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Part I

Theory and Methods

"The natural phenomena of the evolutionary history of man claim an entirely peculiar place in the wide range of the scientific study of nature. There is surely no subject of scientific investigation touching man more closely, or in the knowledge of which he is more deeply concerned, than the human organism itself; and of all the various branches of the science of man, or anthropology, the history of his natural evolution should excite his highest interest."

Ernst Haeckel (1834–1919), *The Evolution of Man* (1892)

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1 Evolutionary Theory

Douglas J. Futuyma

Our contemporary understanding of evolutionary processes builds on theory developed during the "Evolutionary Synthesis" of the 1930s and 1940s, when Darwin's ideas, especially on natural selection, were joined with Mendelian genetics. Since, then, of course, our understanding of evolution has been greatly advanced by the discoveries in molecular genetics, as well as by continuing elaboration of the "neo-Darwinian" theory that issued from the Evolutionary Synthesis (Futuyma, 1998, 2009).

A capsule summary of contemporary theory, to be followed by more detailed explication, is as follows. Elementary evolutionary change consists of changes in the genetic constitution of a population of organisms, or in an ensemble of populations of a species. These genetic changes may be reflected in change of the population mean or variance of phenotypic characteristics. Any change requires that genetic variation originate by mutation of DNA sequences, and/or by recombination. The minimal evolutionary process is an increase in the frequency of a mutation, or a set of mutations, within a population, and the corresponding decrease in frequency of previously common alleles. Such frequency changes are the consequence of random genetic drift (leading to occasional fixation of nearly neutral genetic variants) or of diverse forms of natural selection. Successive such changes in one or more characteristics cumulate over time, yielding potentially indefinite divergence of a lineage from the ancestral state. Different populations of a species retain similarity due to gene flow and perhaps uniform selection, but can diverge due to differences in mutation, drift, and/or selection. Some of the consequent genetic differences can generate biological barriers to gene exchange between populations, resulting in the formation of different biological species.

THE ORIGIN OF VARIATION

Mutational changes in DNA sequences are of many kinds, ranging from single base-pair alterations to insertions, deletions, and rearrangements of genetic material, and even changes in ploidy. Many mutations have no effect on phenotype or fitness (are *selectively neutral*), such as synonymous mutations in protein-coding regions, which do not alter amino acid sequence, and mutations that occur in pseudogenes and other apparently nonfunctional regions. There exists greater potential for fitness effects of nonsynonymous mutations in coding regions, or of mutations in regulatory sequences. The rate of mutation (usually on the order of 10^{-9} per base pair per gamete) is usually too low to be a significant factor in driving allele frequency change within a population, but it can determine the rate of DNA sequence divergence in the long term, and can influence the equilibrium level of standing genetic variation. Considerable contemporary research concerns whether or not rates and directions of phenotypic evolution are often constrained by the supply of suitable mutations (Houle, 1998; Blows and Hoffmann, 2005). Mutation is a random process, in the sense that the probability of occurrence of a particular mutation is not affected by environmental circumstances which would make it advantageous. That is, there is no known mechanism by which the mutational process can be directed by the environment in advantageous directions.

VARIATION WITHIN POPULATIONS

Based on studies of many species, it appears that most populations carry substantial sequence variation in many gene loci, and that there exists some heritable variation in many or most "quantitative" phenotypic traits (continuous traits such as size, as well as the number of highly repeated unit characters, such as hairs or scales). Presence of two or more fairly common alleles or genotypes within a population is referred to as *polymorphism*. The level of variation is enhanced by mutation, recombination (often but not always), gene flow from other, genetically differentiated, populations, and some forms of natural selection (e.g., frequency-dependent selection, below). It is eroded by genetic drift and by most forms of natural selection (including

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directional and stabilizing selection on quantitative traits). The analysis of genetic variation is based on the frequencies of the alleles and genotypes at individual genetic loci (for an introduction to population genetics, see Hartl and Clark, 1997) For sexually reproducing populations, the Hardy–Weinberg (H-W) theorem states that the frequency of each allele (p_i for allele i) will remain constant from generation to generation unless perturbed by mutation, gene flow, sampling error (genetic drift), or selection, and that the frequencies of the several genotypes will likewise remain constant, at values given by the binomial theorem (p_i^2 for homozygote A_iA_i, and $2p_ip_j$ for heterozygote A_iA_j), if mating occurs at random. A single generation of random mating establishes H-W genotype frequencies at any autosomal locus. Furthermore, alleles at two or more polymorphic loci will become randomized with respect to each other (a state of *linkage equilibrium*) due to recombination. These principles have important consequences; for example, at H-W equilibrium, a rare allele exists mostly in heterozygous state, and so is concealed if it is recessive. In fact, rare, deleterious recessive alleles exist at a great many loci in populations of most outcrossing species, including humans. The frequency of heterozygotes ("heterozygosity") at a locus in H-W equilibrium ($2p_ip_j$) is often used as a measure of genetic variation at that locus, since variation is maximized when allele frequencies are equal.

