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P08-02. Genetic variation within the gene encoding the HIV-1 CCR5 coreceptor in two South African population groups

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Background

Polymorphisms within the open reading frame (ORF) as well as the promoter and regulatory regions can influence the amount of CCR5 expressed on the cell surface and hence an individual's susceptibility to HIV-1. In this study we characterize *CCR5* genes within the South African African and Caucasian populations.

Methods

A 9.2 kb continuous region encompassing the CCR5 ORF, its two promoters and the 3' untranslated region was amplified in 5 overlapping sections. Amplified fragments were sequenced and sequences were subsequently analyzed for the presence of single nucleotide polymorphisms (SNPs), indels and intragenic haplotypes.

Results

Full length CCR5 sequences were obtained for 42 individuals (21 South African Africans and 21 South African Caucasians). Within the 9.2 kb region sequenced, 62 SNP positions, 4 indels, as well as the Δ32 deletion mutant, were detected. Three insertion indels appear to be in linkage disequilibrium. A novel SNP (3014A/G) within the ORF leading to a non-synonomous amino acid (Trp → Cys) change was detected in one Caucasian individual. Five complex putative haplotypes spanning the length of the sequenced region have been identified. These haplotypes appear to be extensions of haplotypes previously described within CCR5 (HHA, HHD, HHE and HHG*2). Two haplotypes, found in high frequency in each of the two population groups studied, share 3 indels and 8 SNP positions suggesting an evolutionary link between the two haplotypes.

Conclusion

Sequencing of CCR5 from 42 South African individuals has demonstrated a high degree of genetic variation and has led to the identification of novel SNPs and putative haplotypes extending over the entire gene. The Caucasian and African population groups show differences in both haplotype arrangement as well as SNP profile. Future work will determine whether identified genetic variations in both the promoter and the remaining gene have an impact on CCR5 expression.