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CAN WE REALLY IDENTIFY SPECIES, LIVING OR EXTINCT?

ABSTRACT: Given the profound effects regulatory genes – especially the better known homeobox genes – have on an organism's development, it is imperative that this level of what is obviously a genetic hierarchy is incorporated into our ideas on the origin and identification of species. Since regulatory genes are inherited in the same Mendelian fashion as structural genes, but the consequences of mutations affecting the former are more profound, they are of significance in discussions of species identification. Moreover, because nonlethal mutations typically arise in the recessive state, their spread via heterozygosis throughout the population will be silent. However, when homozygotes begin to appear, the morphologies resultant from the recessive mutations will emerge as if out of nowhere – thereby underscoring the role of morphology in both delineating species and being a potentially faithful reflection of phylogenetic relationships among taxa.

KEY WORDS: Species – Homeobox gene – Regulatory gene – Structural gene – Mutation – Mendelism – Darwinism – Punctuation – Allopatric speciation

INTRODUCTION

In spite of the fact that for centuries now systematists have uniformly agreed in theory that the species is the basic unit of nature, and, therefore, of classification, the most continually vexing question is how, in practice, one defines a species and distinguishes one from another. The matter is further complicated when dealing with fossils because the operational definition (or its variant popular versions) of a species, being based on real or expected properties of extant organisms, is even more impossible to apply to extinct animals, which themselves may be represented by only the smallest fraction of their entire biology. But short of throwing up our hands in utter exasperation and continuing to treat the problem as known but, unfortunately, uncorrectable, it might perhaps be beneficial to review briefly the history of how the species problem has been conceived of and dealt with, especially in the decades leading up to the grand evolutionary synthesis and the solidification of the view of species and speciation that came to dominate the field.

THE SPECIES PROBLEM: PRE-TWENTIETH CENTURY

As we all know, it was Linnaeus who introduced the first standardized and repeatable system of classifying organisms. His binomial approach, defining a presumably biologically real aggregate of individuals by way of a genus and a species, has persisted as the basic format of communication among systematists since the publication of the first edition of the *Systema Naturae* in 1735. The rank of genus had been first used by the sixteenth century taxonomist Conrad Gesner, while the concept of a species as a distinct entity in nature [the word itself being a derivative of "spece" and "spice," both of which refer to a specific kind of something (see Gregory 1910)] had been introduced by the seventeenth century systematic zoologist John Ray.

Formalizing a categorical reference to an aggregate of individuals and demonstrating the biological reality of such an community represent, however, two drastically different concerns. This duality was not lost on Ray, who was

apparently the first to speculate that the criteria of copulation and the production of fertile offspring could serve in evaluating whether individuals belonged to the same species (see Blumenbach 1795). Although Frisch, early in the eighteenth century, restricted Ray's species definition to wild animals because of the ambiguities involved in incorporating domestic animals into such deliberations, Buffon adopted it in its entirety (ibid.). But while these attempts at articulating exactly what constitutes the difference between varieties and species might sound familiar to systematists today, it must not be forgotten that the impetus for these early speculations was a desire to document the Great Chain of Being, in which boundaries between species had to exist because all life on earth arose through divine creation.

In terms of the venue of this conference, it is significant that the consideration of species drew the attention of one of Göttingen's foremost scholars, Johann Friedrich Blumenbach. In 1775, in his doctoral thesis, he discussed hybridization. Later, he (1795) tackled the species problem more directly, concluding that restricting Ray's definition to only wild, rather than wild and domesticated animals, would not resolve the matter. No one, he ventured, could ever fully test the criterion of copulation-leading-to-the-production-of-fertile-offspring because this would at times require bringing together organisms that, in the wild, were widely dispersed. Following Frisch, he rejected domesticated animals as providing clues to understanding nature, citing among his reasons the contradiction of classifying as separate species animals that some scholars might regard only as varieties even if such morphologically different individuals did satisfy the criterion of "being able to copulate."

In spite of the fact that Blumenbach ultimately rejected "copulation" as a defining attribute of a species, he could not replace it with a concept of more constant utility because of taxonomic problems that arose from differences of opinion between taxonomists. For instance, although he perceived albino rabbits with their snowy coats and red pupils as being representative of a variety within a species, other systematists, who were more impressed by the consistency of these rabbits' features, treated them as a true species. Because of this kind of dilemma, Blumenbach resorted to the criteria of analogy and resemblance as the bases for distinguishing species, of which he (1795: 191) offered the following examples:

> "I see, for example, that the molar teeth of the African elephant differ most wonderfully in their conformation from those of the Asiatic. I do not know whether these elephants, which come from such different parts of the world, have ever copulated together; nor do I know any more how constant this conformation of the teeth may be in each. But since, so far in all the specimens which I have seen, I have observed the same difference; and since I have never known any example of molar teeth so changed by mere *degeneration*, I conjecture from analogy that those elephants are not to be considered as mere varieties, but must be held to be different species.
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> "The ferret, on the contrary, does not seem to me a separate species, but must be considered as a mere variety of the polecat, not so much because I have known them [to] copulate together, as because the former has red pupils, and from all analogy I consider that those mammals in which the internal eye is destitute of the dark pigment, must be held to be mere varieties which have degenerated from their original stock."