Phenotypic variation in most quantitative traits is continuous or almost so, because it is *polygenic*, based on segregating alleles at several or many loci, and also includes environmental effects on the development or expression of a character (Falconer and Mackay, 1996; Barton and Keightley, 2002). At many of the segregating loci, the individual effects of alleles on the character typically are small, relative to the range of variation, but substantially larger effects are commonly contributed by segregating alleles at a few loci. The variance in phenotype (V_P) includes a genetic component (genetic variance, V_G) and an environmental component (V_E), and often an interaction effect ($V_{G.E}$) as well. An important component of V_G is the "additive genetic variance" (V_A), which is described by the correlation between the phenotype of parents and their offspring; it is this component of variation that is most important for evolution by natural selection. This component consists of the "additive" effects of alleles, that is, the phenotypic effect of each allelic substitution, averaged over all the genetic backgrounds in which it occurs. V_A depends on the number of loci contributing to the character, on the evenness of allele frequencies at each locus, and on the average magnitude of the phenotypic effect of different alleles. ($V_A = 2\Sigma p_ip_ja^2$ in the simplest case, where a is the average phenotypic effect and Σ indicates summation over loci.) The ratio V_A/V_P is termed the *heritability* (in the narrow sense), defined more narrowly than the "broad sense heritability" V_G/V_P. Because V_A is a function of allele frequencies, and V_P includes the environmental variance V_E, an estimate of the heritability of a trait is valid only for the particular population and the particular environment in which it was estimated, since other populations might differ in both these respects. Although many or most characters are genetically variable, we do not know what fraction of this variation can contribute to evolution by natural selection, since it is possible that a considerable portion of the variation may be deleterious under most circumstances.

The "mapping," or relationship, between a phenotypic character state (e.g., body mass) and the environment (e.g., caloric intake) is a genotype' *norm of reaction*. Genotypes may differ in their norms of reaction; for example, some people may gain more weight on a given diet than others. Such differences give rise to a genotype X environment interaction, expressed at the population level by the variance component V_{GXE}. The "mapping" between genotypes and phenotypes, even within a constant environment, often depends on developmental processes. For example, a trait may simply increase or decrease additively and gradually as + or − alleles (those that increase or decrease the character) are substituted in the genotype; or there may be nonlinear effects, so that the character suddenly changes from one to another discretely different state when the number of + alleles crosses a threshold.

A gene commonly affects two or more characters (*pleiotropy*), and so can contribute to a *genetic correlation* (r_G) between them. Another possible cause of genetic correlation is *linkage disequilibrium*, nonrandom association of certain alleles at two or more loci within a population (e.g., an excess of *AB* and *ab* combinations and a deficiency of *Ab* and *aB*). A genetic correlation caused by pleiotropy may be the net effect of both positive and negative components, since alleles at some loci may affect both characters in the same direction, and at other loci, in opposite directions. The value of r_G depends on the frequencies and phenotypic effects of all contributing loci. It is estimated by the covariance between characters over a set of families, just as the genetic variance is estimated for a single character. Genetic correlations are important because if the population mean of one character is altered, perhaps by natural selection, the other character will also be changed.

GENETIC DRIFT

Random genetic drift is simply random change in the frequency of alleles (and consequently, of genotypes). The genes carried by a generation of newly formed

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zygotes in a population are a sample of the genes carried by the previous generation, to which the parents belong. Because of random *sampling error*, the frequency (p) of an allele, say A_i, among the zygotes is unlikely to be exactly the same as in the previous generation, since there is likely to have been random mortality and random variation in female reproduction (fecundity) and male reproduction (number of mates) among individuals in the previous generation (here we are considering random, not selective, variation in survival and reproduction). So although the allele frequency in a new generation of N zygotes (carrying $2N$ genes if the species is diploid) is p on average (the same as in the previous generation), the frequency distribution of possible allele frequencies has a *variance*, given by the binomial expression Var $(p) = p(1–p)/(2N)$. This may be conceptualized as the variation among a large number of possible samples of $2N$ genes. The greater Var (p) is, the greater the random change in allele frequency is likely to be, from generation to generation, and thus the faster the process of evolutionary change by genetic drift. The expression for Var (p) tells us that this happens faster, the smaller the population size N. N in this theory refers to the *effective size* of the population, which is smaller than the "census size" if individuals vary in reproductive rate, if the sex ratio among breeding individuals departs from 1:1, or if the population fluctuates in size.

Since this variance holds in each generation, p fluctuates at random from generation to generation with no corrective tendency to return to its starting point, in a "random walk" to a boundary from which no return is possible: either loss of the allele A_i from the population or *fixation* of the allele A_i, i.e., attainment of $p = 1$. (Movement away from this boundary is possible, however, if new variation enters the population by mutation or by gene flow from other populations.) Hence, genetic drift results in loss of genetic variation within a population.

