Are Languages Really Independent from Genes? If Not, What Would a Genetic Bias Affecting Language Diversity Look Like?

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Are Languages Really Independent from Genes? If Not, What Would a Genetic Bias Affecting Language Diversity Look Like?

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Abstract It is generally accepted that the relationship between human genes and language is very complex and multifaceted. This has its roots in the "regular" complexity governing the interplay among genes and between genes and environment for most phenotypes, but with the added layer of supra-ontogenetic and supra-individual processes defining culture. At the coarsest level, focusing on the species, it is clear that human-specific—but not necessarily faculty-specific—genetic factors subtend our capacity for language and a currently very productive research program is aiming at uncovering them. At the other end of the spectrum, it is uncontroversial that individual-level variations in different aspects related to speech and language have an important genetic component and their discovery and detailed characterization have already started to revolutionize the way we think about human nature. However, at the intermediate, glossogenetic/population level, the relationship becomes controversial, partly due to deeply ingrained beliefs about language acquisition and universality and partly because of confusions with a different type of gene-languages correlation due to shared history. Nevertheless, conceptual, mathematical and computational models—and, recently, experimental evidence from artificial languages and songbirds—have repeatedly shown that genetic biases affecting the acquisition or processing of aspects of language and speech can be amplified by population-level intergenerational cultural processes and made manifest either as fixed "universal" properties of language or as structured linguistic diversity. Here, I review several such models as well as the recently proposed case of a causal relationship between the distribution of tone languages and two genes related to brain growth and development, *ASPM* and *Microcephalin*, and I discuss the relevance of such genetic biasing for language evolution, change, and diversity.

The relationship between language and genes is extremely complex, and this article aims to analyze some of the reasons for this complexity, viewing it on three interacting organizational and temporal levels (D. Dediu, unpublished): the

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phylogenetic/species, the glossogenetic/population, and the ontogenetic/individual. In particular, I focus on the intermediate level, involving human groups across several generations, representing the level at which the processes relevant for the dynamics of languages and dialects take place, insisting on the concept of *genetically biased language change*. I provide supporting evidence from conceptual, mathematical and computational models, recent experimental results from human artificial language learning and song birds, as well as the "real-world" association between linguistic typology and population gene frequencies, and I argue that not only are such genetic biases affecting the cultural transmission of language active and relevant for some obscure corners of science but also that they are essential for a proper understanding of language evolution, change, and diversity.

Human Language Is a Multilevel Phenomenon

Language is an extremely complex phenomenon ranging across multiple levels on several dimensions, from the individual acquiring his or her native language and the real-time processes involved in producing and understanding it to the synchronic distribution of typological features across today's approximately 7,000 languages and the diachronic processes of language change and evolution. This set of problems requires collaboration across several disciplines, including philosophy, psychology, psycholinguistics, linguistics, anthropology, sociology, neuroscience, biology, and mathematics, to name just a few. But to date, despite intense efforts, fundamental problems still persist.

Even if, as in any other scientific fields, factors pertaining to historical accident and the psychology and sociology of science have played an important role, the main reason for this resilience to understanding resides in the intrinsic properties of the phenomenon under study, which is manifest on multiple, interacting levels (Enfield and Levinson 2006). A first distinction is between the *capacity for language* and any particular *languages* or *varieties*, whereby the first is what makes us capable, as a species, of acquiring and using language, whereas the second refers to specific communicative systems used by circumscribed groups for a specific period and in given contexts. It is currently an open question as to the composition of the capacity for language, some arguing for a language-specific core (Hauser et al. 2002) and others for the reuse of more general, non-language-specific and preexisting mechanisms (e.g., Christiansen and Chater 2008; Pinker and Jackendoff 2005). Nevertheless, it is clear that the existence of particular languages as cultural constructs requires this capacity to be in place, and it is also clear that any particular language is shaped by its properties (Christiansen and Chater 2008).

Language has several *design features* (Hockett 1960), among the best known being the *arbitrariness* of the mapping between signal and meaning, *discreteness*, and *duality of patterning* (meaningful units result by combining meaningless lower level units). A language thus has several interdependent levels of structure, ranging from low-level articulatory (or gestural) coordination,

acoustic (or visual) perception, to phonology, morpho-syntax, the lexicon, and the whole discourse embedded in its communicative, social, and cultural contexts. These multiple levels can—and do—vary between languages, and the laws and constraints subtending this variation are currently a hotly debated topic (Evans and Levinson 2009).

Levels of Relationship between Language and Genes

Therefore, "language" is not a unitary phenomenon, a property that it shares with "genes," and this makes any attempt at providing a single narrative encompassing the relationship between them far from simple. More precisely, it is becoming increasingly clear that not only are the pathways connecting genes to phenotypes nonlinear and difficult to map, and that gene-gene and gene-environment interactions are the norm in the development of most phenotypic aspects, but also that there is no clear-cut difference between "genetic" and "environmental," that "development" is not a discrete, encapsulated, and teleological phase in the life cycle of an organism, and that "genes" are essential to all processes at all times (Minelli 2003; Odling-Smee et al. 2003; West-Eberhard 2003). And these observations apply even more forcefully to such a complex and multifaceted phenotype as language, where, for each level and aspect, the question of the genetic basis is legitimate and the answer will probably contain both unique and shared components.

At the most general level, that of the whole species and encompassing phylogenetic periods (tens or hundreds of thousands to millions of years; Joblinget al. 2004), the question of the genetic bases of the human capacity for language seems to require a positive answer, given that it represents a species-specific trait not shared even with our closest primate relatives. However, even at this level, it is becoming unclear how "species-specific" this trait in fact is (or was), given the recent data suggesting that Neandertals shared with us not only a similar capacity for symbolic culture (Zilhão et al. 2006, 2010) and probably a similar larynx (Arensburg and Tillier 1991; Fitch 2000), but also the currently most "language-related" gene, *FOXP2* (Krause et al. 2007). Nevertheless, irrespective of this debate, human (and possibly Neandertal) language and speech requires a set of special—but not necessarily language-specific—genetic factors.

