

Sequence analysis of the human kallikrein gene locus identifies a unique polymorphic minisatellite element

Abstract:

Minisatellites are repetitive sequences of DNA that are present throughout the genome. Although the origin and function of these minisatellites is still unknown, they found clinical applications as markers of many diseases, including cancer. Also, they are useful tools for DNA fingerprinting and linkage analysis. Kallikreins are serine proteases that appear to be involved in many diseases including brain disorders and malignancy. We have recently characterized the human kallikrein gene locus on chromosome 19q13.4, which includes 15 kallikrein genes. In this study, we examined the kallikrein locus (approximately 300 Kb) for all known repeat elements. About 50% of this genomic area is occupied by different repeat elements. We also identified unique minisatellite elements that are restricted to chromosome 19q13. Ten clusters of these minisatellites are distributed along the locus on either DNA strand. The clusters are located in the promoters and enhancers of genes, in introns, and in untranslated regions of the mRNA. Analysis of these elements indicates that they are polymorphic, thus they can be useful in linkage analysis and DNA fingerprinting. Our preliminary results indicate also that the distribution of the different alleles of these minisatellites might be associated with malignancy