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1. Science. 2010 Jul 2;329(5987):75-8.

Sequencing of 50 human exomes reveals adaptation to high altitude.

Yi X, Liang Y, Huerta-Sanchez E, Jin X, Cuo ZX, Pool JE, Xu X, Jiang H, Vinckenbosch N, Korneliussen TS, Zheng H, Liu T, He W, Li K, Luo R, Nie X, Wu H, Zhao M, Cao H, Zou J, Shan Y, Li S, Yang Q, Asan, Ni P, Tian G, Xu J, Liu X, Jiang T, Wu R, Zhou G, Tang M, Qin J, Wang T, Feng S, Li G, Huasang, Luosang J, Wang W, Chen F, Wang Y, Zheng X, Li Z, Bianba Z, Yang G, Wang X, Tang S, Gao G, Chen Y, Luo Z, Gusang L, Cao Z, Zhang Q, Ouyang W, Ren X, Liang H, Zheng H, Huang Y, Li J, Bolund L, Kristiansen K, Li Y, Zhang Y, Zhang X, Li R, Li S, Yang H, Nielsen R, Wang J, Wang J.

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Comment in:

Science. 2010 Jul 2;329(5987):40-1.

Residents of the Tibetan Plateau show heritable adaptations to extreme altitude. We sequenced 50 exomes of ethnic Tibetans, encompassing coding sequences of 92% of human genes, with an average coverage of 18x per individual. Genes showing population-specific allele frequency changes, which represent strong candidates for altitude adaptation, were identified. The strongest signal of natural selection came from endothelial Per-Arnt-Sim (PAS) domain protein 1 (EPAS1), a transcription factor involved in response to hypoxia. One single-nucleotide polymorphism (SNP) at EPAS1 shows a 78% frequency difference between Tibetan and Han samples, representing the fastest allele frequency change observed at any human gene to date. This SNP's association with erythrocyte abundance supports the role of EPAS1 in adaptation to hypoxia. Thus, a population genomic survey has revealed a functionally important locus in genetic adaptation to high altitude.

PMID: 20595611 [PubMed - indexed for MEDLINE]