July 08, 2019

Evolution & Behavior

Overturning the hypothesis for how humans evolved language

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This Break was edited by Max Caine, *Editor-in-chief* - TheScienceBreaker

ABSTRACT

Few traits are as uniquely human as complex spoken language. Language, therefore, has interested evolutionary biologists and neuroscientists seeking to understand what makes us, and in particular our brains, distinct from other animals.

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Few traits are as uniquely human as complex spoken language. Language, therefore, has interested evolutionary biologists and neuroscientists seeking to understand what makes us, and in particular our brains, distinct from other animals. The first major genetic break in deciphering the underlying biological architecture of speech came in the late 1990s, when researchers identified a region of chromosome 7 housing a gene named '*FOXP2*' as causing an inherited speech disorder in a large English family. This was the first example of a concrete genetic basis for a human-specific behavioral trait, sparking a wave of wet and dry lab experiments to understand what it could mean for the evolution of *Homo sapiens* and our development of spoken language.

In 2002, it was argued that *FOXP2* experienced a recent selective sweep in the human lineage. A selective sweep occurs when a mutation conferring an advantage appears, rising dramatically in frequency over time in the population due to it being favored by natural selection. This led the authors to propose that *FOXP2* played a key role in the development of modern human language. While met with much enthusiasm at its publication, the subsequent sequencing of DNA from other ancient hominin species (Neanderthals and Denisovans) clouded this hypothesis. Specifically, the mutations

proposed as the target of the sweep – selected as such since they were thought to be human-specific – were also seen in these other archaic hominins. This results in irreconcilable timelines between the ancient DNA evidence and the initial evolutionary hypothesis, which had dated the sweep to be recent and unique to modern humans. Namely, the presence of these mutations in other archaic hominins places their origin far deeper in time before our species split from Neanderthals.

Despite extensive discussion over the past 15 years, the initial hypothesis of a recent selective sweep had not been systematically re-evaluated. This is especially concerning given that the model was based on a limited sequencing dataset of only three noncoding areas of *FOXP2* in a small sample of 20 predominantly Eurasian humans.

In our recent paper, we comprehensively reanalyzed *FOXP2* in hundreds of whole genomes from globally distributed populations to test a hypothesis of recent selection. No selection statistics we ran supported a recent selective event in *FOXP2*. The original signal appears to have instead been the result of an unbalanced sample composition; specifically, the relative lack of genomes of diverse ancestry. This highlights the key importance of being inclusive in scientific research. In particular, studies examining our evolution as a species would do well to consider representative modern populations, not just people whose ancestors originate from Europe.

In conducting our scans, we also noticed an exceptional non-coding region of *FOXP2*. It contained a cluster of sites that are variable in humans but that are invariant when looking across vertebrate species. This suggests that this area may have undergone a shift in its functional role that is unique to the human line, matching the textbook story for *FOXP2*. However, the pattern of variation is most compatible with a loss of function in humans rather than the region taking on a new role or undergoing a selective sweep.

To elaborate, since many species have nearly identical DNA sequence here, this area is interpreted as playing a significant functional role that maintains its DNA as is, with natural selection kicking out any mutations that arise and disrupt the function. In humans, however, we see a group of variable basepairs, which suggests that the selection pressure has been relaxed. This implies that whatever role this area used to have in the common ancestors of humans and other primates, it is no longer as important in us. The verdict is still out regarding the exact function of the intronic region, but we find that it has many properties of an enhancer element, suggesting that it could play a role in modulating gene expression. We also find it expressed in low amounts in human brain tissue, so it could historically have been acting in this tissue of particular interest.

To be clear, we do not contradict the extensive work suggesting that *FOXP2* has an important function in the neurological underpinnings of speech. However, we do find that there is no evidence for natural selection targeting *FOXP2* in humans in the timeline relevant to mankind's attainment of spoken language. This represents a substantial update to the understanding of modern human evolution and a major revision to the history of *FOXP2*, one of the most famous genes upon which much research continues to be based.