If a number of separate populations of the species all began with the same initial p, different populations would have different random paths, and in principle A_i would become fixed in some and lost in others; thus, genetic drift results in variation (divergence) among populations. An allele is more likely to be lost than to be fixed if its frequency is near zero, and conversely if its frequency is near 1.0; in fact, the probability, at any time, t, that an allele will eventually become fixed is p_t, its frequency at that time. A new mutation often exists, at first, as a single gene copy among the $2N$ genes carried by the N individual organisms in a population, so its initial frequency is $1/(2N)$, and this is its probability of fixation (if it is selectively neutral).

Since DNA sequence data have become available, another theoretical approach to studying the dynamics of genetic variation, *coalescent theory*, has become prominent (Hein et al., 2005). Looking back in time from the present, the gene copies (at a particular gene locus) in the population today are descended from only some of the genes carried by the previous generation's zygotes, due to sampling error; those zygotes in turn carried genes descended from only some of those in *their* parents' generation; and so on. Pursuing this logic, it is inevitable that all the gene copies in the population today are descended from one single ancestral gene copy (one DNA molecule) at some time in the past. The descendants of that gene form lineages of genes, replicating down through the generations to the present time, the set of lineages forming a gene tree which, like a phylogenetic tree of species, portrays their ancestry back ("coalesces to") the common ancestral (CA) gene, which existed t_{CA} generations ago. That ancestor was one of some number (say, $2N$) of genes in the population at that time, but the descendants of those other genes have not persisted to the present time, due to random genetic drift. (When this history was first described for human mitochondrial DNA, the catchy phrase "mitochondrial Eve" was applied to the female that carried the ancestor of all human mitochondrial genomes. Some people wrongly supposed that this meant the ancestral human population consisted of only one woman [and presumably one man].) The speed of genetic drift depends on population size, so it will not be surprising to learn that for a population of constant effective population size N ($2N$ genes at a diploid locus), the average time back to the common ancestor of all contemporary genes, t_{CA}, is $4N$ generations (e.g., four million if the effective population size is one million individuals).

A gene tree, representing the history of common ancestry of a sample of gene copies from one or more populations of a species, can be estimated by phylogenetic methods, using as characters the mutational differences (e.g., nucleotide substitutions) that have accrued among the lineages during their descent from their common ancestor.

THE NEUTRAL THEORY OF MOLECULAR EVOLUTION

Building on these principles, Motoo Kimura pioneered the development of a neutral theory of molecular evolution that is the basis for analyzing DNA sequence variation within and among species, and is often considered the "null hypothesis" against which alternative hypotheses, such as natural selection, must be compared (Kimura, 1983; Nei and Kumar, 2000). Mutational changes occur at many sites in a DNA sequence,

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at a total rate of, say, u_T per gene per generation. Of these, suppose some fraction f is selectively neutral, so the neutral mutation rate is $u = fu_T$. (The fraction f may depend on the functional role of a DNA sequence or the effect of a nucleotide change; for instance, a synonymous mutation in a functional gene or any mutation in a nonfunctional sequence such as a pseudogene is more likely to be selectively neutral than a nonsynonymous mutation in a gene with a critical function.) Since $2N$ genes are carried by (diploid) zygotes in each generation, the total number of new neutral mutations in the population each generation is $2Nu$, on average. We know from genetic drift theory that the probability of fixation of a new neutral mutation is $1/(2N)$ in a diploid population of constant size N, so $2Nu \times 1/(2N) = u$ new mutations occur each generation that will eventually be fixed. The time to fixation, we have just learned, is $4N$ generations, on average. Since this is the case each generation, u mutations should be fixed in a population every generation on average. In other words, population-wide substitutions of nucleotides in a DNA sequence occur at a roughly constant rate, so DNA sequence evolution theoretically acts as a *molecular clock*, accumulating ut substitutions over the course of t generations. If two populations (or species) are derived from a common ancestor and do not exchange genes for t generations, and if mutations at different sites in the DNA sequence are fixed in each population, the difference D between sequences taken from the two populations will be $D = 2ut$. If u (the neutral mutation rate, which can vary among genes because of functional differences or DNA repair processes) can be calibrated, then the time since the two populations separated can be estimated from the observed difference D, as $t = D/2u$. (Calibration is usually based on geologically dated events, such as fossils of the studied lineage or related lineages, or separation of two land masses on which related taxa reside.)