At the most specific level, that of the individual's development (ontogeny) and life, the influence of genes on language and speech is pervasive and very important. This is amply demonstrated by many behavior genetic studies of various aspects of language (e.g., for a review, see Stromswold 2001) that have found average to large heritabilities (Lych and Walsh 1998). Various language and speech disorders have usually high heritabilities ($h^2 > 0.50$; Bonneau et al. 2004; Felsenfeld 2002; Stromswold 2001). For example, the liability to stuttering is highly heritable ($h^2 = 0.70$; Felsenfeld 2002) and the heritability of specific language impairment (Bishop 2003; OMIM 602081) is also high (Bishop 2003; Bonneau et al. 2004). Also, normal interindividual variation in aspects of speech

and language have a variable genetic component, with, for example, heritabilities ranging from a very low $h^2 = 0.02$ for expressive vocabulary at 14 months to $h^2 = 0.38$ at 24 months and $h^2 = 0.72$ for WISC-R vocabulary (Stromswold 2001).

Currently, not much is known about the genes subtending these large heritabilities (Bishop 2009), which seems to be a general problem of accounting for the "missing heritability" in complex phenotypes (Bogardus 2009; Maher 2008; Slatkin 2009), but some promising findings do exist. Probably the best known is the case of *FOXP2*, a transcription factor whose disruption results in a complex phenotype including language and speech abnormalities (Fisher and Scharff 2009), but there are also other interesting candidates, including *CNTNAP2*, a gene from the neurexin superfamily expressed in the developing human cortex, down-regulated by *FOXP2* and putatively associated with Nonsense-Word Repetition task (Vernes et al. 2008), as well as various genes involved in dyslexia (*KIAA0319*, *DCDC2*; Bishop 2009).

However, at the intermediate level, involving populations across time and space ranges relevant to language change (*glossogeny*; Hurford 1990), the relationship between genes and languages is not very clear. I suggest that this controversy results from the conflation of two different processes involving accidental and, respectively, causal correlations between genetic and linguistic processes. The first type of relationship has been widely popularized by L. L. Cavalli-Sforza and coworkers (e.g., Cavalli-Sforza et al. 1994) and is based on the fact that population-level processes usually affect in parallel ways both the population's languages (and, more generally, culture) and its genes, resulting in correlations between the two. For example, fissions, whereby a parent population splits into two or more daughter populations due to migration, will result, in time, given reduced levels of contact, in increasing divergence both from a linguistic point of view (dialects → languages → branches in a language family → unrecognizable relatedness) and also from a genetic point of view, albeit in a more continuous manner (Cavalli-Sforza et al. 1994; Jobling et al. 2004), a special case being represented by the language/farming codispersal hypothesis (Cavalli-Sforza et al. 1994; Diamond 1997, 1998; Diamond and Bellwood 2003; Renfrew 2002). Nevertheless, even if this nice and clean picture is complicated by extensive cultural and genetic contact as well as by methodological assumptions concerning linguistic and population hierarchical classifications (Bateman et al. 1990; Bolnick et al. 2004; MacEachern 2000; McMahon 2004; McMahon and McMahon 2005; Sims-Williams 1998), it probably reflects a fundamental process affecting linguistic diversity. Importantly for this discussion, the resulting correlation between genes and languages is arbitrary and nonfunctional, in the sense that what particular genetic variants (e.g., haplogroups, single-nucleotide polymorphism alleles, repeats numbers) happen to be associated with what particular linguistic groups (languages or groups of related languages) is down to processes random with respect to this correlation. Therefore, there is nothing, say, "linguistically Germanic" in the genes differentiating Germanic populations from Romance populations within the Indo-European family.

Genetic Biasing of Language at the Population Level

The second type of relationship between genes and languages hypothesizes causality flowing from the first to the second and is manifested by genetic processes biasing linguistic processes across glossogenetic timescales and geographical regions. It is the least well studied and accepted of all correlations between genes and language, but support for its effectiveness and characterization of its properties is growing thanks to conceptual, mathematical, and computational models and also to experimental and correlational studies.

The fundamental idea is that individual-level genetic factors affecting the interindividual variability of various aspects of language and speech (briefly reviewed previously) also can affect differences *between groups* given the right conditions. For example, let's assume that the capacity to articulate a trilled[3] sound, like in Scottish English or Spanish, has a genetic component[4] with a single gene of major effect influencing the ability of individual speakers to produce this sound, with "impairing" allele *A* determining the production of a substitution sound *r*, such as the approximant [ɹ] or the flap [ɾ], and the "trilling" allele *a* (for details, see Dediu 2007: ch. 5.1.1). Depending on the type of language spoken by an *A*-carrier, its effect can be (1) *hidden* (in languages that do not use the trilled nor the substitution sound *r*, in languages in which and *r* are allophones, being used interchangeably, and in languages that use only *r*) or (2) *visible* but not affecting communication, possibly having only a social value (in languages that use the trill but not the substitution *r*), or (3) it can *affect communication* (in languages in which the trill and the substitution *r* contrast phonemically, differentiating meaning or grammatical function). If the frequency and/or the social relevance of *A* carriers is small in a population, then they should have a negligible impact on the language, their phenotype being at most an individual marker with social effects determined by the local culture. However, if for some reason the frequency of *A* increases in population and/or they mate assortatively and form stable communicative networks, or their effectiveness as language models rises, then it is possible that they would affect the language, driving it toward type 2 and especially type 1 above. This is a case of *direct biasing* in Boyd and Richerson's (1985) parlance, and its assumptions are obviously very strong.

However, nearly 30 years ago, Peter Ladefoged—a very important phonetician—suggested a case involving differences between populations in the anatomy of the vocal tract and its effects on speech (also see Ladd et al. 2008). He observed that the otherwise very similar vowel systems of Italian and Yoruba (both have seven vowels /i e ɛ a ɔ o u/) show subtle differences

[3]The IPA (The International Phonetic Alphabet, revised 2005, © International Phonetic Association, www.langsci.ucl.ac.uk/ipa/IPA_chart_(C)2005.pdf) notation is used.