In the absence of evolutionary theory, the notion of degeneration was popular among eighteenth century naturalists for explaining how differences within a species could arise. Indeed, within our own species, Buffon (1749–67) and Blumenbach (1795) explained the rise of human "races" or varieties as a degeneration from an original human "ancestor" of greater perfection.

The recognition of evolution did not, however, bring with it a clarification of how one might deal with species. Even Darwin (1859) was tentative on the topic in *On the Origin of Species by Means of Natural Selection*. Although he did discuss species within the context of variation under nature in Chapter II and the significance of hybrid sterility in Chapter VIII, he remained vague on the matter of their definition in part because of his preoccupation with the minutiae of individual variation, which he saw as linking species (however defined) with other species and varieties within species. Through Darwin the defining of species became tied to ideas on speciation. But because he was unable to articulate a model that dealt with the diversification of species (see Schwartz 1999b), his thoughts on species formation were constrained to linear transformation.

Although circular in reasoning, Darwin (1859: 47) dealt with the species problem by resorting to the wisdom of those supposedly in the know: "Hence, in determining whether a form should be ranked as a species or a variety, the opinion of naturalists having sound judgement and wide experience seems the only guide to follow." When greater doubt about species status exists, he recommended relying on the decisions of a "majority of naturalists." But it was in the many cases of questionable species or variety status, which were for him the result of individual variation blurring lines of taxonomic distinction, that Darwin (1859: 51–2) found the essence of the origin of species: It is from individual differences that slight varieties derive; from the latter "more strongly marked and more permanent varieties"; and from this latter group, "sub-species" and then species by "the action of natural selection in accumulating…differences of structure in certain definite directions." As Mayr (1942) would later argue, but with no mention of Darwin's priority to the basic idea, a subset of a species (Darwin's "a well-marked variety" and Mayr's subspecies) "may be justly called an incipient species" (Darwin 1859: 52). And since, according to Darwin (1859: 95), "natural selection can act only by the preservation and

accumulation of infinitesimally small inherited modifications, each profitable to the preserved being," the transformation of a variety or an incipient species into a new species would inevitably be a long and gradual process.

Darwin also discussed species in terms of hybrid sterility. Because he was as motivated to offer a biological alternative to a biblical interpretation of nature as he was to present his theory of evolution based on natural selection, he (1859: 245) acknowledged that "The view generally entertained by naturalists is that species, when intercrossed, have been specially endowed with the quality of sterility, in order to prevent the confusion of all organic forms." But, he argued, there is a difference between sterility resulting from crosses between species and sterility arising from crosses between hybrids. In the latter case, sterility is supposedly due to imperfections in the sexual organs that result from the crosses themselves. In species crosses, however, "It is certain, on the one hand, that the sterility of various species when crossed is so different in degree and graduates away so insensibly, and, on the other hand, that the fertility of pure species is so easily affected by various circumstances, that for all practical purposes it is most difficult to say where perfect fertility ends and sterility begins" (ibid. 248).

History reminds us that Darwin and other evolutionists laboured to articulate evolutionary ideas in the absence of understanding the basics of inheritance, which the Austrian monk and plant hybridizer, Gregor Mendel, unravelled and published in 1866. Whether or not awareness of the law of segregation would have provoked Darwin and others to develop models of speciation that incorporated theories of diversification is, of course, is impossible to know. But it is clear that the monk was curious as to how selective breeding, of plants, at least, might transform one presumed species into another.

Prompted by his demonstration of morphological stability over multiple generations of (presumed) varieties of garden pea, Mendel (1866: 3–4) was forced to admit that: "If we adopt the strictest definition of a species, according to which only those individuals belong to a species which under precisely the same circumstances display precisely similar characters, no two of these varieties could be referred to one species." Yet, according to the experts, most of the groups of garden peas were supposed to belong to the same species. Since Mendel could successfully cross- and back-breed his garden peas, it was "quite immaterial for the purposes of the experiments in question" (p. 4) if they belonged to varieties or subspecies of the same species or to different species. However, Mendel (ibid.) had to conclude that "[i]t has so far been found to be just as impossible to draw a sharp line between the hybrids of species and varieties as between species and varieties themselves."

In discussing the transformation of one plant species into another, Mendel referred to Gärtner and Kölreuter's work on hybridization, which Darwin (1859) also cited prominently in his discussion of species. That Darwin would have found these experiments important to his argument that crosses between species result in a continuum from fertility to sterility is evident in Mendel's (1866: 37–8) summary of how a species could be altered:

> "If a species *A* is to be transformed into a species *B*, both must be united by fertilisation and the resulting hybrids then be fertilised with the pollen of *B*; then, out of the various offspring resulting, that form would be selected which stood in nearest relation to *B* and once more be fertilised with *B* pollen, and so continuously until finally a form is arrived at which is like *B* and constant in its progeny. By this process the species *A* would change into the species *B*."

