ANALYSES OF AN ARCHAIC ALTITUDE ADAPTATION EPAS1 HAPLOTYPE AMONG EAST ASIAN POPULATIONS

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Tibetans, who have lived on the Himalayan Plateau southwest of China for hundreds of generations, have adapted to the extreme environment of high altitude. A highly differentiated 5-SNP haplotype motif (AGGAA) on a hypoxic pathway gene, EPAS1, is observed in Tibetans and lowlanders. To evaluate the potential usage of the 5-SNP archaic altitude adaption haplotype in ancestry inference for Tibetan or Tibetan-related populations, we analyzed the 5-SNP haplotype in 1053 individuals of 12 Chinese populations residing on the Tibetan Plateau, peripheral regions of Tibet, and plains regions. These data were integrated with the genotypes from the 1000 Genomes populations and populations in a previous reported paper for population structure analyses. We found that populations representing highland and lowland groups have different dominant ancestry components. The core Denisovan haplotype (AGGAA) was observed at frequency of 72.32% in the Tibetan Plateau, with a frequency range from 9.48-21.05% in the peripheral regions of Tibet, and a frequency < 2.5% in the plains area. From the individual perspective, 87.57% of the individuals from the Tibetan Plateau carried the archaic haplotype, while < 5% of the Chinese Han people carried this archaic haplotype. Our findings indicate that the 5-SNP haplotype has a special distribution pattern in populations of Tibet and peripheral regions and could be integrated into ancestry informative SNP panels to enhance ancestry resolution.