In vertebrates, tandem arrays of TTAGGG hexamers are present both at telomeres and at intrachromosomal sites (Interstitial Telomeric Sequence, ITS). An extensive comparative analysis of two primate (human and chimpanzee) and two rodent (mouse and rat) genomes allowed us to describe organization and insertion mechanisms of all the informative ITSs present in the four species. Our results strongly suggest that telomerase was utilized, in some instances, for the repair of DNA double-strand breaks occurring in the genomes of rodents and primates during evolution. Results obtained in collaboration with the ISREC (Switzerland) have demonstrated that mammalian telomeres are actively transcribed into telomeric repeat containing RNA (TERRA). Using a bio-informatic approach we have identified a putative TERRA promoter; we are now constructing an experimental system to test whether the sequence identified can act as promoter for a reporter gene.

Several ITSs were identified from the now available draft of the horse genome and used for the comparative analysis of the sequence organization of loci orthologous to horse ITSs in different horse breeds and from other equidae species. 9 ITS loci showed either VNTR or presence/absence polymorphism. These loci represent excellent molecular markers for genetic analysis, paternity testing and population genetics. The observation of “empty” ITS loci in the horse population is consistent with the hypothesis that equidae genomes are evolving at a relatively fast rate.

References