Eventually, D increases at a lower rate and levels off, because mutational substitutions occur repeatedly at the same sites within the sequence, erasing evidence of previous substitutions. This happens sooner for rapidly than slowly evolving sequences. According to the neutral theory, evolutionary change is more rapid if mutations do not affect organismal function, since mutations that affect protein function are more likely to be deleterious and eliminated by natural selection. Consequently, evolution is predicted, and found, to be more rapid at third-base than second-base positions in codons, because third-base mutations are more often synonymous. Sequence evolution is also more rapid in nonfunctional sequences, such as pseudogenes, than in functional sequences. (Indeed, the rate of sequence evolution between species is now used by molecular biologists to target functionally important versus less important sequences. This and other lines of evidence suggest that some supposedly nonfunctional, "junk," DNA sequences may have unknown functions, perhaps in gene regulation.) The ratio $\omega = k_A/k_S$, where k_A and k_S are the rates of nonsynonymous and synonymous nucleotide substitution, respectively, is often used as an index of the degree to which a protein-coding DNA sequence has been evolving neutrally, relatively free of functional constraint. If all mutations have been selectively neutral, ω should equal 1.

Genetic variation is lost from a population by genetic drift, as we have seen. However, it is regenerated by mutations at many sites in a DNA sequence, and at equilibrium there exists variation in nucleotide sequence within a population, when the rate of input by neutral mutation balances the rate of loss by genetic drift. A measure of polymorphism is the expected proportion of base pairs that differ between two gene copies taken at random (π) from a population. At equilibrium this equals $4Nu$, i.e., it is proportional to the population size and the mutation rate. Consequently, effective population size can be estimated from $\pi/4u$.

Because of polymorphism, the history of population separation may not be the same as the history of any one gene locus. Suppose an ancestral population divides into two populations (or species) A and B at time t_1, and B later separates into populations B_1 and B_2 at time t_2. Populations B_1 and B_2 are more closely related to each other, by definition, than they are to population A. If population B became fixed for a new mutation, and thus for a different sequence than population A, the mutation would be inherited by populations B_1 and B_2 and provide evidence of their sister-group relationship. Suppose, however, that populations A and B, and their common ancestor, have effective size N, and that the time between successive splits (between t_1 and t_2) is less than the $4N$ generations required for one or another sequence variant to be fixed in each population by genetic drift. If the common ancestor is polymorphic for sequences x and y (perhaps differing by a new mutation in sequence x), fixation may not occur until after the three populations have become separate. Then one sequence (say, x) may be fixed in both A and one of the derived B-populations (say B_1), and the other sequence (y) may become fixed in B_2. The phylogeny of genes may be accurate (the gene copies in B_1 are most closely related to those in A), but it would differ from the phylogeny of the populations. Therefore, it is important to use information from several or many independently inherited genes when analyzing the historical relationships among populations or species that have become separated during a short time span.

Summarizing this section, note that for selectively neutral mutations, whose fate is unaffected by natural selection, the theory of genetic drift and the related neutral theory of molecular evolution provide a basis

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for many important inferences: e.g., inferring effective population size, time since separation of populations (or since speciation), historical relationships among populations, and whether or not natural selection has affected DNA sequence divergence and polymorphism.

NATURAL SELECTION

There are so many nuances to the concept of natural selection that a simple, comprehensive definition is difficult to devise, but it may suffice, for present purposes, to define it as *consistent (nonrandom) differences in the rate of survival or reproduction among classes of entities that differ in inheritable characteristics*. The term "reproductive success" is often used for "survival and reproduction," since survival to reproductive age is prerequisite for reproduction. "Entities" is deliberately vague, because selection can (in principle) act among various kinds of biological "individuals," such as genes or larger sections of genetic material, individual organisms, groups of conspecific organisms, species, or clades (Williams, 1992). We speak of "classes" of genes, individuals, etc., because we cannot tell if a difference in reproductive success is nonrandom from information about a single individual of each kind; we require samples of similar genes or individuals in order to see if there is a consistent difference between different types of alleles or phenotypically different organisms. Natural selection, in distinction from genetic drift, is marked by a consistent difference in mean reproductive success within a given environment, not a random, unpredictable difference; thus natural selection is the antithesis of chance.

MODES OF SELECTION

Most analyses of evolution by natural selection are concerned with *individual selection*: differences in fitness, owing to a genetically variable phenotypic character, among individual organisms within a population. In the simplest models, the character is affected by variation at a single locus, and we suppose that the fitness of each genotype can be estimated. In practice, this can be difficult, because fitness, defined as a genotype's relative rate of increase, i.e., growth in numbers from one generation to the next, depends on several life-history parameters. The rate of increase is a complex function of the probability of survival at each age from birth to the oldest reproductive age class, and on the age-specific values of female reproduction (fecundity) and male reproduction (affected by mating success and sometimes by sperm competition). (In some cases, it may be affected also by other complicating factors, such as genetic compatibility among uniting gametes.)