[4]This assumption seems supported by data: "The results of these analyses suggest that articulation of the phoneme /r/ is largely the result of genetic factors, whereas environmental factors play a greater role in the articulation of the phonemes /l/, /w/, and /j/" (Stromswold 2001: 673).

that can be traced back to anatomic differences between their speakers (Disner 1983; Ladefoged 1984):

> Some of the differences between the two languages are due to the shapes of the lips of Italian as opposed to Yoruba speakers. [. . .] [W]ith the exception of /i/ and to a lesser extent /e/, the second formant is lower for the Italian vowels than for the Yoruba vowel. These differences are precisely those that one would expect if Yoruba speakers, on the whole, used a larger mouth opening than that used by the Italian. [. . .] The possibility of overall differences in mouth opening is certainly compatible with the apparent facial differences between speakers of Yoruba and Italian. (Ladefoged 1984: 85–86)

Leaving aside any unfounded accusations of racism, if confirmed, this observation represents a clear case of biasing of speech ultimately due to genetic differences between the speaker populations. Critical, however, is the observation that these differences, this bias, is small at the individual level (as opposed to the previous example involving a large individual bias resulting in the incapacity to produce the trill /r/), because

> [t]his does not, of course, imply that a Yoruba could not learn perfect Italian. Any individual speaker could compensate for the overall, statistical, difference in headshape [. . .]. (Ladefoged 1984: 86)

More precisely, the focus is on types of biases that *do not prevent any individual* from acquiring and using the language(s) of its native community, being easily "overridden" and "masked" by learning. However, importantly, even such an apparently negligible bias, I would argue (together with other researchers), can—and does—influence language and speech given the appropriate conditions.

To make the case clearer, let us entertain a thought experiment (following Ladd et al. 2007): a group of Italian children is teleported at birth to a Nigerian village and adopted by the Yoruba-speaking community.[5] There is no reason to suppose that they would not acquire perfect Yoruba, and that they, assumed to continue living as a closed, mostly endogamous community, would teach their Yoruba to their children, which would acquire it, and so on for several generations. However, a phonetician analyzing the second formant of their vowels would probably notice a tendency to lower them, which would be amplified across generations, resulting, in the end, in a "dialect" of Yoruba with vowels tending toward the Italian vowels. Thus, linguistic differences would, in this case, be the causal result of the iteration of small individual biases amplified by transgenerational cultural transmission in populations of appropriate genetic structure.

[5]To fully use the power of SiFi and control for confounding variables, let us further suppose that their adoptive community does not notice, or consider relevant, any phenotypic differences.

Another important argument in favor of the efficacy of such a genetic bias in influencing cultural evolution is provided by recent work on zebra finches (Fehér et al. 2009), a bird species in which young males learn their songs from adult males. When raised in isolation, they spontaneously develop a deviant type of song, different from the species-specific song. However, when introduced in an iterated learning chain, where the isolates act as the model for the first generation, in a short number of generations (three to four) the birds recover the species-specific song. Fehér et al. (2009) have performed various analyses of the intergenerational changes pulling the song from the isolate toward the species-specific type and have identified several biases—apparently innate in nature—affecting, for example, rhythm and syllable structure. This example, even if not directly related to language, strongly suggests that small individual biases can still force cultural evolution in certain directions.

Concerning specifically the possibility that small individual biases are amplified by transmission across generations in the context of rule learning in human language, a very recent experimental study of the cultural evolution of plural marking in an artificial language (Smith and Wonnacott 2010) shows that the barely detectable individual-level tendencies toward regularization are not enough to explain the massive regularization of plural marking after five generations but requires the iterated application of these weak biases on the products of previous weak biases in a cross-generational cumulative process.

Several mathematical and computational models support these inferences. Kirby et al. (2007) used Bayesian agents (Griffiths and Kalish 2007) in an *Iterated Learning Framework* (Kirby and Hurford 2002) to investigate the influence of learning biases on language evolution. In this context, a Bayesian agent uses Bayes' rule (Press 2003) to update its prior distribution across a set of possible languages in the light of observed language data and then chooses a single language from this posterior distribution, which it then uses to produce speech data for the next generation. Different rules for choosing this language are conceivable (Kirby et al. 2007; Smith and Kirby 2008), but the most important rules are what is called the *sampler*; the *sampler* picks randomly a language with probability given by the posterior, and the *maximizer* picks the language with maximum posterior probability (Griffiths and Kalish 2007). The a priori distribution across languages is equated with the bias, which is assumed to be of innate (Kirby et al. 2007) or of a heterogeneous (Griffiths and Kalish 2007) nature. Nevertheless, the most important results for this discussion are that, in this highly simplified context involving a single agent per generation and therefore purely vertical transmission (Dediu 2009), samplers always converge to their prior, whereas maximizers show a complex behavior influenced by the prior (Griffiths and Kalish 2007). More specifically, Kirby et al. (2007) have shown that maximizers not only amplify very small biases toward compositionality but also that, within limits, the actual strength of this bias does not seem to affect the resulting language. Together, these models suggest that biases do affect language

across generations of cultural transmission and that, given certain assumptions, these biases are expressed (amplified or dampened) in complex ways.

The generalizability of such simple models has been questioned both on grounds of the social and communicative structures considered (Dediu 2008, 2009) as well as in the assumptions behind this Bayesian model of language acquisition and usage (Dediu 2009; Ferdinand and Zuidema 2009). To study the effects of more complex (and realistic) social and communicative settings, I have recently conducted a series of studies (Dediu 2008, 2009) where computational agents learn and transmit language either in (1) the simple, single-agent transmission chain discussed above; (2) a more complex setting involving chains of pairs of agents (allowing vertical, horizontal, and oblique transmission); and (3) a two-dimensional world with complex demography, featuring several populations that migrate and interact. I also have used two types of computational agents, Bayesian (sampler and maximizers) and non-Bayesian (implementing two types of biases: *initial expectation* representing an asymmetric initial state and *rate of learning* representing an asymmetric easiness of acquisition). The simulations showed that Bayesian agents in situations 2 and 3 still produce language influenced by their biases but in a much more complex manner (Dediu 2009) and that non-Bayesian agents implementing a rate of learning bias also show bias amplification even in scenario 3 (Dediu 2008).

Therefore, mathematical and computational models suggest that genetic biasing of language, even if small at the individual level, can act as a forcing factor on the trajectory of language change, leading to universals or distributions of language reflecting, usually in complex ways, these biases. It must be noted, however, that such models, especially when more complex social and communicative assumptions are used, strongly suggest that the actual outcome is influenced by many other factors, including "random" historical accidents and language contact. However, probably the most detailed example of what such a genetically biased influence on linguistic diversity might look like is offered by the recently published relationship between linguistic tone and two brain-related human genes.

