

## The dilemma of dominance

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**Abstract.** The concept of dominance poses several dilemmas. First, while entrenched in genetics education, the metaphor of dominance promotes several misconceptions and misleading cultural perspectives. Second, the metaphors of power, prevalence and competition extend into science, shaping assumptions and default concepts. Third, because genetic causality is complex, the simplified concepts of dominance found in practice are highly contingent or inconsistent. The conceptual problems are illustrated in the history of studies on the evolution of dominance. Conceptual clarity may be fostered, I claim, by viewing diploid organisms as *diphenic* and by framing genetic causality modestly through individual alleles and their corresponding *haplophenotypes*.

### Introduction

The revival of Mendel's work just over a century ago was unquestionably a landmark in studying inheritance. However, one new Mendelian concept encountered substantial criticism at the very outset: dominance. Correns (1900), for example, expressed doubt about adopting a universal either-or principle:

I can not understand why DE VRIES assumes that in all pairs of characters which differentiate two strains, one member must always be dominant. Even in peas, where some characters completely conform to this rule, other character pairs are known, in which neither character is dominant, as for instance the color of the seed coat, being either reddish-orange or greenish hyaline. (p. 34)

Erich Tschermak (1900) echoed his concerns:

The appearance of the dominating or the recessive character is not an all or none phenomenon. In individual cases I was sometimes able to establish, with certainty, a simultaneous appearance of both, i.e. transition stages. (p. 44)

William Bateson, while championing Mendel in England, was equally critical on this score:

*Dominance*, as we have seen, is merely a phenomenon incidental to specific cases. (1902: 25)

He too noted blend forms and forms distinct from either parent (p. 25). Early geneticists, then, viewed the concept of dominance as problematic. Indeed, ambivalence towards dominance persisted throughout the century.<sup>1</sup> Today, geneticists finesse the various exceptions and contexts of dominance with a bevy of supplemental concepts and terminology. Still, one may consider how the core concept functions. Here, I extend the early criticisms of dominance in two ways. First, using the perspectives of language, gender and culture, I explore how the metaphor of dominance leads to misconceptions and misperceptions among non-geneticists (Section ‘From student misconceptions to cultural metaphor’), while also shaping assumptions and norms among geneticists themselves (Section ‘The metaphor in science: assumptions and default concepts’). Then I shift focus and revisit whether a concept of dominance can be framed consistently (phenomenologically and causally) (Section ‘Conceptualizing dominance’). A brief history examines how the concept has affected biological practice (Section ‘Reification: dominance and evolution’). Finally, I consider (more speculatively) alternative concepts and terms for talking about phenotypes, genetic traits and genetic causality (Section ‘Beyond dominance’).

Falk (2001) has recently reviewed the history of dominance, suggesting that the concept has waned in the light of molecular biology – especially with the failure to reduce dominance to a consistent molecular mechanism (Wilkie 1994). Yet the concept of dominance still circulates widely in many arenas: medical genetics, genetic counseling, animal and plant breeding, general education and public media. It would seem appropriate, then, to begin by defining what all these persons mean by dominance. However, this is problematic. The meaning varies for individuals and in different discursive contexts. For some, dominance describes traits; for others, it is a property of alleles; for yet others, it describes a relationship between a trio of genotypes and two phenotypes. In some contexts, dominance is construed as absolute; in others, it describes a continuum. Indeed, these disparities in interpretation (Section ‘Conceptualizing dominance’) reflect what I regard as the dilemma of dominance. Let us begin, then, by turning to introductory biology classes, where one might expect to learn about basic genetics and the concept of dominance.

### **From student misconceptions to cultural metaphor**

My point of departure will be, more specifically, various misconceptions in genetics, common among students even at the post-secondary level. These

<sup>1</sup>See Allchin (2002). For example, Thomas Hunt Morgan did not consider dominance a fundamental principle in his synoptic *Theory of the Gene* (1926: 25). In their 1969 textbook, Kroeber et al. reported dominance as one of Mendel’s laws, then promptly noted exceptions and questioned whether the law in fact existed (pp. 412–413).

views are relevant to philosophers, not just educators, for two reasons. First, they illustrate the epistemic influence of terminology. The power of mere language to shape concepts metaphorically has been widely noted (e.g., Lakoff and Johnson 1980), and this case seems no exception. Second, student conceptions reflect how dominance is understood, not among professional geneticists or clinicians, but in the general culture. While philosophers once regarded ‘public understanding of science’ as relevant only to educators, journalists and museum specialists, it now poses significant epistemological challenges. What is or can be known by various epistemic agents and in various discursive communities, especially for non-experts or non-scientists (Goldman 1999)? That is, how does one interpret the foundations of knowledge *demographically*? Characterizing something as ‘known,’ abstractly in passive voice, is not sufficient. One must also consider *who* knows it. To begin, then, I focus on the philosophical dimensions of biological knowledge as expressed in the general culture. Below I will also profile how the misconceptions may reflect thinking even among professional scientists, albeit more subtly (Section ‘The metaphor in science: assumptions and default concepts’).

Consider several student misconceptions about genetics, familiar to anyone who teaches introductory genetics (Donovan 1997; Allchin 2000):

A dominant trait is ‘stronger’ and ‘overpowers’ the recessive trait.

A dominant trait is more likely to be inherited.

A dominant trait is more ‘fit’ in terms of natural selection.

A dominant trait is more prevalent in the population.

‘Wild type’ traits are inherently dominant, while mutants are recessive.

Male or masculine traits are dominant.

Any well trained biologist knows better, of course. However, the errors themselves are not relevant here. Rather, why do these conceptions emerge at all?<sup>2</sup> No one teaches these concepts. They emerge spontaneously and with stunning consistency (regardless of the teacher’s skill.<sup>3</sup>) What is the *archaeology* of the concepts (Foucault 1972), or the nature of the conceptual framework whereby these ideas can arise? In this case, all these misconceptions embody the metaphor of dominance. That is, each reflects interpreting a trait (or allele) as ‘dominating’ in the vernacular sense of power, prevalence or value. Using no more than the metaphoric overtones of one word, students reason (erroneously) about the mechanics of gene expression, the prevalence of alleles, reproductive fitness, heritability, normality and gender. Here, the language itself engenders false beliefs.

<sup>2</sup>Students might well be weaned from these misconceptions –‘eventually’ as Fritz (1999) notes. However, one must first consider why such misconceptions arise at all and why such additional instruction seems necessary, when the basic concepts are so simple. Teachers and texts often alert students to the possible misinterpretations (e.g., Campbell et al. 1999: 248–249; Rothwell 1993: 4–7). They arise nonetheless. The misconceptions are also amazingly recalcitrant to revision.

<sup>3</sup>*These misconceptions do not result from poor instruction.* Blaming teachers misses the point.

Use of the term ‘dominant’ thus constitutes a linguistic metaphor, whereby meanings from non-biological contexts transfer to genetics (Lakoff and Johnson 1980). Two standard meanings are especially relevant: ‘dominant’ as prevailing, or most frequent, and ‘dominant’ as controlling, or powerful. For example, many students infer that dominant traits are more common in the population *because* they are ‘dominant.’ In addition, the term connotes power. In the biological domain, one allele (or trait) is viewed as controlling or suppressing the other. At first, individuals tend to interpret one trait as actively eclipsing the other. Indeed, one can find phrasing or expressions of this type in definitions of dominance in many introductory biology texts (and course websites).<sup>4</sup> The imagery also resonates with and is reinforced by the use of ‘dominance’ in describing social hierarchies in behavioral biology. The extended meanings of dominance foster misleading images. The language itself poses epistemic problems among non-geneticists.

Misconceptions about genetics at a basic level may well fuel concerns about public understanding of science. The growing importance of genetics in personal and social decision-making only amplifies such concerns. One may want to replace the offending term in education and public discourse (Allchin 2000). Alternative language is certainly available (for example, in discussing multiple alleles). However, this poses a dilemma if professionals continue to use the term. Basic terminology intimately links public and professional discourse.

Deeper concerns, however, are raised by the broader role of the metaphor of dominance. The student misconceptions noted above serve primarily to illustrate that the basic metaphor has substantive influence. To probe this topic further, I adopt critical tools and perspectives from feminist and cultural studies (e.g., Haraway 1989). What meaning does dominance embody culturally? I explore two overtones, each based on the metaphoric meaning of power:

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<sup>4</sup>Here is how ‘Biology Online,’ a website study guide sponsored by a life insurance company, defines dominance for students:

When dominant genes were present, they would supersede the presence of wrinkled and were deemed the dominant gene. (Richard Lees, URL: [www.biology-online.org/2/4\\_crossing\\_over.htm](http://www.biology-online.org/2/4_crossing_over.htm), accessed September 19, 2002).

