classical model analysis assumes an infinite population),
(iii) that the net fitness estimates for particular genotypes refer to the population in which the estimates were made. Although estimation is being done in terms of particular identifiable genotypes, these genotypes are not divorced from the genetic background – epistatic interactions and linkage disequilibria that affect the net fitness in one population may be different in some other population,

(iv) as a further development of (iii), these net fitnesses strictly should be applied within the environment in which the estimation was made, as genotype x environment interactions may well exist.

As more QTL are identified in domesticated species, the problem becomes even more complex, as the question then will relate to the fitnesses of multi-locus genotypes. For an n-loci, two allele model, where cis and trans heterozygotes are distinguished, $2^{n-1}(2^n + 1)$ genotypes exist, e.g. two loci – 10 genotypes, three loci – 36 genotypes.

As noted previously, (phenotypic) fitness may be defined and measured for populations, in terms of their relative survival to enter the breeding population. Thus for example, comparing two livestock breeds and starting with equal numbers of new-borns in each, which are then maintained in the same environment (including the same management, disease control, etc), the numbers of each breed surviving to enter the breeding population are counted. However, this is an incomplete measure as it takes no account of survival during the reproductive period. Further, the aim of breed comparisons will be to determine the 'suitability' of those breeds for production in a given environment – where suitability will mean not only production trait criteria, but also ability to survive and reproduce in that environment. Thus for population comparisons, we need a measure analogous to the net fitness of genotypes. However, there is an important difference – genotypes are compared to one another *within* an interbreeding population, while populations are reproductively isolated from one another. The appropriate measure for populations then is adaptedness.

3.3 Adaptedness (Concept 3)

Although defined by Endler (1986) as 'the degree to which an organism is able to live and reproduce in a given set of environments; the state of being adapted', the measure proposed 'the average absolute contribution to the breeding population by a phenotype or a class of phenotypes' appears inadequate in not including reproduction. The apparent problem hinges on the use of the term 'breeding population', which could be inferred as that of the current generation.

In essence, the 'average absolute contribution' is the number of individuals born in the next generation, which could be expressed relative to the number at birth in the current generation as the rate of change in population size per generation. This ratio is referred to as the net reproductive rate, usually designated as R_0. As a rate per unit of time, it may be measured as the statistic (r_m) variously referred to as the intrinsic rate of natural increase (Lotka 1925), the innate capacity for increase (Andrewartha and Birch 1954) or the Malthusian parameter (Fisher 1930).What this measures is the rate of growth of the population in a given environment, provided that the food supply and space are not limiting, and that the population has a stable age distribution. Two sets of data are required to estimate r_m – the age-specific birth rates (m_x) and survival rates (l_x).

Given these data, r_m may be estimated by solving the equation:

$$\int_0^{\infty} \mathrm{e}^{-r_m x}\ l_x m_x \delta x = 1 \tag{3}$$

While this approach has been used for some laboratory populations (Baldwin and Dingle 1986) and for some natural populations (Berryman and Lima 2006), adaptedness in domestic animal populations may be estimated more simply as the net reproduction rate (R_0). Thus for population (breed) comparisons of adaptedness, the example given above for (phenotypic) fitness is simply extended to a count of the number of newborn in each population in the next generation. One should note, however, that this gives, for the populations being compared, a measure of adaptedness to the specific set of environments during this one generation transition.

In recent years, the importance of adaptedness and the use of adapted breeds (or varieties) in genetic improvement programs have been recognised (e.g. Hammond (2000) for livestock and Namkoong (1998) for crop plant and forest tree breeding). In this context, however, adapted breeds are considered simply as those that live in some particular, usually stressful, environment, and the relative adaptedness of different breeds has not been measured. Choice of the breed (or breeds) for the foundation population of any breeding program is the basic first step (Barker 1967). Consequently, adaptedness should be a component of the characterisation and evaluation of breeds for conservation and for breeding programs.

3.4 Adaptability (Concept 4)

An individual, a population or a species may exhibit adaptability to some range of environments, or may have the capacity to become adapted. Adaptability then is a measure of actual or potential capacity, while adaptation is the process of becoming adapted or more adapted. Present (existing) adaptability could be tested, but potential adaptability cannot be predicted. Natural populations or species that lack appropriate adaptability may become extinct if the environment changes (e.g. global warming), particularly if they cannot respond (or respond fast enough) to track the changes. For domesticated populations, actual adaptability would be important if, for example, a particular breed needed to be used in some range of environments. More usually, however, the necessary criterion will be adaptedness – choice of particular breed(s) for specific environments (horses for courses or cows for climates).

