MUTATION RATES AT 20 STR LOCI IN THE POPULATION FROM SÃO PAULO, BRAZIL

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Short tandem repeats (STRs) are genetic markers largely employed in forensic analysis and paternity investigation cases because they tend to be highly polymorphic and present a low mutation rate. When an inconsistency between parent and child is considered as a possible mutation, the mutation rate should be incorporated into paternity index calculations to have a robust result and to reduce the chance of misinterpretation. The aim of this study was to estimate mutation rate of 20 autosomal STRs loci used for paternity tests. In these loci were analyzed 22,130 parent-child allelic transfers from 767 duos or trios paternity tests carried out during 2013-2015 from São Paulo State, Brazil. Blood samples were collected by capillary puncture and DNA was amplified by multiplex PCR and genotyped by capillary electrophoresis. We identified 30 mutations in 15 loci, and they were more frequent in paternal germline when compared to maternal germline. Results showed mutation rates ranging from 0.9 to 3.5 x 10^{-3} and the overall mutation rate was 1.4 x 10^{-3} (95% confidence interval 0.9-1.9 x 10^{-3}). The locus with highest rate was D18S51, and the ones with lower rates were Penta E, CSF1PO, Penta D and D21S11 and we do not identified any mutation in D16S539, D2S1338, TH01, TPOX and D3S1358 loci. The majority of the mutations consisted by losses or gains of one repeat unit. Comparing our results with the literature, in samples from Portugal, China and Brazilian regions, mutations in paternal germline were also more common. Pernambuco, Rio Grande do Sul (Brazil) and also China population, D18S51 locus also showed high mutation rates when compared with other loci, however this did not occur in Portugal; TPOX and TH01 also showed low rates in all cases. However, in some loci, like D21S11, the values differed significantly from our results. This study concluded that the rates found in São Paulo population have peculiarities when compared with rates described on literature, which justifies the use of regional data base in laboratories. Thus, the results of this study are important for obtaining a robust likelihood ratio when mutation rate is needed to be used in paternity probability.

Keywords: Short tandem repeats; Mutation rates; Paternity testing
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