It is also obvious why it was important that crosses between species would not be fated to produce only infertile hybrids. For Darwin, who embraced the notion of blending inheritance [e.g. see his (1868) theory of pangenesis], novelty derives from the combination of different variations as well as from the inheritance of characters acquired through use or disuse. Since differences between individuals arise through natural selection acting on the available variability, hybridization between species represented another avenue for producing novelty. Unlike Bateson (e.g. 1913) and de Vries (1900), for example, Darwin rejected the importance of "monstrosities" or "sports of nature" as potential sources of novelty.

Although Francis Galton (1875) had earlier developed a model of inheritance based on discrete, non-blending units of inheritance (his stirps), it was Hugo de Vries (1900) who first proposed that the existence of such units meant that variation between individuals as well as between species must be discontinuous (rather than continuous) and that mutations in such units (his pangens) were the basis of novelty. Whereas Darwin envisioned natural selection's acting on an infinite supply of slight variations as leading gradually to the emergence of new species, de Vries argued that novelty sufficient to produce species arose only through mutation producing new pangens during the phase of replication. Once present, natural selection could act on the novelty, rejecting it if its effects were deleterious (rather than retaining it because it might be advantageous). The number or frequency of pangens could change in response to shifts in the climate or environment, but this would only result in low level variability between individuals that would be as transient as the external fluctuations were. For de Vries, new species emerge rapidly.

William Bateson (e.g. 1909), who translated Mendel's publication into English, also rejected "continuous variation" and "the gradual transformation of species" in favour of discontinuous variation and distinctiveness. He did so because his studies on variation had failed to document Darwin's prediction that organisms change in direct adaptive response to their environment (Bateson 1894). Contrary to Darwin's prediction, Bateson found that changes in the environment lead not to the origin of new

features or adaptations, but merely to shifts in the frequency of expression of characters already present in a species.

Bateson agonized over how to define a species. Although he never did produce a satisfactory definition, his (1894: 2) view of species was diametrically opposed to Darwin's:

> "No definition of a Specific Difference has been found...But the forms of living things, taken at a given moment, do nevertheless most certainly form a discontinuous series and not a continuous series. This is true of the world as we see it now, and there is no good reason for thinking that it has ever been otherwise. So much is being said of mutability of species that this, which is the central fact of Natural History, is almost lost sight of, but if ever the problem is to be solved this fact must be boldly faced. There is nothing to be gained by shirking or trying to forget it.
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> The existence, then, of Specific Differences is one of the characteristics of the forms of living things. This is no merely subjective conception, but an objective, tangible fact."

THE SPECIES PROBLEM: THE TWENTIETH CENTURY

Bateson experimented with the details of inheritance, eventually documenting, for example, that nonlethal mutations typically arise in the recessive rather than dominant state, that certain characters are sex-linked, and that some characters are represented not by a unilaterally dominant state, but by a co-dominant one. In a seminal publication, he and Saunders (1902) outlined the basics of inheritance and also discussed their applicability to evolutionary questions. Since, they argued, many characters, and thus the genetic units that underlay them, are passed freely from parent to offspring and of no apparent consequence for existence (e.g. height, weight, colour), there must be some characters and genetic units that are not so easily interchangeable. As such, they would interfere with mating and/or the production of reproductively viable offspring. Since experiments in plant and animal hybridization sought to understand alternative allelic states and the continuity of inheritance from one generation to the next, Bateson and Saunders focused on discontinuity of inheritance between groups of individuals: that is, between species. The reason why most of the hybrid offspring of cross-bred species seemed to produce sterile offspring was, they suggested, because the gametes of cross-species' hybrids could not properly sort out the genetic material inherited from the parental generation.

Although Mayr (1942) criticized Bateson along with Buffon and others for using sterility as a criterion for species recognition, this attack was unjustified. Buffon, for example, began with the assumption that a divine creator had placed life on earth and that hybrid sterility was a reflection of the creation of different species. Bateson, however, sought a genetic explanation at the level of gametic (in)compatibility, which was in general also the concern of various subsequent geneticists, such as Fisher (1930) and Dobzhansky (1935, 1937).

Like de Vries, Bateson (1894, 1909, 1913) denied to natural selection both a role in generating evolutionary change and a utilitarian function that always selected adaptively advantageous features. Convinced that the kind of morphological change on which Darwinism was based was transient and nonadaptive, Bateson (1894) proposed that the discontinuous nature of evolutionarily relevant change was due to the differential expression of repeated or meristic parts, whether they be petals, scales, feathers, vertebrae, teeth, or digits. Rather than features gradually evolving from one state to another, structures would increase or decrease in complexity by whole increments, as is often the case with supernumerary structures in vertebrates.

