

The Relationship Testing Application of GeneMarker® HID: Automated Pedigree and Kinship Analysis with Database Searching Capabilities

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Introduction

DNA typing of individuals using STR analysis is a powerful tool in relationship testing. With natural and man-made mass disasters, casualties of war, and reports of missing persons, it is necessary to identify remains using reference samples from the missing individuals or their primary biological relatives (1). Additionally, verifying relationships is imperative in parentage and immigration cases. Programs which automate and expedite relationship testing are of interest to the forensic examiner; the integration of these types of programs with genotyping abilities would be of great benefit to the human identification community. GeneMarker® HID STR Human Identity Software (SoftGenetics, LLC, State College, PA) is a program designed for STR analysis (2) which meets the definition and scope of an expert system as defined by the National Institute of Justice's Expert System Tested Project (3). The STR fragment sizing and allele calling in GeneMarker® HID is directly linked to a Relationship Testing application. This module expedites kinship analyses by automating pedigree construction and calculating kinship indices for different relationship levels. Additionally, the program allows users to store profiles in a closed database, in which unknown profiles can be searched for possible biological relatives or personal effects.

Pedigree Analysis

In the Relationship Testing application of GeneMarker® HID, pedigrees can be designated manually, or the Family Group tool can automatically draw pedigree trios. Using this tool, established naming conventions are defined and used to automatically construct pedigrees for alleged paternity trios. The child's icon in the pedigree tree is linked to the electropherograms for the entire trio. Using the STR profiles for each individual, the software confirms Mendelian allele sharing (Figure 1). The software program will also automatically display loci of conflict between a parent and child or between two siblings. If a conflict in allele sharing is detected, the child's pedigree icon and conflicting loci are highlighted in red (Figure 2). If any sample contains loci with questionable data, the pedigree icon is tagged with a question mark, and the unresolved loci are listed.

In addition to paternity trios, the program can facilitate construction of large familial pedigrees. Biological parents and children can be added to any node; deceased individuals can be designated. Figure 3 depicts a family pedigree of 44 individuals, created and analyzed with the Relationship Testing application. The red icons indicate conflicts; in this pedigree, four individuals displayed mutational events.

References

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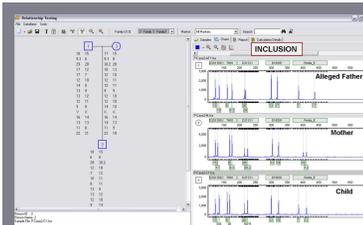


Figure 1: An automated parentage trio where the father is included
This pedigree trio was automatically created using the Family Group tool. Mendelian inheritance of alleles is confirmed automatically and is indicated by the blue pedigree icons. The child's icon is directly linked to the electropherograms of all three individuals.

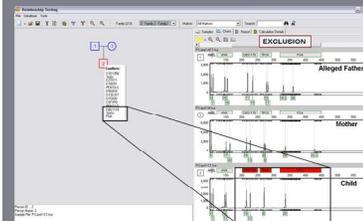


Figure 2: An automated parentage trio where the father is excluded
This figure illustrates a pedigree tree automatically created using the Family Group tool. In contrast to Figure 1, this example illustrates a paternal exclusion.

STR Origin/Allele Agreement

Agreed to this report are possibly related to the following individuals: 2003-029-003-001, 2003-029-003-002, 2003-029-003-003, 2003-029-003-004, 2003-029-003-005, 2003-029-003-006, 2003-029-003-007, 2003-029-003-008, 2003-029-003-009, 2003-029-003-010, 2003-029-003-011, 2003-029-003-012, 2003-029-003-013, 2003-029-003-014, 2003-029-003-015, 2003-029-003-016, 2003-029-003-017, 2003-029-003-018, 2003-029-003-019, 2003-029-003-020, 2003-029-003-021, 2003-029-003-022, 2003-029-003-023, 2003-029-003-024, 2003-029-003-025, 2003-029-003-026, 2003-029-003-027, 2003-029-003-028, 2003-029-003-029, 2003-029-003-030, 2003-029-003-031, 2003-029-003-032, 2003-029-003-033, 2003-029-003-034, 2003-029-003-035, 2003-029-003-036, 2003-029-003-037, 2003-029-003-038, 2003-029-003-039, 2003-029-003-040, 2003-029-003-041, 2003-029-003-042, 2003-029-003-043, 2003-029-003-044, 2003-029-003-045, 2003-029-003-046, 2003-029-003-047, 2003-029-003-048, 2003-029-003-049, 2003-029-003-050, 2003-029-003-051, 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