A college instructor posts this for his students:

Dominant - the allele that expresses itself at the expense of an alternate allele; the phenotype that is expressed in the F<sub>1</sub> generation from the cross of two pure lines. (Phillip McClean 2000, URL: [www.ndsu.nodak.edu/instruct/mcclean/plsc431/mendel/mendel1.htm](http://www.ndsu.nodak.edu/instruct/mcclean/plsc431/mendel/mendel1.htm), accessed September 19, 2002)

And here is a teacher’s aid from the U.S. National Institutes of Health:

If the protein that is coded for the “abnormal” phenotype prevails over the ‘normal’ phenotype it is said to be dominant. (Walter Bryant and Lewis Geer, 2001, URL: [www.ncbi.nlm.nih.gov/Structure/people/lewisg/hs.htm](http://www.ncbi.nlm.nih.gov/Structure/people/lewisg/hs.htm), accessed September 19, 2002)

Note the language of ‘superseded,’ ‘prevails’ and ‘at the expense of.’

first, framing the simultaneous behavior of two alleles as either-or competition, and second, the reduction of diverse factors to an all-or-none dichotomy.

First, dominance metaphorically casts genetics in terms of competition. The two traits (or alleles) are framed as exclusive alternatives, where only one will 'dominate' (see note 4). *Either* one trait (or allele) *or* the other is expressed, not both. The metaphor ostensibly excludes the possibilities that both traits/alleles may be expressed in tandem (generating a 'compound' phenotype) or may interact. For Correns, Tshermak and others (Section 'Introduction'), Mendel's concept of dominance implied no intermediates. This conclusion is unwarranted scientifically (further comment below, Section 'The metaphor in science: assumptions and default concepts'), yet the metaphor conveys this impression. Further, nature models society – metaphorically, not rationally or justifiably. Hence, by casting nature in simple dualities, the concept of dominance reflects and reinforces tendencies to interpret *culture* in simple polarities. The scientific concept embeds a cultural notion into the fabric of nature, making it appear 'natural' (Haraway 1989). That is, if one thinks that nature follows this pattern in forming organisms, then one is less likely to imagine cases of collaboration, cooperation or reconciliation in society – or to see them as viable.<sup>5</sup> In decision-making contexts, equal voice or shared authority become 'unnatural.' A feminist perspective here may highlight how the concept is gendered, by supporting competition in a male/masculine mode. Although no geneticist would endorse the metaphor, *de facto* it can help reinforce an image of competitive society, from athletics and TV game shows to politics and international economic relations. Dominance in genetics serves (along with other concepts) as an implicit natural model. Even where one does not imagine a direct and unequivocal influence, the metaphor of dominance has potent overtones. From a feminist and cultural critical perspective, therefore, the metaphor of dominance introduces unwarranted bias.

The dominance metaphor also casts genetics as an either-or dichotomy. Traits and alleles are reduced to two alternatives, the dominant and the recessive. The effect of any single allele, likewise, is all-or-none. For example, in a heterozygous individual, a dominant trait/allele is fully expressed; a recessive trait/allele fully unexpressed (further comments in Section 'Conceptualizing dominance'). Here, again, the focus on simple dualities or dichotomies, when the phenomena described are much richer, reflects a bias readily identified from a feminist perspective.

The metaphor of dominance would be less a concern if the scope of dominance was clearly limited. (This is the case for geneticists, of course.) Indeed, after discussing dominance introductory textbooks typically introduce incomplete dominance, codominance and multiple alleles. Note, however, that

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<sup>5</sup>Conversely, if culture is interpreted competitively, then scientists will tend to interpret nature on that model, or be less likely to see or give relevance to the exceptions indicated in nature.

dominance is typically introduced *first*. Moreover, these other cases are usually called ‘non-Mendelian’ or ‘extensions’ of Mendel’s initial framework. That is, they are cast as *exceptions*, or special cases, outside the norm. Again, there is an implicit normative message about what is normal or natural, and what is abnormal or unnatural. Despite discussion of other alternatives, dominance retains a strong conceptual priority.<sup>6</sup> The basic metaphor persists.

The irony for biologists, of course, is that Mendel was fully aware of ‘non-Mendelian’ cases where no dominance occurs. Before introducing his seven pairs of traits as *dominirende* and *recessive*, he noted: ‘with some of the more striking characters, those, for instance, which relate to the form and size of the leaves, the pubescence of the several parts, etc., the intermediate, indeed, is nearly always to be seen’ (1866: Section 4). Mendel noted other cases in peas: stem length (hybrids were longer, Section ‘Conceptualizing dominance’), seed coat color (hybrids were more frequently spotted, Section ‘Conceptualizing dominance’), flowering time and peduncle length (hybrids were intermediate, Section 8). For Mendel, dominance was not the exclusive norm (see also Falk 2001: 287–288). Indeed, he may have deliberately *narrowed* his focus to traits he could confidently sort dichotomously as dominant and recessive (Di Trocchio 1991). Ironically, biologists tend to cast absence of dominance as *non-Mendelian*. In doing so, they help perpetuate the cultural metaphor of dominance.

The metaphor of dominance, emergent from the terminology alone, is pervasive. To dismiss such analysis as undue fuss about political correctness trivializes the scope of the metaphor in shaping public belief, about both genetics and the nature of culture more broadly. Educators, genetic counselors and anyone discussing genetic diseases in a public forum, in particular, may well be alert to ways they inadvertently mislead others. Alternative language is certainly one easy solution. Equally important, student and public perceptions illustrate how the metaphor is not idle.

### **The metaphor in science: assumptions and default concepts**

Now, does the critique of the metaphor of dominance also extend to science? One might justifiably suppose that mere linguistic analogies would not influence scientists who are steeped in the facts of genetics. Consider, however, the early history of genetics. Hugo de Vries, as an advocate of mutation theory, was certainly predisposed to see traits as discretely dichotomous. Still, his language was hardly neutral. He characterized pairs of Mendelian traits as ‘antagonistic’ (1900: 30–32), reflecting the competitive metaphor embedded in dominance. Other early geneticists interpreted the *term* in this way initially too, prompting the objections of Correns, Tschermak and Bateson (Section

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<sup>6</sup>At more advanced levels, the concepts of expressivity and penetrance help further modify the basic all-or-none model of dominance. Most students never encounter these qualifications.

'Introduction'). Hence, the metaphor emerged in science at the very outset. Genetics has advanced considerably since 1900, of course, and one may wonder if geneticists haven't neutralized the metaphor's effect. Yet a century later, in the pages of *Science*, Lander and Weinberg (2000: 1779) echoed the competitive view in describing Mendel's key insight in almost political terms:

The two factors governing a trait might carry conflicting instructions, in which case the voice of one might dominate in determining the appearance of the individual.

Vestiges of the metaphor persist. The overtones seem inherent in the language. Below I show how the metaphor is present in scientific conceptions, albeit more subtly.

First, the dominance metaphor resonates with assumptions and default conceptualizations about what is basic, or foundational, and what is extraordinary or atypical. Consider, for example, the practice of labeling allele pairs. Allele pairs fit many patterns, of course, besides the dichotomous dominant-recessive model. Yet other patterns are labeled by reference to dominance. The terms used to describe alternatives – *codominance* and *incomplete* dominance – reflect an assumption that either-or dominance is nonetheless primary. As a conceptual root, dominance is foundational. Moreover, the supplemental concepts express a *modification* of dominance, rather than a *negation* or *absence* of dominance. 'Anti-dominance' or 'non-dominance' are not used, for example. In lieu of either-or dominance, one might call hybrid traits 'double' or 'compound.' The traits might be framed as 'independently manifest,' 'coexpressed,' or 'expressed in parallel.' Indeed, both alleles are typically active at the molecular level. The phenotype at higher levels parallels and reflects the nature of this activity. The plausible alternatives highlight how dominance is a *default* pattern. The concept of either-or dominance remains the primary referent, or norm.