During the early phase of his career, the geneticist Thomas Hunt Morgan (1903) was an ardent anti-Darwinist who embraced the notion of discontinuous variation. Like Bateson, he saw speciation as involving first an interference with genetic continuity and then an acquisition of genetic discjunction. He did not believe that evolutionary novelty could derive from "sports of nature," but he (1903) did subscribe to a version of de Vries' mutation theory: If a new feature could arise as a result of a single mutation, so, too, could a new species. If natural selection played a role, it was in fine tuning the novelty after it had arisen. New features persisted, not because they were advantageous to their bearers, but because they were not disadvantageous or deleterious. In addition to rejecting Darwinism, for awhile, Morgan (1909) also opposed Mendelism, in its populational as well as evolutionary contexts.

Morgan (1916, 1925, also Morgan *et al.* 1915) became both a Darwinian and a Mendelian through his experiments in fruit fly population genetics. His conversion came from two sources: the unexpected appearance of an eyeless strain of fruit fly in his experimental colony, and his collaborator Muller's manipulation of average fruit fly wing length from longer to shorter than body length. The former convinced him that, while mutation introduced evolutionary novelty, it was not significant at the level of the species because the mutants could still breed with normal individuals. From Muller's altering average wing length over many generations of fruit flies, Morgan concluded that evolutionary change, too, was a slow and gradual process. Although he accepted that discrete units of heredity were tied to distinct features, he relegated discontinuous variation to such a low level of importance that, for all intents and purposes, discontinuous variation became continuous – as Yule (1902) had earlier argued mathematically. In one fell swoop, Morgan melded Darwinism with Mendelism: mutation introduced the novelty upon which natural selection subsequently acted. The slow transformation of one species into another could now, it seemed, be explained genetically.

Among those who embraced this perspective without question was R. A. Fisher (1930), who sought to demonstrate mathematically the Darwinian process of accumulating continuous variants and the role of natural selection. Central to Fisher's considerations, as well as to the theories of his intellectual rivals, Wright (e.g. 1932) and Haldane (1932), was the fact that most nonlethal mutations arise in the recessive state. This was important, Fisher argued, because a novelty that arose in the dominant state would be immediately expressed and, probably just as quickly, eliminated from the population, even if it might be advantageous. A recessive allele, however, could spread silently through the population for some amount of time. Because he advocated heterozygosity as being the most advantageous genetic state, Fisher was forced to theorize that the recessive mutation would soon be converted to the dominant state, replacing the original or "wild" dominant allele, which would allow the novelty to be expressed in the preferable heterozygous condition. In Wright and Haldane's models, however, the mutation was allowed to stay in the recessive state until the population was sufficiently saturated with heterozygotes that homozygotes expressing the novelty would emerge. At some later time, the recessive mutant allele would be converted to the dominant state.

Because Fisher believed that variation is continuous and mutations and their effects minute, he perceived the evolution of species as an ongoing, gradual process. Had he favoured discontinuous variation, he would have had available a mechanism for producing taxic diversity – discontinuity, of course, precludes the existence of an unbroken continuum. As an ardent gradualist, however, he had to develop a model of continual transformation that also accommodated branching speciation.

On the grounds that within-species variation is supposed to be slight, Fisher (1930) argued that a species could be defined by its members sharing the vast majority of their gene loci. Given this genetic commonality, a barrier to gene flow between subsets of the species had to be introduced so that the ongoing process of continual change via mutation and natural selection could produce two species from one. Although he invoked geographical isolation as one possible intrusion on gene flow, his primary source was environmental instability ("heterogeneity"), which could be either external to the organism (e.g. a physical barrier) or internal, at the cellular level. Instability arose in populations of a widespread species as they adapted to their local conditions, with natural selection choosing features that would be advantageous only in the context of an individual's specific circumstances. A species thus clinally "stretched" could be easily disrupted and gene flow eliminated between its increasingly dissimilarly adapted populations, which would ultimately find themselves reproductively isolated from one another.

Wright (e.g. 1932) and Haldane (1932) rejected Fisher's basic premise of gene flow being continuous throughout an entire species on the grounds that there are always genetic subsets within a species. They saw speciation as a rapid process that involved the isolation to some extent of relatively small peripheral populations. Experiments in animal husbandry through managed inbreeding made them realize that the smaller the population, the faster mutations (arising in the recessive state) or novel combinations of genetic interactions would spread through the population.

For Wright, the peripheral population, which was typically not completely isolated either genetically or physically from the original population, was the source of the novelty that eventually transformed the species. The small size of the peripheral population drove its members to change quickly, while its continued connection to the parent species allowed it to infuse the latter with new, adaptive genetic combinations (which were Wright's preferred sources of evolutionary novelty). For Haldane, however, not only did the peripheral population constitute a smaller subset of the original species than Wright postulated, but it was always completely isolated from the parent species. Haldane's concept of speciation thus emphasized novelty and diversity rather than linear transformation. In particular, Haldane thought that major clades, at least, tended to emerge abruptly, even if diversification of species within a clade might be more modest or even at times gradual. Taking inspiration from palaeontology and embryology, as well as from developmental genetics, Haldane seems to have been the first evolutionist to express in print a model of rapid speciation followed by periods of taxic stasis (see review in Schwartz 1999b).

