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Epigenetic signatures of high altitude adaptation in Tibetan population

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Abstract

Genetic adaptations in high-altitude populations which provide them survival benefit/advantage in high altitude have been well documented. However, till date, very limited studies on the epigenetic adaptation in high altitude human population. Therefore, we aimed to study the high altitude adaptation in Tibetan population, with respect to epigenetic adaptation. DNA methylation is one of the major epigenetic marks, role of which has been suspected in a spectrum of gene-environment interaction and biological processes. The most common form of DNA methylation in vertebrates is 5-methylcytosine, mostly observed in CpG rich promoter region. Recent advancements in the field of DNA sequencing made it possible to analyse genome-wide methylation rapidly with high resolution, however, study of methylation at population level to explore population specific gene-environment interaction is not in long race. Therefore, the present study has been designed to analyse DNA methylation signatures (using whole genome bisulfite sequencing) in Tibetan population (presently inhabiting Karnataka since last 50-60 years) but were native of Ladakh (above 5000 meters) since generations along with Indian populations who are living at low altitude (~10 meters). DNA was isolated from blood, collected from the subjects after informed consent. DNA was converted using bisulfite reagents and whole genome bisulfite sequencing (WGBS) was performed using Illumina-2500 platform (Medgenome Pvt. Ltd.). Analysis of WGBS data was performed using various statistical/bioinformatics tools such as bedtools, Bioconductor and R package to find out methylation sites that are significantly different. We observed 6 differentially methylated regions in Tibetans, highland population, of which, 5 were hypo methylated and one was hypermethylated. The present study reveals differential hypo methylation of *CYP2E1* and *CRELD1* genes, previously reported to be involved in high altitude adaptation (Simonson *et al*., 2010; Dong *et al*., 2014), which would of greater interest. Besides this, we observed novel epigenetic differences in chromosome 7, 11 and 15. Our study, for the first time reveals genome wide level of methylation difference in Tibetan population (native of high altitude since generations) residing in low altitude with other mainland Indians of low altitude which could be important epigenetic markers of natural selection. Comparison with native high altitude Tibetan would make the scenario clearer, which is in the process.

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