Linguistic Tone As Biased by *ASPM* and *Microcephalin*

One of the many dimensions on which languages differ is represented by the use of voice pitch to convey lexical or grammatical meaning besides the more common sentence-level meanings, such as questions or exclamations (Dediu and Ladd 2007; Yip 2002). In such *tone* languages, such as Mandarin Chinese or Yoruba, the pitch level and/or the pitch contour mark linguistically relevant distinctions, and the number of such *tones* varies from two to about seven (Yip 2002). The typological (Croft 1990) classification of languages as tonal is usually straightforward, but there are borderline cases (such as Swedish or Norwegian) as well. Nevertheless, tone languages make up slightly more than half the world's languages (Haspelmath et al. 2005), with an uneven geographical distribution, mostly clustered in sub-Saharan Africa, Southeast Asia, Papua-New Guinea, and

Central America and Amazonia (Dediu and Ladd 2007; Haspelmath et al. 2005).[6] Like words and many other *typological features* (e.g., word order, number of vowels and consonants; Croft 1990; Haspelmath et al. 2005), tone (or for that matter non-tone) tends to be inherited from parent to daughter languages and can be borrowed across language borders through contact (Dediu and Ladd 2007; Yip 2002). Also, tone can arise through regular historical linguistic processes (*tonogenesis*; Hyman 1978; Yip 2002), and it also can be lost, for example, in situations of usage as *lingua franca* (like in Swahili).

In 2007, together with D. Robert Ladd (Dediu and Ladd 2007), we have proposed that one factor contributing to the distribution of tone languages is represented by the biasing induced by two human genes involved in brain growth and development, *ASPM* and *Microcephalin*. Both genes are responsible (but are not the only genes) for primary autosomal recessive microcephaly (OMIM 608716), characterized by head size much smaller than the average (Cox et al. 2006; Gilbert et al. 2005; Woods 2004), probably by influencing the number of asymmetric cell divisions of the neuronal precursors (Dediu and Ladd 2007). In 2005, the existence of a "derived" haplogroup, one for each gene, was reported (Evans et al. 2005; Mekel-Bobrov et al. 2005), having a skewed geographical distribution and reportedly under recent or ongoing natural selection.[7] The phenotypic effects of these derived haplogroups are seemingly not major and do not seem to be related to, for example, variation in intelligence or head size in the normal population (Mekel-Bobrov et al. 2007; Woods et al. 2006) or to the incidence of schizophrenia (Rivero et al. 2006); however, there seems to be an association between *Microcephalin* and cranial volume in normal Chinese males (Wang et al. 2008) and an independent finding of a sex-specific association between these two genes and normal variation in brain anatomy (Rimol et al. 2010).

The geographical distribution of tone languages and the population frequencies of the derived haplogroups of *ASPM* and *Microcephalin* (henceforth denoted, for brevity, as *ASPM-D* and *MCPH-D*) seem to be similar, as shown in Figure 1. More precisely, tone languages tend to be spoken by populations with low frequencies of both derived haplogroups and non-tonal languages by populations with high frequencies of both, whereas populations with a low frequency of *ASPM-D* and high frequency of *MCPH-D* have equal chances of speaking any type (Figure 2).

Both the correlations between the population frequency of *ASPM-D* and tone (tonal vs. non-tonal language; $r = 0.53$, $p = 9.63 \times 10^{-5}$) and *MCPH-D* and tone ($r = 0.54$, $p = 7.22 \times 10^{-5}$) are large and highly significant, confirmed by the strong relationship between both *ASPM-D* and *MCPH-D* considered together and tone (the logistic regression of type on the frequencies of both

[6]For a map of tone the interested reader should consult WALS online (March 2011): http://wals.info/feature/13?v1=cffff&v2=cf6f&v3=cd00&s=12&z1=2998&z2=2999&z3=3000&tg_format=map&lat=5.5&lng=152.58&z=2&t=m.

[7]The claim of recent natural selection has since been disputed (Currat et al. 2006; Yu et al. 2007), but its status is not relevant to the current discussion (Dediu and Ladd 2007; Ladd et al. 2008).

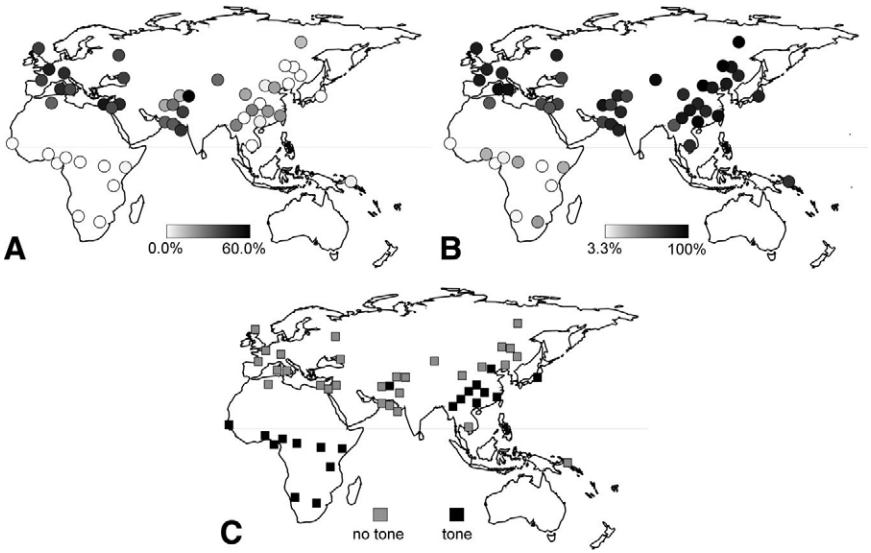

Figure 1. Geographical distribution of the derived haplogroups of *ASPM* (A), *Microcephalin* (B), and tone languages (C). In A and B, shades of gray represent population frequency of the derived haplogroup, from white (the minimum, 0% for *ASPM* and 3.3% for *Microcephalin*) to black (the maximum, 60% for *ASPM* and 100% for *Microcephalin*). In C, black represents tone languages and white non-tonal languages. Please note that the points represented on these maps correspond to the populations for which genetic data are available (for details, see Dediu and Ladd 2007 7), thus accounting for the lack of information in Australia (eminently non-tonal) and Papua-New Guinea (a very interesting mix of tone and non-tonal languages).

derived haplogroups correctly classifies 73% of the languages, Nagelkerke's $R^2 = 0.53$). However, given that it is not clear how the relationship between a typological feature and two human genes looks in general (Dediu and Ladd 2007), we have compared the association between *ASPM-D*, *MCPH-D* and tone to the distribution generated by relating any of 26 typological features to any pair of 983 genetic markers. This comparison provides an appropriate null hypothesis, given that it takes into account the effects of past demographic events or any other spurious (for our purposes) sources of association between language and genes. It turns out that the relationship between *ASPM-D*, *MCPH-D* and tone is in the tail of this empirical distribution (the correlations are in the top 1.5%, whereas the logistic regression is in the top 2.7%), suggesting that there might, indeed, be "special" reasons for this association (Dediu and Ladd 2007; Ladd et al. 2008).