Haldane's eclecticism allowed him to meld insights from areas outside of population genetics, which had increasingly become the central focus of evolutionary biology. As such, he, even more so than Wright, who espoused the importance of interactive gene combinations over mutation in producing evolutionary change, saw value in studies that investigated the effects of differential growth and development on the final outcome of the organism and its parts. Among those who caught Haldane's attention were the embryologist and comparative vertebrate anatomist Gavin de Beer and the developmental geneticist Richard Goldschmidt.

De Beer (e.g. 1930) focused on the relation between phylogeny and ontogeny. He argued that evolutionary novelty can result from altering the relative rates at which an individual achieves sexual and somatic maturity. The sum total – ontogeny – results as much from external factors engendering an internal response from the individual as vice versa. As such, it is not that phylogeny drives ontogeny, but rather that ontogeny can impact phylogeny. By not considering the significance of ontogeny, de Beer felt that population geneticists were ignoring the larger evolutionary picture. The study of inheritance only involves following genes through two cellular generations, whereas, he (1930: 20–1) pointed out, "it takes fifty-six generations of cells to reproduce a body like that of a man out of a fertilized egg (itself a single cell), and during these fifty-six generations

the genes are playing their part in company with the external factors in moulding the animal through the successive stages of ontogeny."

Goldschmidt's (1934) interest in ontogeny, which he explored most fully in his studies of nun and gypsy moths, ranged from the genetics of sexual development to the formation of wing venation patterns and the alteration of normal growth by manipulating external factors (e.g. heat). Although he, like de Beer, was impressed by the latter studies, it was from the observation that both male and female genitalia could be induced to develop from the same cellular *primordium* that he concluded that studies in Mendelian genetics did not fully explain evolution and the origin of species. In developing his theory of evolution, Goldschmidt (1940) drew on Dobzhansky's (summarized in 1937) studies on chromosomal rearrangement and hybrid sterility, which had led the latter (1935: 353) to define a species as "a group of individuals fully fertile inter se, but barred from interbreeding with other similar groups by its physiological properties (producing either incompatibility of parents, or sterility of hybrids, or both)." But while Dobzhansky took experiments at the level of population genetics (providing evidence of microevolution) as demonstrating the mechanisms involved in the origin of species (macroevolution), Goldschmidt saw the two processes as being completely different: i.e. the genetics of variation within species and of the origin of species are not the same. The latter would not involve a change at one or two loci, but rather major systematic mutations, which would derive from a rearrangement of genes. Such genetic re-patterning would alter the early phases of ontogeny, which would have a cascading effect on the organism and produce significant novelty within the space of a generation.

Like Bateson and de Beer, Goldschmidt saw regulation of developmental timing as key to evolution, and, further like Bateson, he found his evidence in meristic differences between organisms. Unfortunately, Goldschmidt's portrayal of the bearers of his systemic mutations as "hopeful monsters" and his inability to explain how more than one hopeful monster (and individuals of opposite sexes) would emerge brought as much denigration of his ideas from his detractors (especially Mayr, Dobzhansky) as did his criticisms of Darwin. His primary source of support came, however, not from another geneticist, but from the palaeontologist Otto Schindewolf (1936, 1950), who rejected not only the concepts of natural selection and adaptation, but also the notion that evolution is cumulative rather than involving "profound transformations in the characters of basic organization" (1950: 352). In Schindewolf's interpretation of the fossil records of major clades of organisms – vertebrate and invertebrate, alike – the ancestor of each bore a novelty or suite of novelties that set it distinctly apart from other organisms. The descendants within the clade evolved within the constraints of their ancestor's reorganization. Thus the origin of a clade constituted a significant "leap," whereas the changes within the clade, albeit less saltational, were nonetheless stepwise and not smoothly transformational.

In their *ad hominem* attacks on Goldschmidt, Dobzhansky (1941) and Mayr (1942) crystallized their opinions on species and gradual evolution. Although Dobzhansky (1941) continued to invoke hybrid sterility and physiological isolating mechanisms as attributes of speciation, he had become an ardent gradualist whose vision of evolutionary change was that of accumulating numbers of small mutations over long periods of time. Although, of course, this mode of species change had never been observed – only extrapolated from laboratory studies on generations of fruit flies – Mayr (1942) was not deterred in his conviction that could he identify species, that evolutionary change was gradual, and that he could distinguish the stages of speciation and emergent species: "That speciation is not an abrupt, but a gradual and continuous process is proven by the fact that we find in nature every imaginable level of speciation" (ibid.: 42). [It was only later, in the face of Eldredge and Gould's (1972) model of punctuated equilibria and their emphasis on peripheral isolates and rapid speciation, that Mayr (e.g. 1982), who in 1942 had only briefly noted this possibility, began to emphasize it.]