Considering either-or dominance foundational might well be justified if, in fact, it was the most common pattern of development in allele pairs. Studies of its relative frequency are rare and typically narrow in scope. As early as 1907, however, C.C. Hurst noted that incomplete dominance was twice as prevalent as complete dominance (Darden 1991: 68). A more recent survey indicates that fewer than one-third of human clinical genetic conditions follow the dominant-recessive pattern (Rodgers 1991). Wright's detailed analysis of coat color in guinea pigs showed only 13 of 29 genes (45%) to exhibit dominance (1968, 1:65). If we accept Di Trocchio's (1991) analysis, Mendel himself found only 7 of 22 traits in peas (32%) exhibiting clearly dichotomous phenotypes. Dominance is a special case, not the most prevalent. Terminology that implicitly casts either-or development as the norm is misleading, while also echoing the prevalence metaphor of dominance.

Contemporary practice in labeling human diseases reflects a slightly different meaning. Here, dominance generally describes any condition found – fully *or* partially – in heterozygotes. The heterozygote need not resemble the homozygote at all, as in the standard textbook characterization. 'Dominant' instead

labels conditions where just one relevant allele is present.<sup>7</sup> ‘Recessive’ conditions are found only where two copies are present. Here, dominance ostensibly refers to the causal efficacy of a single allele (a meaning implied in other contexts, as well). Following now the metaphor of power, dominance is associated with *causal* power. It is active. Recessive alleles, by contrast, are implicitly inert or weak (definitional descriptors may include ‘latent’ or ‘unexpressed’). The metaphor was plainly evident, for example, when R.A. Fisher noted that while labels of dominant and recessive were formally complementary, nevertheless it was more appropriate, with regard to natural processes, to view dominant traits as dominant, rather than recessive traits as recessive (1928a: 115–116). (This interpretation also echoes the once popular presence/absence hypothesis; e.g., Falk 2001: 291–296).

Pragmatically, one seeks simple language to discuss genetically rooted medical conditions. This meaning of dominance, however, tends to overstate the association with the genetic make-up. Here, the genetic causality, rather than the phenotypic effect, is either-or. The trait is caused by *either* the dominant *or* the recessive allele. For example, the language does not readily admit causal contributions from both alleles in heterozygotes, even where their phenotypes differ from homozygotes. That is, in this usage a dominant allele can sometimes ‘cause’ different phenotypes. It depends on the second allele. Again, alternative language may further help highlight the bias of the metaphor. For example, by calling conditions monoallelic or biallelic (e.g., Bix and Locksley 1998), one conveys simply and adequately the number of alleles present. Other labels may well identify causal mechanisms (when known). Haplosufficiency, for example, implicitly explains how two alleles, jointly, act physiologically. (Note, too, how *sufficiency* does not imply causal *necessity*). Framing dominance as a causal property can thus be misleading (see Section ‘Conceptualizing dominance’). Yet it is commensurate with (and perhaps derived from) the metaphor of power in dominance.

The causal asymmetry between two alleles is especially awkward in cases of dominant negative mutants. Here, one allele alone can subvert normal physiological function. For example, collagen is a triple helix protein, derived from two genes. When just one polypeptide chain is altered, the shapes do not fit and the protein cannot assemble. With reduced collagen, the individual becomes acutely susceptible to bone injury. This condition (osteogenesis imperfecta, OMIM #120160) is thus classified as ‘dominant’. Here, the medical view of

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<sup>7</sup>The U.S. National Human Genome Research Institute, for example, gives a definition of dominant as

“a gene that almost always results in a specific physical characteristic, for example, a disease, even though the patient’s genome possesses only one copy. With a dominant gene, the chance of passing on the gene (and therefore the disease) to children is 50–50 in each pregnancy” (URL: [www.genome.gov/glossary.cfm?key=dominant](http://www.genome.gov/glossary.cfm?key=dominant)).

The stated odds clearly indicate that the occurrence of a homozygote with dominant alleles is not considered significant.

dominance implies that the normal allele is eclipsed and causally insignificant in heterozygotes. But a normal protein is produced and is 'vital' to the phenotype. In homozygotes, where no functional peptide chains are produced, the condition is lethal. The normal polypeptide also matches the one in healthy individuals. This similarity is obscured by the dominance label and the either-or metaphor of causal power. The 'negative' label describes the interference by the mutant polypeptide. Everything follows from the negative behavior. Hence, 'dominant negative' mutants are, ultimately, just 'negative' mutants. The 'dominant' label adds nothing.

Another strain of thinking regards dominance in more relative terms. In the spirit of Bateson's 'allelomorphic series' or Muller's (1932) classification, phenotypes are sorted along a physiological continuum from amorphs and hypomorphs to hypermorphs. The dichotomous either-or (sometimes presence/absence) framework of causality is replaced by a physiological scale of degree. Expressing traits on a linear scale from dominant to recessive seems especially apt where proteins catalyze reactions to varying degrees and/or multiple alleles yield varying amounts of metabolic products. Yet this does not exhaust all cases. Here, the metaphor of power persists, merely accommodating levels of causal power. That is, the dominant trait still forms a standard, against which the causal power of all phenotypes is measured. It is also still 'dominant,' in the sense of establishing the functional norm, or baseline of 'full' or 'complete' causality. The recessive is characterized only by its causal efficacy in terms of the dominant. No framework allows the second allele to have unique or separate effects. Consider, for example, malaria resistance as a functional corollary of sickle cell anemia. The sickle cell trait is recessive (on one scale) at the same time malaria resistance is dominant (on another scale) (see also Section 'Conceptualizing dominance'). The linear scale means that the two traits must be assessed on the same axis. Even the more nuanced relative interpretation of dominance reflects the metaphors of power and normality (prevalence).

Each interpretation of the meaning of dominance exhibits traces of the cultural metaphor of dominance, whether of causal power, prevalence or normality, or either-or competition. As a result, each characterization displays its own assumptions, with corresponding conceptual blind spots. Moreover, the various definitions or interpretations do not match each other, although expressed by the same term. These inconsistencies might elicit deep concern about communicating with the term widely. Of course, one might hope to stabilize the meaning of dominance. As I show in the next section this is inherently problematic due to the complexity of genetic causality in diploid organisms. Hence, one may begin to wonder whether any alternative concepts are possible.

### **Conceptualizing dominance**

Discourse in genetics is subtly shaped by the metaphor of dominance (Section 'The metaphor in science: assumptions and default concepts'). The interpretive

elements highlight the general challenge of how one conceptualizes dominance. Here, geneticists do not agree. Assumptions differ, indicating that the concept is not uniform. Each concept is highly contingent or leads easily to inconsistencies. Ultimately, most conceptions are problematic because they simplify dominance or try to reify it (Section 'Reification: dominance and evolution'). While dominance may be clearly observed and described, it cannot be effectively characterized as an entity, nor apart from a suite of many contextual factors. Nor can it do the explanatory work often implied in discourse.

### *Traits and alleles*

The first challenge for an observer is whether dominance refers to traits, alleles, or loosely to both. For some, traits – and *only* traits – can be dominant. For others, alleles – *not* traits – are dominant. Others seem content to talk about dominant traits and dominant alleles. How does one resolve these different perspectives?

First, consider dominance as applied to traits. Mendel certainly used the term this way. This conceptualization (advocates note) draws only on appearances and ascribes no properties to hidden alleles, which cannot be inspected directly. Appearances may be deceiving, however. Or at least complex. Traits are not always reliable benchmarks. Many conditions can be either dominant or recessive, depending on different mutations, even at the same locus (McKusick 1998: *Vol. 1*: page 1, Table 15). Due to such irresolvable ambiguities, Mendelian Inheritance in Man (MIM), the central archive for documenting human genes, discontinued classifying traits as dominant and recessive in 1994 (see OMIM, URL: [www.ncbi.nlm.nih.gov/omim/symbol.html](http://www.ncbi.nlm.nih.gov/omim/symbol.html), note the date May 15, 1994). Editor Victor McKusick explains:

The reason that we stopped classifying autosomal traits as dom. or rec. was that so often the precise same phenotype was dominant when caused by one mutation but recessive when caused by another mutation in the same gene. (personal communication, Jan. 23, 2000)

One cannot always define traits as dominant or recessive consistently. Not much seems at stake in such labeling, however. MIM seems not to have experienced any adverse consequences from abandoning the dominant/recessive classification scheme.

