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THE SIGNATURE OF POSITIVE SELECTION ON HUMAN-SPECIFIC INSERTIONS AND DELETIONS

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Human-specific small insertions and deletions (HS indels, with lengths <100 Bp) are reported to be ubiquitous in the human genome. However, whether these indels contribute to human-specific traits remains unclear. Here we employ a modified McDonald-Kreitman (MK) test and a combinatorial population genetics approach to infer, respectively, the occurrence of positive selection and recent selective sweep events associated with HS indels. We first extract 625,890 HS indels from the human-chimpanzee-macaque-mouse multiple alignments, and classify them into non-polymorphic (41%) and polymorphic (59%) indels with reference to the human indel polymorphism data. The modified MK test is then applied to 100 Kb partially overlapped sliding windows across the human genome to scan for the signs of positive selection. After excluding the possibility of biased gene conversion and controlling for false discovery rate, we show that HS indels are potentially positively selected in about 10 MB of the human genome. Furthermore, the indel-associated positively selected regions overlap with genes more often than expected. However, our result suggests that the potential targets of positive selection are located in noncoding regions. Meanwhile, we also demonstrate that the genomic regions surrounding HS indels are more frequently involved in recent selective sweep than the other regions. In addition, HS indels are associated with distinct recent selective sweep events in different human subpopulations. Our results suggest that HS indels may have been associated with human adaptive changes at both the species level and the subpopulation level.

Keywords: human evolution, human-specific indels, positive selection, recent selective sweep