Nevertheless, to explicitly test the possibility that this association is due to accidents of history, contact, or both (see above), we have further controlled for historical relationships between languages, encoded as historical linguistic distances, and for contact, using as proxy the land distances between populations (Dediu and Ladd 2007). The Mantel correlations (Mantel 1967) between *ASPM-D*, *MCPH-D*, and tone remain strong and highly significant after controlling

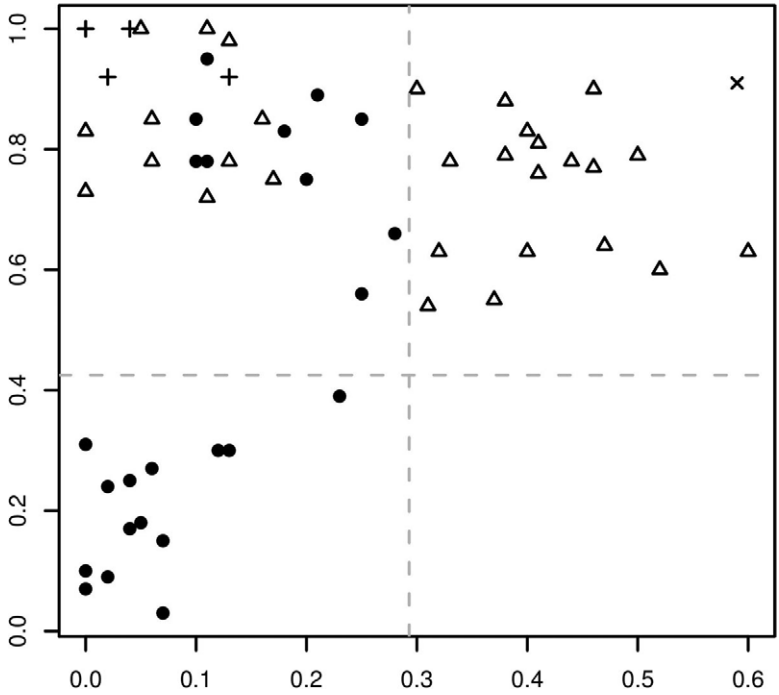

Figure 2. Scatter plot of language type (tone, △; non-tone, ●) by frequency of *ASPM-D* (horizontal axis) and *MCPH-D* (vertical axis). Plus signs (+) represent the American populations that have been excluded from the analyses and used as test cases for the results: they conform to the predicted pattern of low *ASPM-D*, high *MCPH-D*, and a mixture of tone and non-tonal languages (for details, see Dediu and Ladd 2007).

for contact ($r = 0.291$, $p = 0.003$) and, more importantly, after controlling for both contact and shared history ($r = 0.283$, $p = 0.000$). All these, taken together, strongly suggest that the association between these two derived haplogroups and linguistic tone is not only "special" among the 11,582,690 such associations tested (Dediu and Ladd 2007) but that it is not explained by shared history or genetic and linguistic contact, suggesting a direct causal link between them, with the two genes encoding a bias affecting tone. Probably, this bias is an "unintended" side effect of these genes on brain growth and development that most likely does not play a role in the putative recent selection on these genes (for a fuller discussion, see Dediu and Ladd 2007 and especially Ladd et al. 2008). Moreover, this bias is assumed to be very small at the individual level, in agreement with the observation that any normal child can perfectly learn the language(s) of their native community (see also above), ruling out any simplistic "gene for Chinese" theories (Ladd et al. 2008).

The actual mechanisms through which these genes could influence the acquisition, processing, or both of linguistic tone are currently not clear, but several promising hints do exist. It is clear that deleterious mutations of *ASPM*

and *Microcephalin* are causes of primary microcephaly (Cox et al. 2006; Gilbert et al. 2005; Woods 2004), as discussed above briefly, and it also has been recently shown that polymorphisms in *Microcephalin* are associated with normal variation in the cranial volume in a sample of Chinese males (Wang et al. 2008), whereas polymorphisms in both genes show a sex-specific association with brain volume and cortical surface area in a combined Norwegian-North American sample (Rimol et al. 2010). These findings clearly support the idea that normal variants (polymorphisms) of these genes are implicated in normal variation in brain structure, because they probably control the number of symmetric divisions of the neuroepithelial cells during embryogenesis (Bond and Woods 2006; Zhong et al. 2006) resulting in variation in brain size (Caviness et al. 1995; Dediu and Ladd 2007). Given the apparent region specificity of the effects of normal polymorphisms on cortical area[8] (Rimol et al. 2010), it is not improbable to suggest that the derived haplogroups of *ASPM* and *Microcephalin* can affect brain networks involved in the acquisition, processing, or both of linguistic tone. In fact, association studies focusing specifically on these haplogroups and brain areas putatively involved in tone are a natural next step.

Strong support is offered by the recent finding (Christiansen et al., unpublished data) of an association between polymorphisms of *ASPM* and various language measures, some related to phonology. In this same vein, we are currently working on the isolation and operationalization of aspects of the postulated bias affecting tone to conduct more specific large-scale genetic association studies. Not specifically concerning these two genes, but in general supporting putative genetic differences between populations in the processing of tone, it was found that absolute pitch is more common among East Asians than Caucasians (Deutsch et al. 2006; Gregersen et al. 2000) and that this seems to have a genetic basis (Baharloo et al. 1998; Zatorre 2003), but it is currently unclear whether these differences are due to genetic, linguistic, or other cultural factors, or, probably, to a complex interaction between them (Gregersen et al. 2007; Henthorn and Deutsch 2007). A similar advantage for relative pitch in East Asians was found by Hove et al. (2010), who also controlled for the effects of speaking a tone language in addition to ethnicity. Also in general support of this line of arguments is the recent finding that five genes involved in cochlear function, including the protocadherin gene *PCDH15*, show signs of selection in East Asia (Grossman et al. 2010). Of course, all these represent clues and supporting evidence for the postulated role of *ASPM* and *Microcephalin* in shaping a genetic bias affecting linguistic tone, but in the near future I am expecting stronger tests of this hypothesis, some of which I am currently involved in.