Others adopt the alternative of using *alleles*, not traits, as benchmarks. Many textbooks adopt this approach (Tobin and Duscheck 1998: 220, G-11, G-27; Campbell et al. 1999: 242, 248, G-7, G-19; Lewin et al. 2000; Klug and Cummings 2000: 66), which may be traced back at least to the Morgan school's notational system. Biologists outside the field of genetics commonly speak this way, too. However, alleles are not reliable benchmarks either. An allele can appear to be dominant or recessive, depending on how it is paired with other alleles. A familiar example is the 'A' allele for blood type (OMIM #110300). In

conventional frameworks, type A is dominant to type O, in the sense that the blood type of heterozygotes (AO) corresponds to homozygotes (AA). When A is coupled with type B, by contrast, both alleles are expressed, yielding type AB blood. Here, A and B are ‘codominant.’ There is also a less familiar allele, *cis-AB* (110300.0004). *Cis-AB* yields a protein distinguished by *dual* enzymatic function. It can generate *both* A and B antigens. Hence, *cis-AB* yields type AB blood even when paired with O. When A and *cis-AB* are coupled, then, the phenotype is AB. In this case, A is recessive to *cis-AB*. The very same allele – seeding the type A blood antigen – can be dominant, codominant and recessive, depending on whether it is coupled with the O, B or *cis-AB* allele. The irony, of course, is that the behavior of A remains fundamentally the same in all cases. The A allele yields A antigens. The dominance labels obscure this fundamental similarity. Indeed, even at the level of transmission genetics (that is, without knowing the molecular dynamics), the allele contributes ‘type A-ness’ to the phenotype in each case. Dominance labels add nothing. Blood types illustrate the general rule: *dominance depends on how alleles are paired*. It is not a property inherent in any isolated allele, but rather varies with context.

The promise of labeling either traits or alleles as dominant and recessive is seductive. Establishing a simple if-then formula for linking genotype and phenotype would help organize and streamline reasoning in genetics considerably. Dominance seems to offer that, through a 1:1 identity of (single) alleles and (diploid) phenotypes:

dominant allele      phenotype 1  
recessive allele [×2] phenotype 2

(Note how alleles are typically named by reference to diploid phenotypes.) A system of reference based on either traits or alleles inevitably fails, however, because genetic causality is more complex: the second allele *does* matter. Complete diploid phenotypes do not arise from individual alleles. Philosophers have long contended that dominance can only be articulated fully through a relationship of three diploid genotypes and the two phenotypes that arise from them Hull 1974; Lewontn 1992; Schaffner 1993: 440–442; Falk 2001: 313–314):

AA    phenotype 1  
AA    phenotype 1  
aa    phenotype 2

Conceptually, dominance applies minimally to *both* alleles, specified in particular *pairs*, with *both* their respective phenotypes. Attributing dominance to either alleles or traits truncates this mapping. Contextual features are ascribed inappropriately to a single element. A label of dominance thus cannot be justifiably generalized beyond a specified pair of alleles. Each new pair of alleles must be addressed separately, restricting the domain of dominance.

A simple conceptual scheme that maps individual alleles unerringly to particular phenotypes reflects an ideal that cannot be realized. The concept of dominance cannot accommodate the fundamental observation that (in diploid organisms) phenotypes arise from diploid genotypes, not single alleles. Alleles must be considered in tandem, contrary to the either-or metaphor of dominance.

#### *Other contexts of development*

Correns and others objected a century ago that the scope of dominance was too broad. Such questions about proper scope, or domain, persist, albeit on more finely resolved issues. Here, what constitutes minimal context for conceptualizing dominance consistently?

The first dilemma concerns characterizing phenotype. What is a trait? One cannot always describe all aspects of a genetic condition as uniformly dominant or recessive. A familiar case (mentioned earlier) is sickle cell anemia/malaria resistance (OMIM #141900). If one construes the trait as circulation and crude survival, then functional hemoglobin is dominant. If, however, the trait is malaria resistance, then the non-sickle hemoglobin is recessive. If the trait is, by contrast, oxygen physiology, then functional hemoglobin is incompletely dominant. The same condition (genetically) is dominant, recessive and incompletely dominant simultaneously. That is, pleiotropic dimensions of one gene may exhibit different patterns of dominance. That is, whether a trait is dominant or recessive *depends on how one interprets the trait* (e.g., Rothwell 1993: 5–6). To avoid inconsistency, traits must be conceptualized narrowly. In labeling any case of dominance, geneticists would need to specify, in addition to the three-place relation noted above, the boundaries of the phenotype or trait.

In resolving this inconsistency, one meets the other horn of the dilemma. That is, pleiotropic features as a syndrome become fragmented into distinct traits. They lose their identity as simultaneous consequences of a single, complex (or compound) trait associated with a common gene locus. Ironically, in trying to relate genotype and phenotype, the developmental (or causal) connections become splintered. In the case above, sickle-cell circulation, oxygen physiology and malaria resistance must be addressed independently, although they are intimately related genetically. Dominance thus cannot accommodate the etiological unity of pleiotropic features.

A second dilemma concerns the level of phenotype, where contradictions similar to those just discussed emerge again. Tay Sachs disease (OMIM #272800) is a familiar example. At the gross physiological level, heterozygotes generally survive: the trait is recessive. At the biochemical and cellular level, heterozygotes produce an intermediate amount of the relevant enzyme: incomplete dominance. At the level of transcription and translation, mRNA and polypeptides are produced by both alleles: codominance. Tay Sachs, like

many other conditions, is recessive, incompletely dominant or codominant, *depending on the level of phenotype* (Lewontin 1992: 142–144). One could well privilege one particular level. But in this case, for example, referring to the trait as dominant at the physiological level tends to discount features at the molecular level that are important to diagnosis and treatment. When labeling any case of dominance, then, geneticists would need to specify, in addition to the three genotypes and the particular dimension of the phenotype, also the *level of phenotype*: a five-part relation. So conceptualized, dominance may perhaps begin to seem quite bulky for the simple purposes it is often meant to serve.

Here, achieving consistency regarding dominance introduces the other horn of the dilemma: fragmenting the phenotype yet again. Features characterized at distinct levels become separate traits, rather than parallel unfoldings of the same trait. Their relationship to the same genotype becomes eclipsed. Dominance thus fails to accommodate how multiple phenotypic layers simultaneously reflect one genetic make-up.

A third set of dilemmas concerns further context for the phenotypic outcome of heterozygotes: other genes and several environmental features, such as neighboring cells, intercellular chemical signals and extraorganismal physical conditions. All these elements, as highlighted by genocentric critiques, shape a phenotypic outcome and challenge concepts of simple genetic determinism or causality. They are relevant here, however, for their specific role in altering the apparent dominance relationship for a given pair of alleles. That is, even traits characterized narrowly in the terms noted above might still be dominant *or* recessive, depending on genomic context. Cystic fibrosis (CF, OMIM #219700; also CFTR, #602421), significant as the most frequent single-gene dysfunction among Caucasians, may illustrate. As a recessive disorder, CF should appear only in homozygotes. However, mutation analysis reveals that in about 18% of CF cases ‘only 1 abnormal gene will be identified.’ ‘No abnormal gene can be found in about 1% of CF cases’ (OMIM, URL: [www.ncbi.nlm.nih.gov/80/entrez/dispomim.cgi?id=602421](http://www.ncbi.nlm.nih.gov/80/entrez/dispomim.cgi?id=602421), accessed September 24, 2002). Most heterozygotes are free of the disease, while at the same time other heterozygotes are not. CF seems dominant sometimes. These cases are not yet understood fully, but certain alleles at a second gene locus (R553Q) are known to ameliorate or neutralize a major CF diagnostic symptom. Thus, other genes seem to affect the pattern of dominance at the CF locus (CFTR). CF illustrates the general rule: dominance for a particular allele pair *can depend on the suite of alleles at other gene loci*. Such cases where other genes appear to ‘modify’ dominance have long been documented. Indeed, such dominance modifiers were the focus of sustained evolutionary inquiry and debate (Section ‘Reification: dominance and evolution’). The three-part relationship for dominance that has become the standard for philosophers is thus incomplete. Even 4-part and 5-part relationships delineating the trait and its phenotypic level are too narrow. Dominance labels must also specify, at least in some cases, the genetic make-up at other loci, yielding a 6-part definition.

One can adjust the concept of dominance to fit these additional contexts, as in the previous cases. But it may become unwieldy, far from the simple concept of discourse. There are conceptual consequences here, as well. Dominance assigns priority to one gene locus, casting others as ‘modifiers.’ But such ‘modifiers’ modify the phenotype, not the relationship of the alleles in question (see also Section ‘Reification: dominance and evolution’). ‘Modifiers’ are merely additional gene loci that contribute to the same phenotypic feature. Dominance unjustifiably privileges one locus (as in the simple formula above), obscuring the etiology of features with multiple genetic contributions.