Combining uniformitarianism, Darwinian gradualism, and Fisherian genetics, Mayr (1942: 70) asserted the following:

> "First, there is available in nature an almost unlimited supply of various kinds of mutations. Second, the variability within the smallest taxonomic units has the same genetic basis as the differences between the subspecies, species, and higher categories. And third, selection, random gene loss, and similar factors, together with isolation, make it possible to explain species formation on the basis of mutability."

For Mayr (1942: 298), the phenomena and processes involved in the origin of species and even higher categories are the same as those that produce intraspecific variation.

Differing from Dobzhansky and Fisher, who emphasized sterility or reproductive failure in their models of species formation, Mayr favoured a simpler means of disrupting interbreeding: geographical or ecological separation. From the foregoing, he (1942: 120) could then articulate his biological species definition: "Species are groups of actually or potentially interbreeding natural populations, which are reproductively isolated from other such groups."

Although G. G. Simpson (e.g. 1944) discussed speciation – adapting Wright's selective topographic landscape to his model of quantum evolution – geneticists felt that palaeontologists had little evolutionary insight to offer on the level of the species and subspecies. In a negative review of Simpson's (1944) monograph, Wright (1945: 417) was very clear on this point: "For the palaeontologist, the distinction between small scale and large scale evolution is rather that between the origins of

genera and of families." But Eldredge and Gould (1972), both paleontologists, did come to propose a model of rapid speciation via peripheral isolates – punctuated equilibria – that was provoked by the ever-present "gaps" in the fossil record that should not exist if evolution proceeded by the gradual accumulation of micromutations. Gould and Eldredge (1979) later suggested that their model of rapid speciation should be appreciated not in terms of the population genetics of structural genes (micromutation), but in terms of changes in regulatory genes that would engender major organismal change (macromutation). The population geneticists Charlesworth, Lande, and Slatkin (1982: 487), however, soundly rejected Gould and Eldredge's proposal:

> "[P]unctutionalists claim that macroevolution is 'decoupled' from microevolution, and deny that gene frequency changes within populations are the foundation of major morphological changes. This argument seems to neglect the fact that every living or fossil organism owes its existence to a continuous line of descent going back generation by generation into the remote past...[T]here is no evidence suggesting the need for qualitatively new mechanisms to account for macroevolutionary patterns."

THE GENETICS OF SPECIATION AND A NEW THEORY

Although Bateson (1894) and de Vries (1900) were the first geneticists to decouple Darwinian natural selection, which they saw only as generating variation within a species, from the origin of species, it was Goldschmidt (1940) who coined "micromutation" and "macromutation" as the genetic counterparts of micro- and macroevolution, the terms he used for the production of variation versus the process of speciation. Although the latter two terms are still used – but often to refer, respectively, to evolution up to and including species and then beyond – micro- and macromutation were soundly rejected by such notables as Dobzhansky (1941) and Mayr (1942) on the grounds that it is unnecessary to invoke two different processes when one will do nicely to explain both the origin of variation within species and the origin of species themselves. Charlesworth *et al.* (1982) reiterated this sentiment.

Clearly, Eldredge and Gould's (1972, also Gould, Eldredge 1979) portrayal of the fossil record as consistently failing to document gradual transitions between morphologically distinguishable species is incompatible with population geneticists' claims that evolutionary change results from the gradual accumulation of small mutations. But is the largely saltational picture of evolution that paleontology provides necessarily incompatible with Mendelian genetics?

The answer to this question is rooted in the history of the field of evolutionary biology. The early generations of geneticists may have been Mendelians, but they were not Darwinians. For them, Mendelism meant that variation was discontinuous and, therefore, so too must be the boundaries between species. De Vries was the first to suggest that the mutations that produced species differed in order of magnitude from those that produced variation between individuals. But it was largely through Morgan *et al.*'s experimental manipulation of wing length and other attributes of fruit flies that Mendelism and Darwinism became fused together to yield the genetic model of gradualism that informed the Evolutionary Synthesis. If, as de Vries and later Goldschmidt claimed, novelty arises abruptly, how could there ever be enough individuals of both sexes for the new species to continue? Gradual evolution allows large numbers of individuals to change collectively while maintaining reproductive compatibility. Thus a species can either become gradually transformed into another or split into populations that will eventually accumulate enough genetic incompatibility that they become separate species. Under the constraints of this model, it seems to make sense that, as proposed by Gould and Eldredge (1979), a change in a regulatory gene would be too profound and singular an event for the mutation either to occur in more than one individual or to be transmittable to others.

