Using Genetics to Investigate the Evolution of Language and Speech: New Findings on Musicality and Vocal Acoustics

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Genetic data have for over 20 years been recognized as one of the most promising avenues for empirical language evolution research (Christiansen & Kirby, 2003; Fisher, 2017; Fitch, 2017). Genes have been described as the closest thing to "fossils of language", with archaic human DNA akin to a "time machine" (Fitch, 2017). The assumption is that genome-wide association studies (GWAS) and other genetic research can pinpoint genes involved in speech and language, providing a springboard for subsequent analyses of primate and archaic human genomes that can shed light on the timeline for language evolution in our ancestors (Christiansen & Kirby, 2003; Fisher, 2017; Fitch, 2017). GWAS on speech and language has been hampered by a lack of large cohorts with genetic information and relevant phenotypes, although important advances have recently been made (Doust et al., 2022; Eising et al., 2022). Here we discuss new results on the genetics of speech acoustics and musicality traits. Our aim is to illustrate the different ways in which genetic research can test and inform theorizing on the evolution of language and speech broadly construed.

We begin with a new attempt at the "time machine" strategy. To better understand genetic factors influencing speech acoustics, we performed a GWAS on voice pitch (f0) and vowel formants in a population with limited dialectal differences (N = 12,901) (Gisladottir et al., 2023). We discovered sequence variants in ABCC9 that influence voice pitch and other traits, including pulse pressure and the expression of ABCC9 in the adrenal gland (of potential relevance for proposals linking vocal behavior with self-domestication and adrenal gland function; Benítez-Burraco et al., 2018; Ghazanfar et al., 2020; Wilkins et al., 2014). Since the vocal channel plays a relatively more important role in humans than in other great apes (Corballis, 2002; Levinson & Holler, 2014), we compared the human ABCC9 to other primates, identifying a missense change in ABCC9 that is fixed in humans but not present in primate reference genomes. When did this missense change emerge? By examining four genomes from archaic humans, we conclude that the missense change occurred after hominins split from the great apes but before they diverged into modern humans, Neanderthals, and Denisovans. The implications of this finding are far from clear. Voice pitch is a simple acoustic

measure without direct relevance for vocal learning or cooperative behavior. Nevertheless, this study is a reminder that the more we know about the genetic components involved in human communication at all levels, the better we will be able to sketch how speech and language evolved in our ancestors.

There are several limitations of the strategy above. A single gene account for a trait is implausible, given the messy mappings between genetics and complex traits (Fisher & Vernes, 2015). However, there are ways to leverage the general genetic architecture behind a trait, which we illustrate with a study on human musicality. Since Darwin, several authors have proposed that the origins of language can be traced to a musical or prosodic proto-language, with the evolution of vocal imitation for singing as a key stepping stone (Darwin, 1871; Fitch, 2010). Fitch has pointed out some testable predictions that emerge from this account, noting that "because the neural mechanisms underlying song were precursors of phonological mechanisms in spoken language, we expect considerable overlap between phonological and musical abilities (within individuals) and mechanisms (across individuals)," (Fitch, 2010, p. 506). To test this prediction, we performed a GWAS of musicality traits, using tests of musical pitch and beat perception (Peretz & Vuvan, 2017) and self-reported music perception and training (Müllensiefen et al., 2014) (N = 20,440, age 18-95 years). We found that musicality traits correlate with speech and language traits at the phenotypic level. To test overlap of the genetic mechanisms, we then estimated the genetic correlation of the musicality traits with 26 other cognitive traits. Besides genetic correlations with intelligence and personality for some measures, we found that all musicality traits show substantial genetic correlation with verbal working memory, also known as the phonological loop (rg = 0.43 to 0.30, $P < 1.3 \times 10^{-5}$). Verbal working memory is necessary to learn complex utterances and thus relevant for vocal learning (Aboitiz, 2018). While the causal scenarios underlying genetic correlations are difficult to entangle, these findings are in line with the view that musicality and spoken language share genetic roots.

Finally, we will turn back to the GWAS on speech acoustics. We estimated the heritability of voice pitch and vowel formants, providing an estimate of phenotypic variance explained by common sequence variants (SNP-based heritability). We discovered that even vowel formants have a small-to-modest SNP-based heritability, particularly F_2 (14%). This finding has bearing on the proposal that genetic biases influencing the vocal tract can be amplified through language transmission, ultimately contributing to linguistic diversity (Dediu et al., 2017, 2019).

Each of these strategies discussed above brings numerous complexities. However, the promise of GWAS for language evolution remains tantalizing, and it is now more attainable than ever due to fast developments in population genomics.

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