#### *Descriptions and explanations*

Finally, is dominance descriptive or explanatory? That is, does dominance merely *describe* patterns in transmission genetics, or does the relationship *explain* phenotypes? Mendel, of course, was only modestly descriptive. Yet the metaphor of dominance implies power and fosters conceiving dominance as causal (Section ‘The metaphor in science: assumptions and default concepts’) and explanatory. Falk (2001) recently profiled efforts throughout the last century to interpret dominance in explanatory terms. All have failed, he claims, and so he concludes that dominance and recessivity have, once again, become ‘strictly descriptive terms’ (p. 318). Still, many persons – including some philosophers – say that a certain phenotype results *because* one allele or trait is dominant (e.g., Schaffner 1993: 440; Campbell et al. 1999: 242). Particular instances (tokens) of dominance may indeed be interpreted in molecular terms. However, dominance (as a type) has no single general mechanism. The phenotypic pattern may reflect haplosufficiency (for a regulatory protein or a rate-limiting metabolic enzyme), disruption by a toxic protein, exon shuffling, competition for enzyme substrate, interference of multimeric assembly, genomic imprinting, among others (Wilkie 1994: 91). In terms of reduction, the relationship is many–many (Schaffner 1993: 440–445). Dominance cannot serve as shorthand for a consistent causal explanation.

The widespread tendency to treat dominance causally is generally coupled with ascribing dominance to individual alleles. But as the analysis above shows, one allele cannot do the explanatory work of dominance. In particular, both alleles contribute to development. While *differences* in individual alleles may correlate with identifiable phenotypic consequences (Sterelny and Kitcher 1988), traits in diploid organisms are rarely caused by individual alleles *simpliciter* (exceptions may include X-linked genes or other instances of functional monoploidy). A *causal factor* here does not substitute for a *complete causal explanation*. Here, discourse about dominance intersects with notions of genetic causality. Critics of genocentrism and advocates of integrated developmental approaches (e.g., Oyama 2000a, b) may parade inadequacies of dominance explanations as yet another reason to jettison the framework of genetic causality and the genotype–phenotype duality. While I sympathize with

such efforts, my focus here is more modest in scope. For geneticists working with inheritance of certain conditions, crude genetic causality may be a pragmatic heuristic or discursive tool, even if inappropriate elsewhere. Even so, identifying one contributing causal factor is not sufficient. When causal accounts accommodate the role of both alleles, the explanatory role for dominance dissolves.

The examples I have used as illustrations in the foregoing analysis are hardly esoteric. My observations may strike many geneticists as commonplace. In practice, however, geneticists rarely articulate the contexts that elsewhere they might acknowledge are relevant. I hope, in part, to underscore the many unstated, sometimes obscured assumptions. Dominance is conceptually complex, however simple it may appear in discourse or textbooks (Section ‘From student misconceptions to cultural metaphor’). It is highly contingent. One may try to accommodate the various contexts of dominance, but only through substantial conceptual gerrymandering. Philosophically, one may seek concepts, perhaps more narrow in scope, that involve fewer problematic assumptions (Section ‘Beyond dominance’). A concept that guides geneticists in reasoning simply from genes to phenotypes and back again would greatly facilitate reasoning and discourse. Dominance seems to offer that. But the dilemma is that it brings either inconsistencies or a concept unduly constrained by context and background assumptions.

### **Reification: dominance and evolution**

Does the conceptualization of dominance matter to biological practice? Here, I consider debates surrounding the evolution of dominance, noteworthy (perhaps notorious) in many histories of 20th-century biology (Falk 2001: 302–308; Skipper 2002). The history illustrates another dimension of how dominance has commonly been conceptualized: its reification. For example, dominance has frequently been construed as a property that affixes to either traits or alleles, rather than as an incidental feature of certain allele pairs (Section ‘Conceptualizing dominance’). It has been treated as a heritable property, rather than epiphenomenal. It has been regarded as a biologically meaningful adaptation or function, rather than primarily an artifact of perspective.

One contributing factor has likely been the framing of ‘dominance’ as a noun. Mendel used the term *dominirende* thirty times. Following the proper grammatical form, Stern and Sherwood (Mendel 1966) translated the term as the gerund ‘dominating’. By contrast, Druery (1902) translated it as ‘dominant’, subtly closer to ‘dominance’ linguistically. A noun-form never appears in Mendel’s paper. Nonetheless, since de Vries, ‘dominance’ has become a benchmark of Mendelian terminology. The noun form fosters reification. One tends to conceptualize dominance as an object: a concrete entity or property. Moreover, a noun-like identity implies a stable referent that obscures the many contingencies and relativity of dominance (Section ‘Conceptualizing domi-

nance'). Once dominance is reified, it (note the objectified 'it') would seem to express a uniform property or some core essence. One might imagine, for example, that dominant traits were all dominant for the same reason or causal mechanism (Section 'Conceptualizing dominance'). Eventually, one might ask whether dominance evolved. If one instead conceptualized dominance as an incidental feature of certain allele pairs, one might be less inclined to consider it a general or abstract concept.

Reification matters, I contend, for interpreting historical debates about the evolution of dominance. What made these sustained debates possible? Here, my ultimate concern is not how individuals argued for divergent positions (see Falk 2001: 302–308). Rather, what assumptions or conceptual framework enabled the very discourse such that the participants could disagree (Foucault 1972)? All agreed that dominance was something that could meaningfully evolve. I question whether the concept so central to these debates was even necessary.

Consider, first, R.A. Fisher's interpretation of dominance as an evolutionary response to mutations. Some have critiqued Fisher's adaptationist stance. I am concerned instead with his more fundamental posture towards dominance itself – why he would apply his adaptationist views to it. His language reflects strong values about dominance itself. For example, mutant alleles are 'imperfectly' dominant when compared with the wild-type trait, while in the symmetrical case where a mutant is dominant, dominance is instead 'incomplete' (Fisher 1928a: 121, 124). The evolutionary process that establishes the 'normal' viability of a heterozygote is 'pristine' (p. 123). Absence of dominance is a 'lack,' or deficiency (p. 124). Wild-type genes, 'in their dominance' are 'clearly of a different nature' than the recessive traits they give rise to (although Fisher did not specify how, p. 116). The dominant wild traits have positive connotation; the recessive mutant, negative. The metaphor of dominance, in multiple terms of power, value and prevalence, is evident. For Fisher, a eugenicist who endorsed stratified social classes, dominance might seem like a natural mechanism for actively shaping genetic influence. While Fisher presented his ideas in the context of a theoretical problem, 'how do new mutants fare in an evolving population?', his puzzle emerges only when one first regards mutations as disadvantageous (pp. 118–119). Fisher clearly valued dominance as a regulator or control of threats to the organism's integrity or fitness. For Fisher, dominance was a thing: an observable force in the 'politics' of the genes.

Fisher (1928a) 'solved' his puzzle through dominance modifiers. For Fisher, the modifiers could alter the expression of heterozygotes, without the alleles themselves changing. But Fisher did not discuss the concerted effect of many independent genes, each acting directly on the phenotype. Rather, he framed his solution in terms of subsidiary genes modifying the dominance at a primary gene locus. Fisher's proposal thus reflected an important assumption: dominance had been abstracted as a heritable property *distinct* from the constituent alleles. That separate property, not the alleles themselves, would

be subject to natural selection. Fisher's notion of dominance modifiers extended the conception of dominance: it added something more to gene expression than the paired alleles themselves. Fisher's dominance modifiers helped reify dominance more deeply.

Fisher (1928b) soon presented his 'decisive evidence' for modifiers (p. 571). If different lineages were hybridized, one could sometimes observe 'breakdown of dominance' in the second generation. Simple Mendelian ratios disappeared. All 'degrees' of dominance were visible. For Fisher, this showed how multiple modifiers, each uniquely evolved in their own lineage, had segregated and how their moderating influence was diluted or lost. However, Fisher did not consider plain polygenic inheritance as a possible explanation. That is, Fisher did not distinguish between modifiers, which would affect dominance, and other genes which contributed to the phenotype directly (also see Falk 2001: 304–305). At the same time, he misplaced the property by attributing the causal role of the modifier locus to the dominance of the locus in question. Earlier geneticists had acknowledged that many genes could affect certain phenotypes and that as they evolved they altered apparent 'dominance,' viewed at one locus. But Fisher never showed that any of the 'modifying' genes *functioned* specifically to *modify dominance* rather than the trait itself (Caplan and Pigliucci 2001). That is, he did not distinguish between *selection of* dominance modifiers and *selection for* dominance modification (Sober 1984: Section 3.2). Fisher's evidence, far from being 'decisive,' was shaped by his reification of dominance.

