

DNA ANALYSIS OF AN EXTENDED PEDIGREE AND EVALUATION OF KINSHIP SOFTWARE TOOLS

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Kinship analysis has become very important in forensic sciences and paternity testing. Having a pedigree of known individuals provides a vast amount of information which can aid in the interpretation of kinship statistics. To further our understanding of how kinship statistics applies to related individuals, biological samples from a large extended family were obtained to specifically study kinship likelihood ratios, mutations and our ability to accurately designate familial relationships. For this kinship study, profiles were generated for 17 autosomal STRs, 17 Y-STRs, and mitochondrial DNA (mtDNA). The results were generated with the PowerPlex 16[®] System (Promega Corporation, Madison, WI), the AmpFLSTR[®] MiniFiler™ PCR Amplification Kit (Applied Biosystems, Foster City, CA) and the AmpFLSTR[®] YFiler™ PCR Amplification Kit (Applied Biosystems). For mtDNA, both HV1 and HV2 were sequenced with BigDye[®] Terminator v.1.1 (Applied Biosystems).

DNA samples from a pedigree containing 46 related individuals were evaluated for various levels of relationship. The primary parental line, P0, consisted of a mother and father that had 12 children. These 12 children (8 males and 4 females) constitute the F1 pedigree. All of their children and grandchildren were also tested, consisting of an additional 22 individual. Five mutations were observed in the analysis of the 46 individuals in this extended family. Autosomal mutations were observed in CSF1PO, D21S11 and Penta E; each of these mutations occurred in a different family member. Interestingly, one male offspring displayed two mutational events, the one in Penta E, and another in DYS635. One point heteroplasmy at 16,093 in HV1 of the mitochondrial genome was observed in the P0 father and compared to his two siblings.

Kinship indices were calculated for various configurations of subsets of the pedigree using two different software programs that are being developed for relationship testing, missing persons and mass disaster identification analyses. These software programs include the analytical module of Laboratory Information System Application (LISA) (Future Technology Incorporated, Fairfax, VA) and GeneMarker[®] HID (SoftGenetics, State College, PA). By studying large family pedigrees such as this one, we are able to have a better understanding of the effectiveness of the software tools available and their utility in complex familial relationship studies.