Let us consider selection among individual organisms in a sexually reproducing population that differ in genotype at a single locus with two alleles, *A* and *a*. In the simplest case, the fittest of the three genotypes *AA*, *Aa*, and *aa* is a homozygote. If *aa* is rare, because the environment previously favored *AA* and has only recently changed so that *aa* is now the fittest genotype, we speak of *directional selection* for *aa*. Once *aa* becomes the prevalent genotype, allele *A*, as well as any other disadvantageous alleles that may arise by mutation, are reduced in frequency, and selection is often termed *purifying*. These are two faces of the same coin, selection that fixes the allele that, in homozygous state, maximizes fitness. The frequency q of the advantageous allele *a* attains the deterministic equilibrium $q = 1$ if only selection is operating, but if other alleles repeatedly arise by mutation, the equilibrium frequency will be set by the mutation rate relative to the strength of purifying selection ("mutation/selection balance"). Similarly, if a locally disadvantageous allele (perhaps *A*) that is advantageous in a different geographic population enters the population by gene flow, the genetic equilibrium is determined by the relative strength of gene flow and purifying selection. Gene flow from other populations can sometimes severely diminish the degree of adaptation of populations to their local environment.

Suppose the advantageous allele *a* is very rare, either because it has recently originated by mutation or because it has formerly been disadvantageous but nevertheless persisted in the population due to mutation/selection balance. If the frequencies of *A* and *a* are p and q respectively, the Hardy–Weinberg frequencies of the two genotypes that contain the *a* allele, *Aa* and *aa*, are $2pq$ and q^2, and the vast majority of the *a* genes are carried by heterozygotes. (For example, if $q = 0.01$, $2pq = 0.0198$, $q^2 = 0.0001$, and the ratio of heterozygotes to homozygotes is 198:1.) Whether or not the *a* allele can increase (or "invade" the population) depends almost entirely on the fitness of *Aa* relative to the prevalent homozygous genotype (*AA*); at this stage the fitness of *aa* is almost irrelevant because it is so rare. This means that even if *aa* is the fittest genotype, the *a* allele will not increase if it reduces the fitness of the heterozygote. This illustrates that natural selection acts only in the present, and cannot look forward toward the best possible outcome. It also shows the value of the Hardy–Weinberg principle.

Directional (or purifying) selection eliminates genetic variation, but several other modes of selection (*balancing selection*) may maintain genetic polymorphism. The simplest model is *heterozygous advantage*, in which the fitness of *Aa* is greater than that of either *AA* or *aa*, and all three genotypes segregate each generation due to random mating. Several hemoglobin polymorphisms in human populations, including sickle

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cell hemoglobin, are the best-known of the few well-documented examples of this mode of selection. Unquestionably more important is *frequency-dependent selection*, in which the fitness of each genotype is a decreasing function of its own frequency in the population, relative to other genotypes; that is, each genotype is more and more advantageous, the rarer it is. Many biological phenomena, including competition for resources, social interactions, and resistance to different genotypes of parasites, can give rise to such frequency-dependent effects. Mathematically, this is a powerful way of maintaining multiple alleles in a population, and cases are known in which 100 or more alleles appear to be maintained this way. Variable selection, in which different homozygotes are advantageous at different times or in different microhabitats within the area occupied by a breeding population, can also maintain polymorphism, although this is by no means guaranteed: mathematical models show that even if both homozygotes (*AA* and *aa*) are advantageous in different environmental states, only a rather narrow range of combinations of selection intensities and environmental frequencies will maintain all the genotypes indefinitely. (Note that persistence of both homozygotes because of their variable fitnesses also implies persistence of heterozygotes, due to random mating.)

The phenotypic implications of these genetic models depend on the relation between genotype and phenotype. In simple cases, in which there is either complete dominance of one allele or additive inheritance (in which the heterozygote's phenotype is intermediate), persistent genetic polymorphism implies persistence of two or three phenotypic classes, respectively. Most of the consequences of the single-locus models carry over into thinking about the effects of selection on a polygenic phenotypic trait, in which each variable locus contributes a small amount to overall variation. We consider the simplest case, an additive character, measured in, say, millimeters, for which "+" and "–" alleles at each of k loci add or subtract the same amount. The mean and variance of the character are determined by the frequency of the alleles at all of the loci; the mean will clearly be higher (and the variance lower) if most of the + alleles have high frequency. However, an intermediate mean could result from many possible allele frequency arrays, ranging from a highly variable population with $p = 0.5$ (i.e., + and − equally frequent) at each locus, to fixation of a single genotype that is homozygous for + at half of the loci and for − at the other half.

Directional selection on the character occurs when there is a monotonic relationship (at least over part of the range of possible phenotypes) between phenotype and fitness. For example, selection may favor larger phenotypes, namely those with more + alleles in their genetic make-up, so + alleles rise in frequency. If the fitness/phenotype relationship is "open-ended" (e.g., the unlikely circumstance that bigger is always better), selection will ultimately favor the genotype with + alleles only (and subsequently, any mutations with still greater effects), so + alleles become fixed at all loci, genetic variation is eliminated, and evolution ceases except insofar as variation continues to arise by mutation. Thus the magnitude of the "mutational variance," the per-generation increment in the variance of the character due to new mutations, will then limit the rate of subsequent response to selection.