3.5 Durability (Concept 5)

As noted earlier, Thoday (1953, 1958) defined fitness in terms of durability. It could be estimated as the expected time to extinction, discussed in detail by Endler (1986). However, it seems that little attention has been paid to this concept in the evolutionary literature, and it is not relevant to breeding programs in domesticated populations. However, it is a component that needs to be considered in setting priorities for conservation of endangered breeds (Ollivier and Foulley 2005).

4 Genetics of Fitness

In domestic animal populations, can fitness be improved, or at least maintained if there are negative genetic correlations between fitness and any of the production traits? That is, does fitness show genetic variation? Fisher's (1930) fundamental theorem of natural selection states 'The rate of increase in fitness of any organism at any time is equal to its genetic variance in fitness at that time'. A naïve interpretation of the theorem would be that there should be no genetic variation in fitness as alleles conferring high fitness would rapidly increase to fixation under natural selection. However, the theorem depends on many assumptions which generally will not apply in natural populations (see, for example, Charlesworth 1987).

While there are no estimates of the heritability of net fitness or adaptedness in domestic animals, the heritabilities of various components of fitness (survival, fertility, fecundity) have been estimated to be around 5–15% (Nicholas 1987, p. 405). For laboratory and natural populations, Roff and Mousseau (1987) and Mousseau and Roff (1987) summarised estimates of heritability for four categories of traits – morphology, behaviour, physiology, and life-history (i.e. fitness components). Life-history and behavioural traits had low heritability, while morphological and physiological traits had high heritabilities. But in a later analysis including more data, Hoffmann (2000) found no differences in the heritabilities of morphological, physiological or life-history traits, although life-history traits tended to exhibit a greater range and higher variance of the estimates. In an extensive analysis of a natural population, Merilä and Sheldon (2000) found that while lifetime reproductive success (as a surrogate for fitness) and various fitness traits had low heritabilities, they had higher levels of additive genetic variance than traits more distantly associated with fitness. But in addition, fitness and the fitness traits had much higher levels of residual variance (environmental and/or nonadditive genetic variance), leading to the low heritabilities. Similarly, in a domestic animal population (Norwegian dairy cattle), Andersen-Ranberg et al. (2005) found low heritabilities for fitness components, and again the residual variance was high, but substantial desirable genetic changes had been obtained.

However, while individual fitness components show significant, albeit low, heritability, there are two other aspects to be considered in developing breeding programs that include fitness – genetic correlations among the components and the genetic architecture of fitness. If individual fitness components were to be included in the breeding objectives, any negative genetic correlations among them would need to be known and taken into account. Equally, we need to acknowledge the ample evidence (briefly reviewed by Barker 2001) that epistatic interactions and dominance are substantial components of the genetic architecture of fitness. Can we identify and select for specific gene complexes?

5 Quantitative Genetics, Animal Breeding and Fitness

Animal breeding seeks to change the phenotypic value of traits that are of economic importance. Assuming a positive correlation between production traits that are being selected for and fitness (phenotypic or genotypic), there will be indirect selection for fitness. However, a negative correlation means that there will be indirect selection against fitness. In either case, an optimum breeding program would include genotypic fitness (for known QTL) and fitness traits such as fertility and longevity, together with production traits, in the breeding objectives.

Barker and Thomas (1987) noted that workers in evolutionary biology had rediscovered quantitative genetics, with a substantial development of quantitative genetic theory in relation to natural populations and evolution (e.g. Lande 1976, 1980). They also urged that empirical knowledge from selection in laboratory organisms and domestic plants and animals should provide part of the background to any consideration of quantitative traits in natural populations. I would now emphasise the reverse – that animal breeders would be well advised to include in their armoury (particularly in relation to fitness) recent developments in relation to microevolution and quantitative genetics in natural populations. This literature is quite vast, but recent papers (primarily reviews) that provide an introduction to these developments include papers in the special issue of *Genetica* (Hendry and Kinnison 2001 – particularly Agrawal et al. 2001, Arnold et al. 2001 and Wade 2001), Blows (2007), Blows and Hoffmann (2005) and Carter et al. (2005).