Fisher's claims about the evolution of dominance precipitated a sustained debate with Sewall Wright, widely recounted since (e.g., Provine 1985, 1986: Chap. 8).<sup>8</sup> Wright's 'physiological' theory, now largely accepted as closer to the mark, stressed shifts in dominance as a result of selection on individual alleles. He usually regarded dominance as epiphenomenal. In disagreeing with Fisher, however, Wright seemed to agree that dominance was biologically significant, could be explained, and could meaningfully evolve. Moreover, given the historical emphasis on the vehement disagreement, one might assume that Wright rejected Fisher's notion of dominance modifiers. But he did not. He merely disagreed about their prevalence and role. Thus, Wright shared Fisher's concept of dominance as a thing that *could* be modified by external factors (Provine 1986: 246). For example, Wright distinguished between dominance modifiers for favorable traits and those for deleterious ones. He even discussed whether the modifiers themselves were dominant or recessive, indicating that they might evolve, as well (1969, 2: 69–71; Provine 1986: 247–248). Even Wright seemed sometimes to treat dominance as a relevant, modifiable property.

E.B. Ford, leader of the influential Oxford school of ecological genetics, followed Fisher's theorizing on dominance (1964: 87–89, 120–121, 127,

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<sup>8</sup>The debate lingers (e.g., Mayo and Bürger 1997; Otto and Bourguet 1999), although many regard Orr's (1991) experiment with haploid and diploid strains of *Chlamydomonas* as a definitive 'crucial experiment.'

269–271). Ford focused on Fisher’s notion that modifiers could eventually make a recessive mutant dominant if it was selectively advantageous. Even pleiotropic features, he claimed, would be modified separately:

Selection will tend to make the favourable effects of the gene dominant and the unfavourable ones recessive. (p. 87)

Ford also endeavored to ground the new genetic interpretations of evolution in studies of natural populations. In (1955) he reported on a presumed modifier for melanism in the lesser yellow underwing moth, *Triphaena comes*. Ford’s evidence became part of the canon on dominance modifiers reported by others in texts (e.g., Berry 1965: 117–119) and therefore deserves careful examination. Ford searched, as Fisher had earlier, for breakdown of dominance. He crossed individuals from isolated populations of moths on the Barra and Orkney Islands. In doing so, Ford assumed that the genetics of coloration reduced to two alleles (1955: 257), even though the existence of several local forms indicated substantial genetic variation. Ford also assumed that the alleles were similar among his two isolated populations (although he had considered the possibility of two distinct gene loci, p. 261). When Ford crossed heterozygote forms from the two locales, a spectrum of forms appeared (B.O.IV., p. 258). Ford interpreted this as breakdown of dominance, due to modifier genes losing their locally adapted influence. Similar spectrums appeared, however, within the same generation of broods from each local population (B.IV.,O.IV., p. 258) – that is, *the same result occurred without hybridization*. Moreover, by Ford’s interpretation, he would have expected a flattened 1:2:1 (p. 261), with substantially more intermediates (type *curtisii*-to-*comes*). But the distribution was almost flat – a pattern possibly produced by recombining four distinct alleles at the same locus from two heterozygous parents (a 1:1:1:1 ratio). Ford did not exclude these interpretations of his results. Nor did Ford discuss the prospective segregation and recombination of the purported modifier genes. In these key crosses, each pigmentation allele would be accompanied by one allele from each corresponding modifier locus (having drawn the F<sub>1</sub> heterozygotes from each local population). Their partial effect (on 1 their ‘own’ pigment alleles) is never addressed. The impression (here, as elsewhere) is that the ‘failure’ of dominance modifiers in hybrids leads to an inchoate ‘unclassifiable series’ (p. 260), rather than discrete cases of non-dominance. (Note, too, how the lack of dominance is associated with chaos, resonating with the metaphor of power.) Ford (like Fisher) did not exclude other explanations. One could also interpret the results through multiple alleles or other genes that affected the phenotype directly (rather than by affecting the differential behavior of the original two alleles). In not addressing these possibilities, Ford exhibited his assumptions about dominance, its priority and its nature as something concrete that could experience ‘breakdown.’

Bernard Kettlewell, widely renowned for his field studies of peppered moths, in turn followed Ford. He assumed that selectively advantageous traits would become dominant. Thus he puzzled why melanistic moths were dominant in

populations from non-polluted environments (Ford 1964: 269). (He thereby echoed confusions about prevalence, fitness and dominance found among biology students today, Section 'From student misconceptions to cultural metaphor'.) Kettlewell, too, crossed forms from distant localities to expose the supposed effect of dominance modifiers accumulated in isolated populations. He, too, reported 'breakdown of dominance' (1973: Plate 19.1).<sup>9</sup> However, he never distinguished clearly the ostensive effects of dominance modifiers from standard polygenic inheritance.

Kettlewell certainly fostered an impression that the genetics of melanism in peppered moths was 'black and white.' He typically portrayed only two forms, resulting from dominant and recessive alleles (1959: 49–51, 1973: 106). But Kettlewell also tallied intermediate phenotypes (type *insularia*; 1973: Plate 9.1 and Fig. 9.1, p. 135), 'specimens grading imperceptibly from light to black' (1973: 106). These forms he did not attribute to the variable effects of dominance modifiers. (Why not?) Here too, Kettlewell rejected the signals of multiple genes, postulating instead many alleles with dominance in a 'descending scale, from the darkest to the lightest' (p. 107; Majerus 1998: 119–121). He did not address how the modifiers he identified elsewhere would work with so many different alleles.

Some geneticists continue to cite the 'ample evidence' on dominance modifiers (cf., Mayo and Bürger 1997: 101–102; Otto and Bourguet 1999: 562). All these cases seem to involve pigmentation and coloration – appropriate candidates for multiple alleles and/or independent genes affecting a common feature. While the results may seem consistent with the existence of dominance modifiers, they do not exclude other interpretations. Often cited in this vein is Clarke and Sheppard's (1960, 1963) work on mimetic polymorphism in *Papilio* (swallowtail butterfly). In their early study, they echoed Fisher's language about the evolution of dominance. Later, such language disappeared. Instead, they discussed coadapted gene complexes and 'polygenes,' not once referring to dominance. The paradox of dominance modifiers was well illustrated in their earlier observation that:

modifiers for the reduction of a character (tail length) can also lead to the evolution of dominance even in the absence of the character (taillessness) whose dominance is being affected. (Clarke and Sheppard 1960: 85)

Of course, modifier genes can hardly be selected for in the absence of the trait they modify. Yet they can evolve for other reasons. Genes can evolve independently of other genes. Here, the concept of dominance modifiers seems redundant. Even in discussion of transmission genetics (that is, molecular mechanisms aside), what matters are the gene loci, their alleles and the ultimate phenotype, not the differential relationship of a specific trait or allele pair.

<sup>9</sup>West (1977) and Mikkola (1984) were not able to replicate Kettlewell's results on breakdown of dominance.

In the five cases above – all considered landmarks – dominance is an unnecessary assumption or interpretive element. While ostensibly central to these debates, the concept of dominance was ultimately redundant. Throughout, dominance was reified apart from or beyond the alleles themselves. Hence, evolutionary biologists might be led to conflate selection of dominance with selection for dominance (Sober 1984). Selection for dominance (as a special relation between two traits or alleles) cannot be teased out from selection for phenotypes. Reifying dominance, even in the physiological approach, gives the outcome unwarranted significance. One may debate whether dominance evolves, but the question only becomes relevant when one first reifies dominance. The historical debate on the evolution of dominance, extending over more than half a century, thus reflects how the conceptualization of dominance has shaped biological practice.

### **Beyond dominance**

With the conceptual analysis (Sections ‘The metaphor in science: assumptions and default concepts’ and ‘Conceptualizing dominance’) and the review of the history of evolution of dominance (Section ‘Reification: dominance and evolution’), one may wonder whether the concept of dominance is pragmatically coherent and, ultimately, fruitful. I will briefly sketch two elements of a possible alternative conceptualization: how to begin characterizing the genotype–phenotype relationship and how to delineate traits.