Although the early geneticists were compelled to spell out the basics of inheritance, beginning with the realization that mutations tend to arise in, and subsequently spread while still in the recessive state, the post-synthesis language of evolution came to portray a mutation as being immediately available for selection to act on it. Of course, as Fisher (1930) argued, if this were the case, any mutation of consequence would be eliminated. However, not only is this not the case, there is no practical difference between regulatory and structural genes in terms of Mendelian inheritance (Schwartz 1999a,b). For example, as has been demonstrated in the development of the eye and orbit in mice (Mathers *et al.* 1997) and synpolydactyly in humans and mice (Johnson *et al.* 1998, Muragaki *et al.* 1996, Zákány, Duboule 1996), mutations affecting homeobox or any other regulatory genes behave in a straight-forward Mendelian fashion: Heterozygotes phenotypically express the dominant state and homozygotes the recessive condition. When the *Rx* gene was experimentally mutated to the recessive state, heterozygotes were normal in eye and orbit formation, whereas homozygotes lacked both structures. A spontaneous mutation involving the *Hoxd-13* gene in a laboratory colony of mice, which expanded the polyalanine stretch and ultimately caused synpolydactyly, arose, as would be expected, in the recessive state and was eventually expressed in multiple homozygotes.

On the basis of mutations typically arising in the recessive state and not being expressed in a population until homozygotes for it are produced, I (1999a,b) suggested the following model: Prior to the expression of the recessive state, the mutation, which might arise only

in one individual, may spread "silently" through the population until there is a sufficient number of heterozygotes that the production of multiple homozygotes will be a natural occurrence. Clearly, this prediction is borne out in the spontaneous mutation cited above and is no doubt also the explanation for the appearance of eyeless mutants in Morgan's fruit fly colony (Morgan *et al.* 1915).

CONSEQUENCES OF THE THEORY

The evolutionary implications of this model are profound. The spread of a mutation may take some number of generations – that is gradually, although its spread can be accelerated in small populations (Haldane 1932, Wright 1932), as probably happened in the case of Morgan's fruit flies and the colony of synpolydactylous mice cited above – but the expression of the feature or features which they underlie would be instantaneous. If the mutation occurs in a structural gene, the effect on its bearers will be of the sort most population geneticists study (e.g. fruit fly wing length, thorax bristle or segment number, eye colour). This level of genetic change corresponds to the differences we attribute to individual variation within a species. If, however, the mutation affects a regulatory gene, such as a homeobox gene, the effect will be phenomenal, as in the production of synpolydactyly, an eye in a bony orbit, a chordate-like body plan, a tripartite brain, or a lifetime of teeth (Schwartz 1999a,b). A change in homeobox gene activity will, therefore, produce the magnitude of difference that we associate with species.

From this realization emerge two important considerations: 1) Morphology derived from homeobox or any other regulatory gene activity is surely an indicator of evolutionary change, especially at the level of species; and 2) the use of morphology in phylogenetic reconstruction is completely justifiable. Since most molecular phylogenies are based on mitochondrial DNA (which consists only of structural genes), the non-coding or structural gene regions of nuclear DNA, or the products of structural genes, it is no wonder that conflicting phylogenetic hypotheses arise when morphological and molecular data sets are applied to the same taxa. With regulatory genes controlling structural gene activity, changes in the former that result in a species (A) that differs markedly from its closest relative (B) will also be reflected in significant structural gene differences. Thus, to elaborate on an earlier suggestion of Gould and Eldredge's (1979), if the phylogenetic relationships of species B are analyzed only in terms of how similar it is in its structural genes (or structural gene products) to other taxa, it will be united with taxa on the basis of symplesiomorphy. If there is a competing hypothesis based on morphological synapomorphy, it would be the more robust theory of relatedness. While advocating the analysis of regulatory genes for phylogenetic reconstruction, the caveat must be that synapomorphy, not overall similarity, has to be the criterion of relatedness. Otherwise, a species (A) that differs markedly from its closest relative (B) (with which it would share synapomorphy) would not be united with it; rather, taxa that shared a common ancestor with A and B would be united with B because they all retained more unchanged regulatory genes.

Since all multicellular organisms – invertebrate and vertebrate alike – studied to date share many of the same regulatory genes, yet there are clear differences between taxa (e.g. bilaterally versus radial symmetry, exo- versus endoskeletons, antennae versus bony legs, multiple versus deformable single lenses), mutation can take the form of activation versus deactivation of a regulatory gene, in addition to the replication of homeobox genes or the insertion into or deletion from a regulatory gene of a molecular component (Schwartz 1999a). Rather than always being the result of the introduction of new genetic material that then introduces novelty in small pieces, it is more likely that mutation of the sort that we associate with species differences is often the differential expression in combination and timing of extant regulatory genes. Autapomorphy, and if retained in descendants as synapomorphy, therefore, derives from internal regulation.

From this perspective, the dilemma of homoplasy that besets systematists becomes more understandable. Organisms can have the same structures because they have the same regulatory genes – even if they are distantly related and otherwise very different from each other. Certainly, this consideration impacts our concept of homology: Structures that are not themselves present in any common ancestor can be identical in unrelated taxa because the underlying regulatory genes, which are inherited from some common but distant relative, are the same. This possibility does not, however, eliminate the need to test hypotheses of homology. For after all, at some point, the morphologies that organisms share must reflect their evolutionary relationships.