What if the relationship between fitness and phenotype is not monotonic, but instead has an intermediate maximum ("optimum") that lies above the current mean phenotype? Directional selection will increase the frequency of + alleles and bring the mean to the new optimum. The character then becomes subject to *stabilizing selection*: deviations in either direction from the mean are disadvantageous. Many different combinations of + and − alleles can add up to give the same optimal intermediate phenotypic value; some of them are highly heterozygous, and others are homozygous for + alleles at some loci and for − alleles at other loci. Mathematical theory has shown that one or another of the homozygous genotypes will eventually replace all the other genotypes, so that genetic variation will be eliminated by stabilizing selection.

Studies of natural populations have shown that the most common forms of selection on quantitative phenotypic characteristics are stabilizing selection and *disruptive* (also called *diversifying*) selection, in which two or more phenotypes have higher fitness than do the intermediates between them (Endler, 1986). Disruptive selection at a single locus generally implies that the heterozygote for two alleles A and a has lower fitness than both homozygotes. Such a polymorphism is unstable, however, and the population will become fixed for the initially more common allele. In models of disruptive selection on an additive polygenic character, variation is not maintained indefinitely; instead, the population mean evolves to one or the other of the superior phenotypes, and stabilizing selection then takes over and reduces variation. In both the single-locus and polygenic models, variation is maintained only if disruptive selection is frequency-dependent, such that the fitness of the superior genotypes declines as they become more abundant. The simplest example would be if the genotypes are each adapted for a different food or other limiting resource, so that competition becomes more intense, and fitness declines, as a particular genotype becomes more abundant and depletes its resource.

I have introduced frequency-dependent selection as a negative feedback loop that can maintain stable coexistence of different genotypes in a single breeding

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population. It is possible, however, to imagine that the fitness of individuals of a particular genotype increases as the genotype's frequency increases. This would be a form of positive feedback that hastens fixation of the genotype, eliminating variation. Such selection is easily envisioned for many social behavioral traits, in which conformity to a predominant behavior pattern might be advantageous.

COMPONENTS OF FITNESS

Genotypes may differ in fitness due to one or more components, most of which are generally considered life history features (Stearns, 1992). These components contribute to the rate of increase (numbers/time) of a genotype, relative to others. One may think of a population of organisms as consisting of subpopulations of different genotypes (or of alleles) that are all growing like a bank account, with compound interest. All else being equal, a difference (in, say, survival probability or fecundity) expressed at an earlier age generally has a bigger impact on growth in numbers (fitness) than a similar difference expressed at a later age. Suppose individuals reproduce from age three until ten, and then die. A mutation that increases the chance of survival from age eight to nine has a smaller selective advantage than one that provides a similar survival advantage from age two to three, because survival enhancement in the older age classes will have much less effect on the number of offspring they might yet produce (and the number of genes passed on). Similarly, a mutation that increases reproductive output at age three has a greater impact on the increase of the mutation's frequency than if it affects reproduction at age ten, because (a) fewer individuals survive to age ten, so they don't get the benefit of the mutation; and (b) the mutation expressed at the younger age effectively shortens the generation time, so more descendants (grandchildren, great-grandchildren . . .) are produced per time unit than are produced by the genotype whose reproductive capacity is enhanced only at an older age.

Consequently, mutations that enhance survival or the number of offspring (e.g., number of eggs or young) are expected to increase fitness, but the magnitude of increase depends on the age at which these effects come into play. Moreover, there may exist trade-offs between different fitness components, or between a given component expressed at different ages, partly because an organism must partition energy or nutrients (e.g., protein) among different functions (the *principle of allocation*). For example, if reproduction reduces growth, it may be advantageous to delay reproduction until the individual is larger, which may ensure a longer life and higher fecundity that together more than make up for the reproduction foregone at an earlier age. On the other hand, if abundant inescapable predators make death at an early age virtually inevitable, selection will favor early reproduction, and mutations that defer senescence or enhance fecundity at advanced ages may well be disadvantageous (if these effects reduce early fecundity). The evolution of *intrinsic* senescence and mortality may therefore be affected by the age distribution of *extrinsic* mortality factors. Many potential adaptations have both benefits and costs, which may be environment-dependent.