The first problem posed by dominance is how to address the highly contextual relationship between genotype and phenotype: crudely, how one frames genetic causality. Dominance embodies the linear, billiard-ball model of causality, which emphasizes singular causes, thereby obscuring context and overstating effects (Section ‘Conceptualizing dominance’). Hence, much philosophical critique of genetic determinism. A more modest sense of causality focuses on *causal factors*, rather than complete *causes*. Dominance raises the special problem of articulating the role of two parallel genomes. If one allele does not cause the full phenotype, but contributes causally, then perhaps one should focus more narrowly just on its partial causal contribution or potential. For example, one may link each single allele with its causal potential:

allele D   haplophenotype potential D/

Here, the potential effect of a single allele is not overstated (compare to formulae in Section ‘The metaphor in science: assumptions and default concepts’). It does not yield a full phenotype on its own. It contributes to only a ‘half-phenotype,’ or *haplophenotype*.

This approach entails a major reconceptualization of phenotype. The either-or framework of dominance highlights how phenotype is currently conceived as *singular* or *homogeneous*. Yet a full diploid phenotype is *double*, just like the

genotype. That is, alleles are active in pairs. Diploid organisms are essentially *diphenic*. Each allele functions partly independently, in parallel with its homolog. A phenotype is thus *compound* – two traits expressed *in tandem* and variously overlapping or intersecting.

A challenge remains, of course, in characterizing how two haplophenotypes combine. Language based on dominance assumes a causal asymmetry as a default: one phenotype implies only one of the two alleles/traits is expressed (Section ‘The metaphor in science: assumptions and default concepts’). For most cases, this is biologically misleading. Namely, each allele is generally active. Cases where one allelic homolog is wholly suppressed are possible: through promoter mutations or regulatory mechanisms (such as in immune cells). However, they prove the exception, not the rule. At the level of transmission genetics, therefore, the appropriate null hypothesis is dual phenotypic development. One thus interprets haplophenotypes as overlapping or coextant, not competing. The diploid organism is *diphenic*.

How does one conceptualize haplophenotype, since it is not directly observable in organisms? Mendel characterized his traits by referring to true-breeding strains as benchmarks. Geneticists today tend follow the same model, characterizing *single* alleles based on *diploid* phenotypes. Here, focus shifts from determinism to potentiality in a multifactorial system. A haplophenotype maps the branch of potential causal threads associated with the allele. As a *haplophenotype*, it may be clear that it cannot yield a complete causal account. Namely, ‘where is the other half?’ Characterizing alleles in terms of partial contributions and potentials may thereby help further dissolve the image of genetic determinism.

The haplophenotype concept functions accommodates well cases where the number of alleles in the genotype is important. That is, some conditions are monoploid or triploid or associated with specific forms of aneuploidy. Some conditions are triallelic (e.g., Katsanis et al. 2001). Some alleles are expressed differently when in single vs. double copies (see Rodgers 1991: 5). Some loci exhibit gene imprinting. The concept of dominance does not apply well in these instances. Haplophenotype units, however, seem well suited to such discourse. Discussions of dominance function most smoothly only when the two alleles may be assumed and one can avoid the burden of specifying context (Section ‘Conceptualizing dominance’). As the number of alleles (and recombinations increase), however, it becomes increasingly cumbersome. The economy of the language of dominance evaporates. Reducing discussion of diploid genotypes to their component haplophenotype units thus seems especially appropriate where allelic variation is high and gene pairings are highly variable.

Conceptualizing diploid organisms as diphenic and treating each allele causally as a haplophenotype potential poses a complementary challenge: how to conceptualize traits. Currently, a genetic trait seems to be whatever feature is deemed relevant for the occasion. This can potentially disguise important genetic or developmental relationships and possibly other relevant genes. Thus, one problem of conceptualizing dominance is that it tends to fragment a unified

phenotypic ensemble into separate traits (Section 'Conceptualizing dominance'). Ideally, genetic traits reflect genetic etiology (to the extent possible), thereby facilitating reasoning from phenotype to genotype. For example, pleiotropic traits are appropriately regarded genetically as *one* complex trait. A genetic *trait* may thus be a syndrome with many discernible phenotypic *features*. While encompassing many parts or causal threads, it is still one trait based on its genetic roots. For example, the cystic fibrosis transmembrane conductance regulator gene (CFTR, OMIM#602421) yields a protein that functions as a chloride channel. The ultimate effect differs in pancreatic, sweat gland and mucosal cells (among others), and dysfunctional alleles lead to particular pathologies in each cell type. In the same way, genetic traits should not be viewed as restricted to one level of organization or development. All the levels occur at once and parallel each other, even if one level is of immediate concern. The trait should thus be viewed as a developmental cross-section including all levels of phenotype *simultaneously*. A genetic trait (or haplophenotype) may thus be conceived as a richly branching tree of potentiality, rooted in a particular gene (problems defining 'a' gene notwithstanding).

This approach to delineating genetic traits gives central place to etiology. Its reduction of the organism's phenotype may thus differ substantially from one founded just on the observer's interest. One virtue of the ill-fated presence/absence hypothesis was that it highlighted the 'additive' nature of a gene product, in contrast to the apparent end-state. For example, a gene might provide (or not provide) potential for green pigment in pea seeds. This contrasted with specifying the seed-color trait as green vs. yellow. The baseline yellow is appropriately characterized as due to other gene loci. An etiological approach to delineating traits echoes this theme by specifying just what the gene can contribute causally to the organism (see Schwartz 2002). Thus, sickle cell hemoglobin and cystic fibrosis, among others, are best viewed as syndromes. One might endorse a *trait/feature distinction*, where the trait reflects etiology and the feature is the immediately relevant fragment of phenotype. It may seem awkward, therefore, to speak of polygenic *genetic* traits. While many *traits* may contribute to an observable *feature*, the ideal would be to parse any feature into its suite of genetic components, each regarded more properly as a genetic trait. A solution to the many awkward dimensions of dominance (Section 'Conceptualizing dominance') may be, in part, guided by a rigorously etiological approach to genetic traits.

The value of any alternative conceptualization ultimately depends on how one views the dilemma of dominance. For example, many geneticists respond adversely to any hint that the concept of dominance may be less than ideal. The name of Mendel is often invoked (e.g., Fritz 1999), suggesting that deference to Mendel may shape assessment of the concept (Allchin 2002, 2003). Perhaps conceptual clarity is not needed pragmatically. Geneticists in various professional contexts may feel they encounter no problem in everyday practice and that the cultural overtones are irrelevant to them. Some acknowledge that dominance is not the most ideal term, but that 'it is 150 years too late.' Some

contend that students are better served by using the established technical jargon. By this reasoning, of course, biologists would still be talking about pangenes, mesosomes, soluble RNA, etc. The current notion of dominance is steeped in a culturally and scientifically inappropriate metaphor (Sections ‘From student misconceptions to cultural metaphor’ and ‘The metaphor in science: assumptions and default concepts’), leads to inconsistencies or awkward contingencies (Section ‘Conceptualizing dominance’), and has been misleading historically (Section ‘Reification: dominance and evolution’). Concepts such as diphenism, haplophenotype and a feature/trait distinction can be introduced alongside existing concepts, even if they might ultimately displace them. The language of dominance might readily dissolve. As I see it, even small changes in conceptualization and language have potentially profound consequences, for both science and culture.

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### References

- Allchin D. 2000. Mending mendelism. *Am. Biol. Teacher* 62: 632–639.
- Allchin D. 2002. Dissolving dominance. In: Parker L. and Ankeny R. (eds), *Mutating Concepts, Evolving Disciplines: Genetics, Medicine, and Society*. Kluwer, Dordrecht, pp. 43–62.
- Allchin D. 2003. Scientific Myth-conceptions. *Science Education* 87: 329–351.
- Bateson W. [1902] 1928. The problems of heredity and their solution. In: Punnett R.C. (ed.), *Scientific Papers of William Bateson*. Cambridge University Press, Cambridge, UK, pp. 4–28.
- Berry R.J. 1965. *Teach Yourself Genetics*. English Universities Press, London.
- Bix M. and Locksley R.M. 1998. Independent and Epigenetic Regulation of the Interleukin-4 Alleles in CD4<sup>+</sup> T cells. *Science* 281: 1352–1354.
- Campbell N., Reece J.B. and Mitchell L.G. 1999. *Biology*, 5th ed. Benjamin Cummings, Menlo Park, CA.
- Caplan J.M. and Pigliucci M. 2001. Genes “For” phenotypes: a modern history view. *Biol. Philos.* 16: 189–213.
- Clarke C.A. and Sheppard P.M. 1960. The evolution of dominance under disruptive selection. *Heredity* 14: 73–87.
- Clarke C.A. and Sheppard P.M. 1963. Interactions between major genes and polygenes in the determination of the mimetic patterns of *Papilio dardanus*. *Evolution* 17: 404–413.
- Correns C.G. [1900] 1950. Mendel’s law concerning the behavior of progeny in varietal hybrids (Eng. trans.), *Genetics* 25(5,2): 33–41.
- Darden L. 1991. *Theory Change in Science: Strategies from Mendelian Genetics*. Oxford University Press, Oxford.