If my argument for justifying the use of morphology in phylogenetic studies is correct – because, at least at the level of species differences, morphology is the end-product of regulatory gene activity – then we can hypothesize that the synapomorphies that would unite a hierarchy of nested clades, while fundamentally a function of mutation affecting regulatory genes in a sequence of ancestors, are reflected in the morphologies whose development these genes help to orchestrate. The task of sorting out synapomorphy from homoplasy – that is, which shared, seemingly derived similarities between taxa are due to recent common ancestry and, therefore, which others are not – would remain as it currently is: the act of testing and either corroborating or falsifying such theories. For example, mice and humans with synpolydactyly have this condition for the same reason: a mutation involving their *Hoxd-13* gene, which expanded the polyalanine encoding region, also orchestrated this ultimate developmental effect. If synpolydactyly is a derived condition within vertebrates (which, after digital reduction from the condition in the Devonian tetrapods *Acanthostega* and *Ichthyostega*, it would seem to be, as, e.g. in mosasaurs and ichthyosaurs), then one could suggest that humans and mice with the condition, and thus with the *Hoxd-13* mutation, are sister taxa. Other morphologies, indicating different potential relationships of *Homo* and *Mus*, obviously contradict this hypothesis.

Another consequence of my theory is that we must rethink Mayr's biological species definition, which is based on members of one species being reproductively incompatible with those of another species. Theoretically, at least, there is no reason why individuals that are homozygous for a mutation affecting a regulatory gene could not mate with individuals of either their own or any preceding or successional generation. Since heterozygotes for a mutation can mate and produce viable offspring with one another as well as with homozygotes for the normal (wild) condition (as in experiments with mice for/without eye and bony orbits and in the case of the spontaneous mutation for synpolydactyly in a colony of mice), then there is nothing genetically that would impede homozygotes for the mutation doing the same with any of these individuals.

If the morphological change – which could actually be anatomical, physiological, or behavioural – emerging in the homozygotes for the mutation were to be sufficiently different in these individuals, then the barrier to reproduction could simply be one of mate recognition (see Paterson 1985). Mutation affecting reproductive organs or behaviour would certainly be a deterrent to mate accessibility. In this context, hybrid zones [e.g. in baboons (Jolly 1993) or crayfish (Roush 1997)] reflect instances where morphological differences that we perceive as being noteworthy are not so to their bearers as far as reproduction is concerned. Although one cannot always reconstruct whether hybrid zones became established because populations that diverged morphologically did not become physically separated from each other, or if once separated populations became sympatric (as in the case of the rusty and blue crayfish), their very existence highlights one of the important implications of my theory: Genetic and concomitant morphological divergence does not necessarily mean that differing populations will be reproductively incompatible.

Another consequence of this theory is that it becomes unnecessary to posit geographical separation as essential to the process of speciation (Schwartz 1999a,b) which is an idea that Endler (1977) had hypothesized, but for different reasons, and which Smith *et al.* (1997) believe to have demonstrated. Essentially, although it has been popular to promote the periphery of a species' range as being the source of selection pressures that could push individuals there genetically and subsequently morphologically away from the rest of the species (e.g. Eldredge, Gould 1972, Mayr 1942, Smith *et al.* 1997), theoretically at least, it matters not if this population is on the periphery of its species' range because the stimulus (e.g. temperature, chemistry, light, or radiation) that provokes the mutation might affect individuals anywhere in the geographical arena of the species. With regard, for example, to Smith *et al.*'s (1997) identification of twelve morphologically and genetically different groups of little greenbul at the periphery (ecotones) of its species, it might simply be the case that the vicissitudes of mate recognition and newly emergent morphologies played a role in the present-day distributions of morphs, rather than that the environments in which they now find themselves provoked their morphological differences. Given the time it would take, even if accelerated through increased inbreeding (e.g. Haldane 1932, Wright 1932), for the mutation to be expressed via homozygotes, one would not expect an adaptive link between the provocation of the mutation and the resultant morphology. Smith *et al.*'s observation that significant gene flow still exists between the twelve morphs and the bulk of the species is consistent with my theory that mutation, leading to morphological novelty, does not preclude interbreeding between the newly emergent homozygotes for the mutation and the extant heterozygotes and homozygotes for the wild alleles.

CONCLUSION

A model of evolution that takes into consideration mutation at the level of structural genes versus mutation at the level of regulatory (especially homeobox genes) can account for both intraspecies variation and the origin of species. Given that each is a gene, but with significantly different effects, a mutation in either, which will most likely arise in the recessive state, will spread "silently" through the population via simple Mendelian mechanisms of inheritance until there is a sufficient number of heterozygotes for the chance production of homozygotes for the mutation. At this point, without geographical isolation as a factor, more than one individual will emerge in the population with the novelty fully expressed. This model is consistent with the fossil record: novelty appears abruptly, not through a long chain of transformation. Since the homozygotes for the mutation can theoretically still interbreed with heterozygotes and homozygotes for the wild allele in the species, other mechanisms – such as species mate recognition – must be involved in the delineation of species. Furthermore, the model provides justification for using morphology in phylogenetic reconstruction and raises questions about the applicability of currently popular molecular analyses for such purposes.

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