Dokumenttyp: journal article

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Titel des Beitrags: A common haplotype and the Pro582Ser polymorphism of the hypoxia-inducible factor-1 alpha (HIF1A) gene in elite endurance athletes.

Abstract: Hypoxia-inducible factor-1 alpha (HIF1A) is a transcription factor regulating several genes in response to hypoxic stimuli. HIF1A target genes code for proteins involved in oxygen transport, glycolytic enzymes, and glucose transporters. We investigated whether single-nucleotide polymorphisms and haplotypes in the HIF1A gene are associated with endurance performance in the Genathlete cohort, which includes 316 Caucasian male elite endurance athletes (EEA) with a maximal oxygen uptake of 79.0+/−3.5 ml.kg(-1).min(-1) (mean+/−SD) and 304 Caucasian male sedentary controls with a maximal oxygen uptake of 40.1+/−7.0 ml.kg(-1).min(-1). Six single-nucleotide polymorphisms (rs1951795, rs11158358, rs2301113, rs11549465, rs115494657, rs17099207) were genotyped with the TaqMan system. We found a nominal significant tendency for a difference between the two groups for HIF1A Pro582Ser (rs11549465) genotype distributions (Pchi2=0.017). Homozygotes of the Pro genotype were slightly more frequent in athletes than in controls (84 vs. 75%). Compared with Ser carriers, the odds ratio (OR) of being an EEA in Pro/Pro homozygotes was 1.77 [95% confidence interval (CI): 1.18-2.67, P=0.006] compared with the other genotypes. A common HIF1A haplotype (frequency: 15%),
including the rs11549465 Pro allele and the minor A allele of rs17099207 in the 3' flanking region of the gene, showed a significant association with EEA status (OR: 2.37, 95% CI: 1.21-4.66, P=0.012), whereas the most prevalent haplotype (frequency: 59%) comprising the rs11549465 Pro allele and the major G allele of rs1709920 showed no association with EEA status (OR: 0.93, 95% CI: 0.58-1.50, P=0.769). We found preliminary evidence that the HIF1A Pro582Ser polymorphism and a common haplotype of the HIF1A gene may be associated with EEA status in Caucasian men.