- de Vries H. [1900] 1950. Concerning the law of segregation of hybrids. *Genetics* 35(5,2): 30–32.
- Di Trocchio F. 1991. Mendel's experiments: A reinterpretation. *J. Hist. Biol.* 24: 485–519.
- Donovan M.P. 1997. The vocabulary of biology and the problem of semantics. *J. College Sci. Teach.* 26: 381–382.
- Druery C.T. 1902. Experiments in plant hybridisation [translation of Mendel]. *J. Roy. Hort. Soc.* 26: 1–32. Also available at URL: [www.netspace.org/MendelWeb/Mendel.plain.html](http://www.netspace.org/MendelWeb/Mendel.plain.html) (updated Feb 22, 1997; accessed Dec 15, 2000).
- Falk R. 2001. The rise and fall of dominance. *Biol. Philos.* 16: 285–323.
- Fisher R.A. 1928a. The possible modification of the response of the wild type to recurrent mutations. *Am. Nat.* 62: 115–126.
- Fisher R.A. 1928b. Two further notes on the origin of dominance. *Am. Nat.* 62: 571–574.
- Ford E.B. 1955. Polymorphism and taxonomy. *Heredity* 9: 255–264.
- Ford E.B. 1964. *Ecology Genetics*. Methuen, London.
- Foucault M. 1972. *The Archaeology of Knowledge*. Pantheon, New York.
- Fritz G. 1999. Allchin's mistake. *BioQUEST Notes* 10(1): 9–12.
- Goldman A.I. 1999. *Knowledge in a Social World*. Oxford, Clarendon Press.
- Haraway D. 1989. *Primate Visions*. Routledge, New York.
- Hull D. 1974. *Philosophy of Biological Science*. Prentice Hall, Englewood Cliffs, NJ.
- Katsanis N., Ansley S.J., Badano J.L., Eichers E.R., Lewis R.L., Hoskins B.E., Scambler P.J., Davidson W.S., Beales P.L. and Lupski J.R. 2001. Triallelic inheritance in Bardet-Biedl syndrome, a Mendelian recessive disorder. *Science* 293: 2256–2259.
- Kettlewell H.B.D. 1959. Darwin's missing evidence. *Sci. Am.* 200(March): 48–53.
- Kettlewell H.B.D. 1973. *The Evolution of Melanism: The Study of Recurring Necessity; with Special Reference to Industrial Melanism in the Lepidoptera*. Clarendon Press, Oxford.
- Klug W. and Cummings M. 2000. *Concepts of Genetics*, 6th ed.. Prentice Hall, Upper Saddle River, NJ.
- Kroeber E., Wolff W.H. and Weaver R.L. 1969. *Biology*, 2d ed. D.C. Heath, Lexington, MA.
- Lakoff G. and Johnson M. 1980. *Metaphors We Live By*. The University of Chicago Press, Chicago, IL.
- Lander E.S. and Weinberg R.A. 2000. Genomics: journey to the center of biology. *Science* 287: 1777–1781.
- Lewin B. 2000. *Genes VII*. Oxford University Press, New York.
- Lewontin R.C. 1992. Genotype and Phenotype. In: Keller E. and Lloyd E.A. (eds), *Keywords in Evolutionary Biology*. Harvard University Press, Cambridge, MA, pp. 137–144.
- Majerus M.E.N. 1998. *Melanism: Evolution in Action*. Oxford University Press, Oxford.
- Mayo O. and Bürger R. 1997. The evolution of dominance: a theory whose time has passed? *Biol. Rev.* 72: 97–110.
- McKusick V.A. 1998. *Mendelian Inheritance in Man: A Catalog of Human Genes and Genetic Disorders*, 12th ed. Johns Hopkins University Press, Baltimore, MD.
- Mendel G. [1866] 1966. Versuche über Pflanzenhybriden [experiments on plant hybrids]. In: English translation in Stern C. and Sherwood E. (eds), *The Origin of Genetics: A Mendel Source Book*. W.H. Freeman, San Francisco, pp. 1–48.
- Mikkola K. 1984. Dominance relations among the melanic forms of *biston betularia* and *Odonoptera bidentata* (Lepidoptera, Geometridae). *Heredity* 52: 9–16.
- Morgan T.H. 1926. *The Theory of the Gene*. Yale University Press, New Haven, CT.
- Muller H.J. 1932. Further studies on the nature and causes of gene mutations. In: Jones D.F. (ed.), *Proceedings of the Sixth International Congress of Genetics*. Brooklyn Botanical Garden, Menasha, WI, pp. 213–255.
- Olby R.C. 1997. Mendel, Mendelism and Genetics. MendelWeb, URL: [www.netspace.org/MendelWeb/MWolby.intro.html](http://www.netspace.org/MendelWeb/MWolby.intro.html) (updated Feb 22, 1997; accessed Dec 15, 2000).
- OMIM™ 2000. Online Mendelian Inheritance in Man, McKusick-Nathans Institute for Genetic Medicine. Johns Hopkins University (Baltimore, MD) and National Center for Biotechnology

- Information, National Library of Medicine, Bethesda, MD. URL: [www.ncbi.nlm.nih.gov/omim/](http://www.ncbi.nlm.nih.gov/omim/) (accessed Dec 15, 2000).
- Orr H.A. 1991. A test of Fisher's theory of dominance. *Proc. Natl. Acad. Sci. USA* 88: 11413–11415.
- Otto S.P. and Bourguet D. 1999. Balanced polymorphism and the evolution of dominance. *Am. Nat.* 153: 561–574.
- Oyama S. 2000a. *The Ontogeny of Information*, 2d ed. Duke University Press, Durham, NC.
- Oyama S. 2000b. *Evolution's Eye*. Duke University Press, Durham, NC.
- Provine W.B. 1985. The R.A. Fisher-Sewall Wright controversy and its influence upon modern evolutionary biology. In: Dawkins R. and Ridley M. (eds), *Oxford Surveys in Evolutionary Biology*. Oxford University Press, Oxford, 2: 197–219.
- Provine W.B. 1986. *Sewall Wright and Evolutionary Biology*. The University of Chicago Press, Chicago, IL.
- Rodgers J. 1991. Mechanisms Mendel never knew. *Mosaic* 22(3): 2–11.
- Rothwell N.V. 1993. *Understanding Genetics: A Molecular Approach*. Wiley-Liss, New York.
- Schaffner K. 1993. *Discovery and Explanation in Biology and Medicine*. The University of Chicago Press, Chicago, IL.
- Schwartz S. 2002. Characters as units and the case of the presence and absence hypothesis. *Biol. Philos.* 17: 369–388.
- Skipper R.A. Jr. 2002. The persistence of the R.A. Fisher-Sewall Wright controversy. *Biol. Philos.* 17: 341–367.
- Sober E. 1984. *The Nature of Selection*. MIT Press, Cambridge, MA.
- Sterelny K. and Kitcher P. 1988. The return of the gene. *J. Philos.* 85: 339–361.
- Tobin A.J. and Duscheck J. 1998. *Asking About Life*. Saunders, Fort Worth, TX.
- Tschermak E. [1900] 1950. Concerning Artificial crossing in *Pisum sativum*. *Genetics* 35(5,2): 42–47.
- West D.A. 1977. Melanism in biston (*Lepidoptera: Geometridae*) in the rural central Appalachians. *Heredity* 39: 75–81.
- Wilkie A.O.M. 1994. The molecular basis of genetic dominance. *J. Med. Genet.* 31: 89–98.
- Wright S. 1968–69. *Evolution and the Genetics of Populations*, Vols. 1–2 The University of Chicago Press, Chicago, IL.