A fitness component of particular interest is reproductive success achieved through success in mating, which Darwin termed *sexual selection* (Andersson, 1994). In many species of animals, the variation in reproductive success, and therefore the potential intensity of sexual selection, is greater in males than in females. This difference is generally ascribed to the smaller and far more abundant gametes of males than females, but sexual selection acts more strongly on females of some species (e.g., phalaropes and some pipefish and seahorses), in which investment in paternal care of offspring limits the number of a male's potential mates. (Thus the "choosier" sex, that exerts stronger sexual selection on the opposite sex, usually expends greater parental effort.) The two most commonly discussed modes of sexual selection are conflict between males, with winners gaining access to more females, and female "choice" of some males over others, based on one or more characteristics that usually are actively displayed to females. (In many cases, the same trait seems to play a role in both male–male and male–female interactions.) There is considerable evidence that conflict between males selects for larger size, greater weaponry, and many other kinds of traits that are used to establish dominance. The equilibrium mean value of such a trait will be set by balance between the reproductive advantage it provides and disadvantages such as its energy costs or effects on susceptibility to predation. Indeed, male investment in features that enhance mating success, such as mating activity, weaponry, or display features, may reduce investment in maintenance (e.g., immune system) and survival.

There is considerable evidence from birds, insects, and other animals that female "choice" imposes sexual selection, but there is considerable uncertainty about why females choose particular male phenotypes, such as males with more vigorous displays or more highly elaborated ornaments or vocalizations. According to one hypothesis, exaggerated male features indicate high physiological vigor that may stem from superior genetic constitution (the "*good genes*" hypothesis), and females that choose such males will have fitter offspring. There is some support both for this hypothesis

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and for several contenders. In models of *runaway sexual selection*, a nonrandom association (linkage disequilibrium) develops between genes that affect a male ornament and genes that affect the degree of female preference for this character. Females that prefer more highly ornamented males have daughters that inherit this preference (as well as unexpressed genes for large male ornamentation) and sons that inherit larger ornaments (as well as unexpressed genes for heightened female preference). (Note that most features expressed by a single sex are encoded in the genome of both sexes.) Therefore, any increase in the average male ornament in the population will cause a correlated increase in the average female preference, and vice versa, ratcheting both toward more extreme values until the process is halted either by counteracting selection or by running out of genetic variation.

In a twist on sexual selection theory, females and males are engaged in sexual conflict: males reduce females' fitness in various ways (e.g., incessantly attempting to mate), females are selected to resist, and selection favors males with ever more stimulating characteristics that can overcome female resistance (Arnqvist and Rowe, 2005). The scope for such interactions appears greater than was formerly supposed, because it is clear that females of many species mate with multiple males, even in species that form a supposedly monogamous pair bond. Thus males have the potential of siring offspring by mating not only with unmated, but also with previously mated, females. The consequences include competition between sperm from different males. Probably because of the strong, long-continued selection exerted by sperm competition and sexual selection, reproductive characteristics, including male display features, genitalic morphology, proteins from accessory reproductive glands, sperm morphology, and cell-surface proteins of gametes, are rapidly evolving characteristics that often are the major differences among closely related species.

MODELING ADAPTATION

In considering components of fitness, we have moved from the very general theories of population and quantitative genetics, which apply to unspecified genes and characters, to models of the evolution of specific classes of characters, such as life history features. Evolutionary analyses of adaptive evolution of specific classes of characters employ several approaches to modeling (Bulmer, 1994). The evolution of some features is best analyzed by *genetic models*. This is true of models of sexual selection by female choice, for example, because linkage disequilibrium is an essential component and it requires an explicit genetic approach. The major alternative is *optimization*, an approach that attempts to specify what the optimal character state ought to be, given some assumptions about benefits, costs, and constraints. This approach assumes that there has been enough time and enough genetic variation for natural selection to bring the mean character state in a population nearly to its optimum value, and that the genetic details do not matter very much. Whether or not these are reasonable assumptions may depend on empirical information about such things as genetic variation and evolutionary history (e.g., inferences about how long a species has probably been subject to consistent environmental selection).

Optimization is a common approach in the fields of functional morphology and physiology, in which it is assumed that fitness is enhanced by maximizing some function, subject to constraints such as costs in energy or materials, or compromises with other functions. For example, aerodynamic models have been used to model flight and optimal wing morphology in birds, in which compromises among speed, maneuverability, and energy expenditure are taken into account. Among nonsocial aspects of behavior, models of optimal foraging describe when a foraging animal should give up searching in one patch and move to another.

Social interactions entail complexities that make genetic modeling difficult, and have been analyzed almost entirely by optimal models. The complexity arises from the frequency-dependent fitness of different trait values: the optimal behavior of an individual often depends on the behavior of other members of the population. Among the most widely used approaches is game theory (Maynard Smith, 1982). Suppose, for example, that the problem is whether or not parental care, by either or both mated partners, will evolve by individual selection. One might postulate two "strategies," "*S*tay and provide care to offspring" and "*D*efect and attempt to reproduce again." For each possible pair (S♀/S♂, S♀/D♂, D♀/S♂, D♀/D♂), one postulates for each partner the expected reproductive "payoff," which depends on both the benefit to each partner (in terms of surviving offspring from this mating) and the costs to each (in terms of the likely reproductive success sacrificed). The average fitness of each strategy, for each sex, is then its payoff averaged over the possible pairings, and weighted by their frequency in a random-pairing population. The best strategy, within the set of strategies considered (here, S and D), is the one that, if fixed in the population, will remain fixed even if individuals with alternative strategies attempt to invade. This is the *evolutionarily stable strategy*, or *ESS*.