Finally, the rationale for this Symposium was to promote a better understanding of common problems, and better appreciation of the complementary studies in natural populations and in animal breeding. My hope is that this introduction to the Symposium will contribute in some small way to that objective.

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Genetic Architecture of Reproductive Fitness and its Consequences

Richard Frankham

Dedicated to Stuart Barker in recognition of his many important contributions to evolutionary genetics and for his support as teacher, supervisor, mentor, and colleague

Abstract Reproductive fitness characters are crucial in animal and plant breeding, evolutionary genetics and conservation biology. However, the nature of their variation is not well understood. I review evidence on the comparative architecture of genetic variation for fitness and peripheral characters for both wild populations and domestic livestock. Fitness characters differ from peripheral characters in typically having lower heritabilities, directional dominance, higher levels of gene interactions, asymmetry of selection response, inbreeding depression and heterosis, declines in mean from mutation, and non-linear changes in genetic variation with inbreeding. These differences are a reflection of the different forces of natural selection operating on the two classes of traits. Genetic diversity for fitness in wild populations is partially due to rare, deleterious, partially recessive alleles in mutation-selection balance, with a further component due to alleles at equilibrium due to balancing selection, but there is no consensus about the relative contributions. In contrast, livestock populations are unlikely to be in equilibrium, due to changes in their environments, artificial selection, small effective population sizes and in some cases to crossing of populations. These have major effects on the genetic architecture of fitness and especially on the proportion of polymorphic loci exhibiting overdominance. In general, long-term directional artificial selection on peripheral traits is expected to move their genetic architecture towards that of fitness traits. Whilst the breeders' equation provides good predictions of selection response for peripheral characters, it does not predict asymmetrical responses to selection as observed for fitness characters, and levels of inbreeding depression and heterosis remain unpredictable. Given the importance of fitness characters and the uncertainties on many important issues, they deserve a higher priority in quantitative genetics research.

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1 Introduction

Reproductive fitness traits represent the most difficult to study and least understood traits in quantitative genetics, yet they are crucial in animal and plant breeding, evolutionary genetics and conservation biology. In layer chickens, the commercial output (eggs) is a reproductive fitness trait. In other domestic animals raised for meat, milk, or fibre, reproductive output contributes to the cost of producing market animals, while survival rates are crucial to the cost of production in all livestock.

In spite of the importance of reproductive fitness, the nature of the genetic variation for fitness characters is not well understood and they have received less scientific investigation than peripheral characters (ones not closely related to reproductive fitness). In this contribution, I review evidence on the comparative architecture of genetic variation for fitness and peripheral characters for both wild populations and domestic livestock and consider the implications of this for animal breeding. I use genetic architecture here to mean the array of loci, alleles, frequencies, effects, dominance, pleiotropy and interactions affecting a quantitative character. The perspectives I bring to this are derived from having worked in quantitative genetics and animal breeding for the first 26 years of my career, but having worked predominantly in conservation genetics since 1990. While it is often assumed that genetic architecture is similar in wild populations and captive/domestic ones, I will argue that there are often substantial differences between them.

1.1 Different Characteristics of Fitness and Peripheral Quantitative Characters

Reproductive fitness and peripheral quantitative traits differ in a range of characteristics that are a reflection of the different architectures of their genetic variation (Table 1). Fitness characters exhibit strong inbreeding depression, while peripheral characters show little (DeRose and Roff 1999). Fitness characters typically show lower heritabilities than peripheral characters (Mousseau and Roff 1987; Roff and Mousseau 1987). Almost all quantitative characters exhibit genetic variation in almost all naturally outbreeding populations (Lewontin 1974). However, a few characters important to survival have essentially zero additive genetic variation in a small number of wild populations, in spite of ample genetic variation for other quantitative characters and for microsatellites (Hoffmann et al. 2003; Kellermann et al. 2006). Fitness characters typically show more selection response for low than high fitness (Frankham 1990), whilst responses are closer to symmetrical for peripheral

Table 1 Differences in characteristics of fitness and peripheral quantitative traits

	Quantitative character	
	Fitness	Peripheral
Inbreeding depression and heterosis	Strong	Weak
Asymmetry of selection response	Strong	Weak
Heritabilities	Relatively low	Intermediate
Non-additive genetic variation	Higher	Lower
Change in V_A with inbreeding	Curvilinear	Linear decline
Effect of mutation on mean	Reduces	Minimal change
Genotype x environment interactions	Stronger	Weaker
Natural selection	Directional	Stabilising

characters (Falconer and Mackay 1996). Fitness characters typically have much higher levels of non-additive genetic variation than peripheral characters (Kearsey and Kojima 1967; Barker 1979; Crnokrak and Roff 1995).