This genetic bias is but one of the many factors affecting the trajectory of language change, together with contact phenomena, system-internal constraints, and pure historical accidents; but, on average, it will result in differences between

[8]Which is to be expected given the complexity of brain development and the general context sensitivity of gene expression.

languages to correlate with genetic differences between the populations speaking them (Dediu and Ladd 2007; Ladd et al. 2008). Another consequence of this genetic biasing it that, on average, linguistic tone should be more stable than expected on purely cultural grounds, a prediction that seems supported by recent work (Dediu 2011).

Conclusions: Genetic Biasing and Language Evolution, Change, and Diversity

I hope that the previous arguments, building on conceptual, mathematical, and computational models; some recent experimental evidence; and the association between the population frequency of two brain growth and development-related genes, *ASPM* and *Microcephalin*, and the distribution of tone languages, have succeeded in showing that genetic biases can affect the trajectory of language change. This can result either in relatively stable patterns of linguistic diversity or in the fixation of properties of language at the expense of their alternatives. Therefore, genetic biases potentially are factors explaining the distribution of language features across the world, but, most importantly, they can play a role in explaining features shown by most languages as well.

It is increasingly clear that the notion of a language universal, seen as an absolute property of all human languages, is too strong to explain the complex reality of human languages (Evans and Levinson 2009) and that the most productive approach is probably represented by a statistical stance, whereby different properties have different probabilities of being found. In this context, the idea of genetic biasing can account both for persisting (or transient) patterns of diversity as well as for the degrees of universality shown by language. For example, assuming the genetic biasing of tone by *ASPM* and *Microcephalin*, the fixation of the derived haplogroups across the entire species would presumably increase the frequency of non-tone languages toward 100%, transforming it into a language universal.[9] A more extreme scenario would involve the possibility that the biasing gene is, in turn, under natural selection due to its effects on language, so that it would increase in frequency toward fixation, at the same time altering the universal properties of language in the process.

It is hard to accurately estimate the importance of the hypothesis of genetic biasing on the future study of language, and it is even harder for me to be objective, but it seems that this proposal—if supported by further, more direct evidence—has the potential to raise new research questions and directions. One important question concerns the generality of such a mechanism: What aspects of language are more plausibly under genetic biasing? What other aspects of culture, more or less directly related to language, show such biasing? Can we find a better model than linguistic tone? What is the range of plausible genetic,

[9]Of course, it is entirely possible that the fixation of the two derived haplogroups would not completely eliminate tone languages but would simply reduce their frequencies.

molecular, neuronal, psychological, and sociocultural mechanisms behind this type of biases? How are they developmentally realized? And so on.

Below are some of my tentative answers to these questions: I tend to assume that phonetic features would be the easiest to study from this perspective but it might as well turn out that other, more "cognitive" aspects are under such strong biases relating to, for example, working memory constraints. Thus, I would suggest that a productive line of inquiry will be represented by the operationalization of as many aspects as possible of the production and perception of language (speech and gesture) followed by studies of their heritability, genetic association/linkage studies as well as large-scale, cross-cultural correlational studies of the type introduced in Dediu and Ladd (2007). Concerning other aspects of culture under biasing, a good starting point is represented by the suggestions and models pioneered by Boyd and Richerson (1985) and Cavalli-Sforza and Feldman (1981), but a domain that looks very promising to me is represented by color terminology in the world's languages (Berlin and Kay 1969), which might, in fact, turn out to be easier to study than linguistic tone but still representative of a complex interaction between genetics and cultural transmission. However, to the last question I think currently the answer is very vague by necessity: such genetic biases can become "manifest" in several manners, at several scales and during various stages of development, ranging, for example, from subtle differences in the processing of fast temporal sequences during the early stages of development to stable anatomical differences in the vocal tract across the whole life span. Therefore, I would propose that instead of trying to constrain the class of possible answers to this question we should keep an open mind and study each individual case of genetic biasing on its own and hope that we will, indeed, arrive at valid generalizations at a later stage.

Moreover, this mechanism of genetic biases affecting (and possibly being affected, in turn, by) language can provide an explanation for the rise of certain seemingly universal properties of language in a gradual manner, through the interplay between genetic and cultural processes acting on different but related timescales.

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Literature Cited

Arensburg, B., and A. M. Tillier. 1991. Speech and the Neanderthals. *Endeavour* 15:26–28.

Baharloo, S., P. A. Johnston, S. K. Service, J. Gitschier, and N. B. Freimer. 1998. Absolute pitch: An approach for identification of genetic and nongenetic components. *Am. J. Hum. Genet.* 62:224–231.

Bateman, R., I. Goddard, R. O'Grady et al. 1990. Speaking of forked tongues: The feasibility of reconciling human phylogeny and the history of language. *Curr. Anthropol.* 31:1–13.

Berlin, B., and P. Kay. 1969. *Basic Color Terms: Their Universality and Evolution*. Berkeley, CA: University of California Press.

Bishop, D. V. M. 2003. Genetic and environmental risks for specific language impairment in children. *Int. J. Pediatr. Otorhinolaryngol.* 6751:S143–S157.

Bishop, D. V. M. 2009. Genes, cognition, and communication: Insights from neurodevelopmental disorders. *Ann. N. Y. Acad. Sci.* 1156:1–18.

Bogardus, C. 2009. Missing heritability and GWAS utility. *Obesity* 2:209–210.

Bolnick, D., B. Shook, L. Campbell et al. 2004. Problematic use of Greenberg's linguistic classification of the Americas in studies of Native American genetic variation. *Am. J. Hum. Genet.* 75:519–523.

Bond, J., and C. G. Woods. 2006. Cytoskeletal genes regulating brain size. *Curr. Opin. Cell Biol.* 18:95–101.

Bonneau, D., C. Verny, and J. Uzé. 2004. Les facteurs génétiques dans les troubles spécifiques du langage oral. *Arch. Pediatr.* 10:1213–1216.

Boyd, R., and P. -J. Richerson. 1985. *Culture and the Evolutionary Process*. Chicago, IL: University of Chicago Press.

Cavalli-Sforza, L. L., P. Menozzi, and A. Piazza. 1994. *The History and Geography of Human Genes*. Princeton, NJ: Princeton University Press.

Cavalli-Sforza, L. L., and M. W. Feldman. 1981. *Cultural Transmission and Evolution: A Quantitative Approach*. Princeton, NJ: Princeton University Press.