LEVELS OF SELECTION

Natural selection was defined above as "consistent (nonrandom) differences in the rate of survival or

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reproduction among classes of entities that differ in inheritable characteristics." These "entities" may be at different, nested levels, and the effects of selection at different levels may be opposite (Okasha, 2006). Consider, for example, the level "individual organism" and the level "somatic cell lineage" within a multicellular organism. If a cell lineage experiences a mutation that causes rapid, unrestricted cell division, that lineage has a "selective advantage" relative to other cells, and will constitute an increasing proportion of cells within the domain of the single organism (Nowak, 2006). This proliferation – cancer – is clearly disadvantageous to the higher-level entity (the organism), if it occurs before or during the organism's reproductive ages. Selection among genetically variable individual organisms will favor genotypes that have the ability to suppress cancerous tumors.

We may likewise distinguish selection among individual organisms with different genotypes (the level of selection assumed so far in this chapter) from selection at the level of the individual gene (locus). In asexual organisms, there is little conflict between selection at these levels, because the fate of a gene (survival, passage to subsequent generations) depends on that of the rest of the genotype to which it is bound. But in sexually reproducing organisms, conflicts can arise. A famous example is the "*t* locus" in house mice. More than 90% of the sperm of males heterozygous for a normal allele (*T*) and one of several recessive alleles (*t*) carry the *t* allele (an example of *meiotic drive*). Some of the recessive alleles cause embryonic death, and others male sterility, in homozygous condition. The differential transmission of *T* and *t* alleles constitutes differential "reproduction" at the gametic level (*genic selection*), opposing differential survival of individual mice (*individual selection*). Genic selection accounts for many phenomena, such as the proliferation of transposable elements ("selfish genetic elements"): DNA sequences that replicate more frequently than most of the genome.

Genic selection provides one way of viewing the evolution of co-operation, which stands in contrast to the selfish individualism that generally characterizes individual selection (Dawkins, 1982; Sober and Wilson, 1998). Cells in multicellular organisms co-operate because they are (generally) genetically identical: a gene in a liver cell is replicated by virtue of the replication of identical copies in the germ cell line – and the fate of the germ cell line depends on the gene copies functioning in the liver. Likewise, the rate of increase of a parent's gene over generations depends on the survival and replication of copies of that gene in the parent's offspring – and so alleles that program parental care may increase in frequency. This is an example of *kin selection*: selection in which alleles differ in fitness by influencing the effect of their bearers on the reproductive success of individuals (kin) who carry the same allele due to common descent. (In this case, the "bearers" are parents, and the "kin" are their offspring.) In the same way, genes that enhance their bearers' propensity to help more distant relatives may increase in frequency – but the consequent increase in the relatives' fitness must be greater, since their probability of sharing the "helping allele" is lower. William Hamilton formalized this relationship in what has become known as Hamilton's rule, which states that "altruism" spreads if $rb > c$: an altruistic trait can increase in frequency if the benefit (b) received by the donor's relatives, weighted by their relationship (r) to the donor, exceeds the cost (c) of the trait to the donor's fitness. The relationship, r, between donor and recipient is the fraction of the donor's genes that, on average, are identical by descent with any of the recipient's genes. For example, $r = ½$ between parent and offspring, so an allele for parental care should spread, even if it costs the parent her life and subsequent reproduction, as long as her care results in survival of more than two extra offspring (compared to a parent that does not provide care). (Kin selection is only one of several explanations of the evolution of co-operation among genes, cells, or conspecific organisms. For example, reciprocity ["reciprocal altruism"] may evolve if individuals recognize one another and can benefit others or not, depending on their history of behavior.)

Because of kin selection, the family (mated pair and associated offspring) is an obvious context in which co-operation may evolve. Nevertheless, intrafamilial interactions are riddled with conflict. *Sexual conflict* inevitably arises from the sex difference in gamete size (and some other features in certain species): male fitness can be increased by mating with many females, whereas all of a female's eggs can usually be fertilized by a single male. Female fitness is more likely to be enhanced by her offspring's survival, which may be increased by parental care or by "genetic quality." Parental care increases the fitness of both parents, but it entails costs, including lost reproductive opportunities. If offspring were as likely to survive with uniparental care as with biparental care, selection would favor defection by one sex – the one for which parental care is more costly (Clutton-Brock, 1991). A further complication is that if a caregiver were not actually the parent of some or all of the offspring, he (or she) would have less of a genetic interest in their survival. "Extrapair copulation," common in seemingly monogamous species of birds, therefore alters the costs and benefits of parental care. In some species of primates and other mammals, a male that replaces another male kills his new mate's offspring, since he has no genetic interest in them, and killing them enables him to father his own offspring faster. (Killing some offspring can

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