Peripheral characters show an approximately linear decline in additive genetic variation with inbreeding (Gilligan et al. 2005; Van Buskirk and Willi 2006). Conversely, a meta-analysis based on 22 studies revealed that fitness characters show a non-linear change in additive genetic variation (V_A) with inbreeding, increasing to more than double base population levels at intermediate levels of inbreeding before declining (Van Buskirk and Willi 2006). New mutations cause the mean to decline for fitness traits, but have little directional impact on means for peripheral characters (Mackay 1989; Garcia-Dorado et al. 1999; Yang et al. 2001). Finally, fitness characters generally show greater susceptibility to genotype x environment interactions than peripheral characters (Frankham and Weber 2000).

These different characteristics reflect different regimes of natural selection on peripheral and fitness characters (Falconer and Mackay 1996). Natural selection in a stable environment is directional on fitness and usually stabilising on peripheral characters (Endler 1986; Falconer and Mackay 1996; Kingslover et al. 2001; Zhang and Hill 2005).

2 Equilibrium Theory for Wild Populations

Quantitative genetic variation for fitness is maintained in two ways, either from the equilibrium between the occurrence of deleterious mutations and their removal by natural selection, or from balancing selection, whether via heterozygote advantage, rare allele advantage or selection that varies in space or time (reviewed by Charlesworth and Hughes 2000). The former prediction leads us to expect low frequencies of deleterious alleles at many loci in the genome and that most of these will be partially recessive. Models of balancing selection typically involve alleles at more intermediate frequencies than those for mutation-selection balance. Antagonistic pleiotropy may also lead to maintenance of genetic diversity if the two homozygotes at a locus have effects in different direction for different fitness components. If deleterious allelic effects on life-history traits are sufficiently recessive, such antagonism can lead to heterozygote advantage for net fitness and a stable polymorphism (Charlesworth and Hughes 2000). All mechanisms will be affected by genetic drift in small populations, but the balancing selection component will generally be less affected. The relative importance of mutation-selection and balancing selection in maintaining genetic variation is a matter of controversy, as detailed below. Hill and Zhang (2009) considers some issues concerning the maintenance of quantitative genetic variation for fitness characters in another chapter.

For peripheral characters, quantitative genetic variation can be due to the mechanisms described above for fitness characters, and in addition by (a) the balance between neutral mutation and random genetic drift, (b) the equilibrium between mutation, stabilising selection and drift, or (c) the balance between mutation, drift and natural selection operating by means of pleiotropic effects of mutations on fitness related to their deviations from the mean for peripheral characters (Barton and Turelli 1989; Falconer and Mackay 1996). Heterozygote advantage on loci affecting a quantitative character can generate apparent stabilising selection (Robertson 1956). The ability of mutation-selection balance to explain quantitative genetic variation for peripheral characters is controversial with Zhang and Hill (2005) concluding that it can, whilst Johnson and Barton (2005) are sceptical.

3 Empirical Data

Much of the information on the nature of quantitative genetic variation for wild populations comes from studies in *Drosophila*. Before discussing the conclusions, it is important to point out that there are potential flaws in essentially all the data. I know of no quantitative genetic studies on the architecture of fitness that have been done under wild conditions. Studies on recently caught wild populations have been done under different conditions in the laboratory. Even studies on long term captive populations often test fitness under different conditions to those the flies have been adapted to e.g. cage adapted flies from high densities may be tested under uncrowded conditions in vials. These differences allow genotype x environment interactions to affect the conclusions reached. Further, I will argue that there are likely to be differences between wild adapted and laboratory adapted populations in genetic architecture of fitness.

There is overwhelming evidence that mutation-selection balance contributes to genetic variation for fitness. Wild outbreeding populations of essentially all species contain loads of rare recessive morphological mutations (presumably deleterious) at many loci (see Lewontin 1974; Hedrick 2005). Wild populations of the few species that have been studied (several *Drosophila* species, plus blue krillifish and zebrafish) contain loads of rare recessive lethal mutations at many loci (see Lewontin 1974; Hedrick 2005). All species examined also contain rare deleterious mutations at many loci that we typically refer to as genetic diseases (see Lewontin 1974).