Analysis and Interpretation of Genetic Mutations Using Thirteen Short Tandem Repeats in Fifty-Seven Parent/Child Allelic Transfers

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The utilization of Short Tandem Repeats (STR) for paternity identity has gained popularity over the past few years. Current reports regarding parent/child allelic transfer data suggest that STR genetic markers have different mutation rates. The literature cautions that paternity exclusions should not be based on STR typing results from a single STR locus, that at a minimum, two STR mutations constitute an exclusion. In order to determine if there were parent/child allelic transfer mutations within several generations of a single family, the Palm Beach County Sheriff's Office (PBSO) and the Texas Department of Public Safety (TDPS) analyzed a total of fifty-seven individuals. Three generations of Family A (PBSO) consisting of thirty-five individuals and four generations of Family B (TDPS) consisting of twenty-two individuals were analyzed for thirteen STR loci including FGA, vWA, D3S1358, CSF1PO, TPOX, TH01, D18S51, D21S11, D8S1179, D7S820, D13S317, D5S818, and D16S539. Each individual was tested for all genetic markers using PowerPlex™ 1.1 and PowerPlex™ 2.1, Profiler Plus™ and Cofiler™ multiplex systems. Allele detection was accomplished using both the Hitachi FMBIO® fluorescent scanner (PBSO) and the ABI CE310 (TDPS). Six mutations were detected during this study, three individuals from Family A and three individuals from Family B. Mutations were observed for three individuals at D21S11, one individual at CSF1PO, one individual at D8S1179, and one individual at D7S820. All alleles were verified using ABI and Promega STR multiplex systems thus eliminating the possibility of primer mutations. This data supports the recommendation that at least two STR loci must be considered for paternity exclusions.

Reports suggest that most mutations come from the father and although genetic sequencing was not done, it appears that three of the observed mutations were from the mother and three from the father. Interestingly, no single base pair mutations were detected in any of the individuals tested. In fact, all of the mutations identified were of a single repeat length difference of four base pairs as determined both visually and using the ABI GenoTyper® Software and Hitachi STaR Call® software programs. This type of mutation is in concordance with the literature as ninety percent of STR mutations are single step.