Study of the Loci A, B and C from the STR System MBP based on PCR and Enzyme Restriction Analysis: Populational Data from North Portugal and S. Tomé Príncipe

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The polymorphism of the STR MBP located 5' to exon 1 of the myelin basic protein gene, is primarily determined by variation of the tetranucleotide motif TGGA. The use of a single pair of primers (Polymeropoulos *et al*., 1992) results in the co-amplification of two regions: locus MBPA (209-237 bp) and locus MBPB (121-145 bp), this last one contained inside the first. Treatment of the largest amplified fragments with the restriction enzyme Nla III, allows the individual analysis of the other region that complements MBPA and that is designed as locus MBPC. In this way the study of the three loci turns possible to conduct simultaneously haplotypic analysis, once MBPA is the sum of MBPB and MBPC.

Additionally, it has been described a G→A substitution at position 124 (Gusmão *et al*., 1996; Nelleman *et al.* 1996), whose discrimination obviously increases the global informativeness of the MBP system.

These features confer to this STR a special interest either for use in population studies or forensic casework as well as for understanding patterns of STR evolution.

In this work, we present allelic frequencies concerning MBPA, MBPB and MBPC from North Portugal and from the African population of S. Tome e Principe.

As expected, haplotypic analysis revealed strong gametic disequilibrium between MBPB and MBPC in both populations.

With respect to the a G→A substitution, that we detected by double amplification with two sets of primers, the corresponding frequency estimate was 4.1% in North Portugal and 0.5% in S. Tome e Principe. In either population we also found that the substitution was exclusively associated with haplotypes B10/C11 and B12/C9.