ESTIMATING MUTATION RATES IN 20 MICROSATELLITE AUTOSOMAL LOCI IN ARARAQUARA POPULATION, SAO PAULO STATE, BRAZIL

Fernanda Silva Polverari, Isabela Brunelli Ambrósio; Simone Busko Valim, Luiz Gabriel Sarone Gone, Regina Maria Barretto Cicarelli, Universidade Estadual Paulista
Laboratório de Investigação de Paternidade - Núcleo de Atendimento à Comunidade –
Faculdade de Ciências Farmacêuticas – Campus de Araraquara

The occurrence of mutations in germ cells is passed on to offspring and can affect the results of DNA paternity tests. Due to the high degree of polymorphism, resulting from their high mutational rate, STRs are widely used as markers for paternity testing. For the estimation of these rates is possible to identify the correct genetic profile of individuals and determine the minimal number of mutations that can be regarded as cases of exclusion of paternity. The present study had the aim to estimate the mutation rates at 20 STR autossomal loci in Araraquara population, Sao Paulo State, attempting to generate a genetic database of interest for statistical calculations, such as in paternity tests. We collected blood samples on FTA cards (Whatmann) and used one punch (1.2 mm) directly in PCR amplification with PowerPlex 21 System (Promega); the analysis was carried out in ABI3500 (Applied Biosystems). Germline mutation of 20 STR loci was studied for 9,120 parent-child transfers from 228 paternity testing cases, all of them carried out in Araraquara, Sao Paulo State, from January to December of 2013. The mutations occurred at least in 7 loci (D7S820, Penta D, Penta E, D12S3911, D13S3172, D8S11792, D1S1656), but more loci are expected. The results are in processing to estimate the mutation rates and the origin of the mutation, if maternal or paternal, if there was loss or gain of units repetition, and the number of mutations per loci mutational rates.