Caviness, V. S., Jr., T. Takahashi, and R. S. Nowakowski. 1995. Numbers, time and neocortical neuronogenesis: A general developmental and evolutionary model. *Trends Neurosci.* 18:379–383.

Christiansen, M. H., and N. Chater. 2008. Language as shaped by the brain. *Behav. Brain Sci.* 31:489–509.

Cox, J., A. P. Jackson, J. Bond et al. 2006. What primary microcephaly can tell us about brain growth. *Trends Mol. Med.* 12:358–366.

Croft, W. 1990. *Typology and Universals*. Cambridge, U.K.: Cambridge University Press.

Currat, M., L. Excoffier, W. Maddison et al. 2006. Comment on "Ongoing adaptive evolution of *ASPM*, a brain size determinant in *Homo sapiens*" and *Microcephalin*, a gene regulating brain size, continues to evolve adaptively in humans. *Science* 313:172.

Dediu, D., and L. R. Ladd. 2007. Linguistic tone is related to the population frequency of the adaptive haplogroups of two brain size genes, *ASPM* and *Microcephalin*. *Proc. Natl. Acad. Sci. USA* 104:10944–10949.

Dediu, D. 2007. *Non-Spurious Correlations between Genetic and Linguistic Diversities in the Context of Human Evolution*. Ph.D. dissertation, University of Edinburgh, Edinburgh, U.K.

Dediu, D. 2008. The role of genetic biases in shaping language-genes correlations. *J. Theor. Biol.* 254:400–407.

Dediu, D. 2009. Genetic biasing through cultural transmission: Do simple Bayesian models of language evolution generalize? *J. Theor. Biol.* 259:552–561.

Dediu, D. 2011. A Bayesian phylogenetic approach to estimating the stability of linguistic features and the genetic biasing of tone. *Proc. R. Soc. B.* 278:474–479.

Dediu, D. (in press). Genes - Interactions with language on three levels: Inter-individual variation, historical correlations and genetic biasing. In *The Language Phenomenon*, K. Smith and P. Binder, eds. The Frontiers Collection, Springer.

Deutsch, D., T. Henthorn, E. Marvin et al. 2006. Absolute pitch among American and Chinese conservatory students: Prevalence differences, and evidence for a speech-related critical period. *J. Acoust. Soc. Am.* 119:719–722.

Diamond, J. 1997. The language steamrollers. *Nature* 389:544–546.

Diamond, J. 1998. *Guns, Germs and Steel: A Short History of Everybody for the Last 13,000 Years*. London, U.K.: Vintage.

Diamond, J., and P. Bellwood. 2003. Farmers and their languages: The first expansions. *Science* 300:597–603.

Disner, S. F. 1983. Vowel quality: The contribution of language particular and language universal factors. *UCLA Working Pap. Phon.* 58:1–158.

Enfield, N. J., and S. C. Levinson, S. C. 2006. *Roots of Human Sociality: Culture*, *Cognition and Interaction*. Oxford, U.K.: Berg Publishers.

Evans, N., and S. C. Levinson. 2009. The myth of language universals: Language diversity and its importance for cognitive science. *Behav. Brain Sci.* 32:429–448.

Evans, P., S. Gilbert, N. Mekel-Bobrov et al. 2005. *Microcephalin*, a gene regulating brain size, continues to evolve adaptively in humans. *Science* 309:1717–1720.

Fehér, O., H. Wang, S. Saar et al. 2009. De novo establishment of wild-type song culture in the zebra finch. *Nature* 459:564–568.

Felsenfeld, S. 2002. Finding susceptibility genes for developmental disorders of speech: The long and winding road. *J. Commun. Disord.* 35:329–345.

Ferdinand, V., and W. Zuidema. 2009. Thomas' theorem meets Bayes' rule: A model of the iterated learning of language. In *Proceedings of the 31st Annual Conference of the Cognitive Science Society*, N. A. Taatgen and H. van Rijn eds. Austin, TX: Cognitive Science Society, 1786–1791.

Fisher, S. E., and S. Scharff. 2009. FOXP2 as a molecular window into speech and language. *Trends Genet.* 25:166–177.

Fitch, W. T. 2000. The evolution of speech: A comparative review. *Trends Cogn. Sci.* 4:258–267.

Gilbert, S. L., W. B. Dobyns, and B. T. Lahn. 2005. Genetic links between brain development and brain evolution. *Nat. Rev. Genet.* 6:581–590.

Gregersen, P. K., E. Kowalsky, and W. Li. 2007. Reply to Henthorn and Deutsch: Ethnicity versus early environment: Comment on "Early Childhood Music Education and Predisposition to Absolute Pitch: Teasing Apart Genes and Environment" by Peter K. Gregersen, Elena Kowalsky, Nina Kohn, and Elizabeth West Marvin [2000]. *Am. J. Med. Genet.* 143:104–105.

Gregersen, P. K., E. Kowalsky, N. Kohn et al. 2000. Early childhood music education and predisposition to absolute pitch: Teasing apart genes and environment. *Am. J. Hum. Genet.* 98:280–282.

Griffiths, T., and M. Kalish. 2007. Language evolution by iterated learning with Bayesian agents. *Cogn. Sci.* 31:441–480.

Grossman, S. R., I. Shylakhter, E. K. Karlsson et al. 2010. A composite of multiple signals distinguishes causal variants in regions of positive selection. *Science* 327:883–886.

Haspelmath, M., M. S. Dryer, D. Gil et al. 2005. *The World Atlas of Language Structures*. Oxford, U.K.: Oxford University Press.

Hauser, M. D., N. Chomsky, and W. T. Fitch. 2002. The FACULTY OF LANGUAGE: What is it, who has it, and how did it evolve? *Science* 298:1569–1579.

Henthorn, T., and D. Deutsch. 2007. Ethnicity versus early environment. *Am. J. Med. Genet.* 143:102–103.

Hockett, C. F., 1960. The origin of speech. *Sci. Am.* 203:88–96.

Hove, M. J., M. E. Sutherland, and C. L. Krumhansl. 2010. Ethnicity effects in relative pitch. *Psychon. Bull. Rev.* 17:310–316.

Hurford, J. 1990. Nativist and functional explanations in language acquisition. In *Logical Issues in Language Acquistion*, I. M. Roca, ed. Banbury, Oxfordshire, U.K.: Foris Publications, 85–136.

