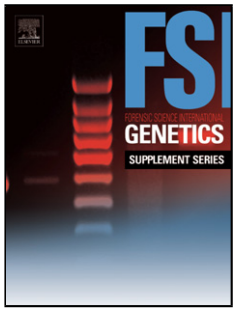

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Mutation Rates for 29 Short tandem repeat Loci from the Ecuadorian Population.

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Abstract

Short tandem repeats (STRs) are sequences that involve repetitive units of 1-6 bp. Therefore, this study recently analyzed a total of 16,310 paternity tests performed from 2012 to 2017 in the Genetic Laboratory of Cruz Roja Ecuatoriana in which 523 mutations were identified in the STRs included in PowerPlex ® 16, PowerPlex® CS7, PowerPlex® Fusion, GlobalFiler® and VeriFiler® Systems Furthermore, among the identified mutations 80 were from maternal, 357 of them were from paternal inheritance and 86 were undetermined. Additionally, the highest number of mutation events occurred in FGA, D21S11 and D12S391. The observed mutational behavior has important implications in forensic DNA analysis for instance determine the exclusion criteria in paternity testing for the correct interpretation. Thus, this study presents the first report of STR's mutation rates for the Ecuadorian population.

Keywords: Mutation rates, Ecuadorian, STRs

Introduction

Short tandem repeats (STRs) constitute a powerful tool for human identification, population genetics, among many others [1] [2]. Mutations in STRs are generally greater (10^{-6} to 10^{-2} nt per generation) in comparison with other genetic markers. The principal mutation occurs in the slippage of the polymerase during DNA replication. When a repetitive region is in replicated, DNA strands are associated incorrectly giving as a result the insertion or deletion of repetitive units [3]. The aim of the present study is to report the mutation rates for 29 autosomal STRs in Ecuadorian population.

Materials and Methods

Our study was performed at the Genetic's Laboratory of Centros Médicos Especializados Cruz Roja. A total of 16310 relationship cases that were done since 2012 to 2017.

DNA was extracted from whole-blood samples on FTA cards and processed using PowerPlex® 16, PowerPlex® CS7, PowerPlex® Fusion, GlobalFiler® and VeriFiler® according to the manufacturer's protocol. Amplified PCR products were analyzed using ABI 3130 and 3500 Genetic Analyzers, Data Collection v2.0, v4.0 and Gene Mapper v3.2 and v5.

The mutation rates were calculated following the AABB recommendations [4]. Moreover, we assume the shortest step and if the mutation could be paternal or maternal we declared as Paternal/Maternal mutation.

Results and Discussion

Among the 16.310 cases, we observed a total of 523 mutations in 29 autosomal STRs (Table 1). Mutations were not detected for PENTA C, D2S441, FESFPS, F13B and LPL.

Conclusion: The present study, we report the first data for mutation rates for 29 autosomal STRs for the Ecuadorian population. We found that the paternal mutation events and rate is 4.97 times higher than the maternal. The results are usefulness for statistical analysis of paternity testing on Ecuadorian population.