

## THE RELATIONSHIP TESTING APPLICATION OF GENEMARKER® HID: AUTOMATED PEDIGREE AND KINSHIP ANALYSIS WITH DATABASE SEARCHING CAPABILITIES

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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, there is a need to identify remains using reference samples from the missing individuals or their biological relatives. Additionally, verifying relationships is imperative in paternity and immigration cases. With advances in software technology, programs designed to automate and expedite relationship testing are of great interest to the forensic examiner.

GeneMarker® HID (SoftGenetics, State College, PA) is a software program designed for STR analysis which meets the definition and scope of an expert system as defined by the National Institute of Justice's Expert System Testbed Project (<http://forensics.marshall.edu/NEST/NEST-Intro.html>). GeneMarker® HID has been evaluated as an expert system using PowerPlex® 16 System (Promega Corporation, Madison, WI) and AmpFLSTR® Identifiler® PCR Amplification Kit (Applied Biosystems, Foster City, CA) data electrophoresed on an ABI Prism® 3100 Genetic Analyzer and ABI Prism® 3130xl Genetic Analyzer, respectively (study submitted to the Journal of Forensic Sciences).

In addition to the STR fragment sizing and allele calling, GeneMarker® HID offers a Relationship Testing application. This module features pedigree and kinship analysis tools. Pedigrees can be designated manually or the Family Group Tool can automatically draw pedigree trios when naming conventions are defined. The user can also designate a deceased individual. In the pedigree, the software will use the STR sample files of defined relatives to display consistent allele sharing. The user can click the symbol of any individual in the pedigree tree or the entire trio, and the corresponding electropherogram(s) will be displayed in the adjacent pane. More importantly, the software highlights conflicting allele calls with a color indicator on the pedigree symbol. By clicking the conflicting allele, the electropherogram for that locus is launched. That is, the software program will automatically display loci of conflict between a parent and child or between two siblings. The pedigree also displays a question mark for any sample that contains loci with unresolved rule firings. The Relationship Testing application also includes a Kinship Analysis tool. This module uses STR allele frequencies to determine the likelihood that two individuals have the designated relationship versus the likelihood that the two individuals are unrelated. Statistical analyses are automated with GeneMarker® HID, reporting the probabilities and kinship indices for different relationship levels (e.g., parent(s) to child, sibling to sibling, uncle to nephew, child to grandparent). These statistics are calculated using allele frequencies from major U.S. populations (Budowle et al., 2001) and mutation rates specified by the American Association of Blood Banks (AABB, 2000).

An additional feature of this software program is that it enables the user to create a database of STR profiles, such as those assembled for Missing Persons/Mass Disaster Programs at the University of North Texas Center for Human Identification. By having a closed database, a search can be conducted to rank likelihood ratios of duplicate and potentially related samples. Multiple report files can be created and saved, and the pedigree trees can be exported as bitmap images for inclusion in reports and other documents. This program automates the time-consuming searches and complex statistical analyses for identifying human remains and relationship testing.