Jobling, M. A., M. E. Hurles, and C. Tyler-Smith. 2004. *Human Evolutionary Genetics: Origins*, *Peoples and Disease*. London and New York: Garland Science Publishing.

Kirby, S., and J. Hurford. 2002. The emergence of linguistic structure: An overview of the Iterated Learning Model. In *Simulating the Evolution of Language*, A. Cangelosi and D. Parisi, eds. London, U.K.: Springer, 121–148.

Kirby, S., M. Dowman, and T. L. Griffiths. 2007. Innateness and culture in the evolution of language. *Proc. Natl. Acad. Sci. USA* 104:5241–5245.

Krause, J., C. Lalueza-Fox, L. Orlando et al. 2007. The derived *FOXP2* variant of modern humans was shared with Neandertals. *Curr. Biol.* 17:1908–1912.

Ladd, D. R., D. Dediu, and A. R. Kinsella. 2008. Languages and genes: Reflections on biolinguistics and the nature-nurture question. *Biolinguistics* 2:114–126.

Ladefoged, P. 1984. "Out of chaos comes order": Physical, biological, and structural patterns in phonetics. In *Proceedings of the Tenth International Congress of Phonetic Sciences*, Dordrecht, Netherlands: Foris, 83–95.

Lynch, M., and B. Walsh. 1998. *Genetics and Analysis of Quantitative Traits*. Sunderland, MA; Sinauer Associates.

MacEachern, S. 2000. Genes, tribes, and African history. *Curr. Anthropol.* 41:357–384.

Maher, B. 2008. Personal genomes: The case of the missing heritability. *Nature* 456:18–21.

Mantel, N. 1967. The detection of disease clustering and a generalized regression approach. *Cancer Res.* 27:209–220.

McMahon, A., and R. McMahon. 2005. *Language Classification by Numbers*. Oxford, U.K.: Oxford University Press.

McMahon, R. 2004. Genes and languages. *Commun. Genet.* 7:2–13.

Mekel-Bobrov, N., S. Gilbert, P. Evans et al. 2005. Ongoing adaptive evolution of *ASPM*, a brain size determinant in *Homo sapiens*. *Science* 309:1720–1722.

Mekel-Bobrov, N., D. Posthuma, S. Gilbert et al. 2007. The ongoing adaptive evolution of *ASPM* and *Microcephalin* is not explained by increased intelligence. *Hum. Mol. Genet.* 16:600–608.

Minelli, A. 2003. *The Development of Animal Form: Ontogeny, Morphology, and Evolution*. Cambridge, U.K.: Cambridge University Press.

Odling-Smee, J. F., K. N. Laland, and M. W. Feldman. 2003. *Niche Construction: The Neglected Process in Evolution*. Princeton, NJ: Princeton University Press.

Pinker, S. and R. Jackendoff. 2005. The faculty of language: What's special about it? *Cognition* 96:201–236.

Press, S. J. 2003. *Subjective and Objective Bayesian Statics*. Wiley Series in Probability and Statistics, 2nd ed. New York, NY: Wiley.

Renfrew, C. 2002. "The emerging synthesis": The archaeogenetics of farming/language dispersals and other spread zones. In *Examining the Farming/Language Dispersal Hypothesis*, P. Bellwood and C. Renfrew, eds. Cambridge, U.K.: The McDonald Institute for Archaeological Research, 3–16.

Rimol, L. M., I. Agartz, S. Djurovic et al. 2010. Sex-dependent association of common variants of microcephaly genes with brain structure. *Proc. Natl. Acad. Sci. USA* 107:384–388.

Rivero, O., J. Sanjun, M.-D. Molt et al. 2006. The microcephaly *ASPM* gene and schizophrenia: A preliminary study. *Schizophr. Res.* 84:427–429.

Sims-Williams, P. 1998. Genetics, linguistics, and prehistory: Thinking big and thinking straight. *Antiquity* 72:505–527.

Slatkin, M. 2009. Epigenetic Inheritance and the missing heritability problem. *Genetics* 182:845–850.

Smith, K., and S. Kirby. 2008. Cultural evolution: Implications for understanding the human language faculty and its evolution. *Philos. Trans. R. Soc. Lond. B Biol. Sci.* 363:3591–3603.

Smith, K., and E. Wonnacott. 2010. Eliminating unpredictable variation through iterated learning. *Cognition* 116:444–449.

Stromswold, K. 2001. The heritability of language: A review and metaanalysis of twin, adoption, and linkage studies. *Language* 77:647–723.

Vernes, S. C., D. F. Newbury, B. S. Abrahams et al. 2008. A functional genetic link between distinct developmental language disorders. *N. Engl. J. Med.* 359:2337–2345.

Wang, J.-K., Y. Li, and B. Su. 2008. A common SNP of *MCPH1* is associated with cranial volume variation in Chinese population. *Hum. Mol. Genet.* 17:1329–1335.

West-Eberhard, M. J. 2003. *Developmental plasticity and evolution*. New York, NY: Oxford University Press.

Woods, C. G. 2004. Human microcephaly. *Curr. Opin. Neurobiol.* 14:112–117.

Woods, R., N. Freimer, J. D. Young et al. 2006. Normal variants of *Microcephalin* and *ASPM* do not account for brain size variability. *Hum. Mol. Genet.* 15:2025–2029.

Yip, M. 2002. *Tone*. Cambridge, U.K.: Cambridge University Press.

Yu, F., R. S. Hill, S. F. Schaffner et al. 2007. Comment on "Ongoing adaptive evolution of *ASPM*, a brain size determinant in *Homo sapiens*." *Science* 316:370.

Zatorre, R. J. 2003. Absolute pitch: A paradigm for understanding the influence of genes and development on cognitive function. *Nat. Neurosci.* 6:692–695.

Zhong, X., G. P. Pfeifer, and X. Xu. 2006. Microcephalin encodes a centrosomal protein. *Cell Cycle* 5:457–458.

Zilhão, J., D. E. Badal-García, F. d'Errico et al. 2010. Symbolic use of marine shells and mineral pigments by Iberian Neandertals. *Proc. Natl. Acad. Sci. USA* 107:1023–1028.

Zilhão, J., F. d'Errico, J.-G. Bordes et al. 2006. Analysis of Aurignacian interstratification at the Châtelperronian-type site and implications for the behavioral modernity of Neandertals. *Proc. Natl. Acad. Sci. USA* 103:12643–12648.