GeneMarker® HID
Expert System Human Identification Software with Linked Post-Identification Database Search, Kinship, and DNA Mixture Applications

Promega Webinar
July 24, 2013
SoftGenetics LLC

- Founded: 2001
- Mission:
  “Provide Biologists with highly accurate, sensitive & user-friendly analysis tools.”
- Founders:
  CS Liu, PhD. VP Development. VP Software Development, Spectrumedix 17 Issued Patents
  John Fosnacht, VP Sales & Marketing VP Sales & Marketing, Spectrumedix ; 2 years President, AccuStandard, 4 years General Manager EMD Reagents USA, 17 years
Product Portfolio:

**GeneMarker®/MTP Genotyping**
- AFLP®/TRFLP
- Microsatellite, Trisomy
- MLPA®, MS-MLPA
- LOH/ MSI
- SNaPshot®

**GeneMarker®/HID STR Analysis**
- Human Identity Expert System
- Mixture Applications
- Familial and Exact Match Database Search
- Parentage and Kinship Testing

**Mutation Surveyor® for Sanger Sequence**
- mtDNA Analysis and SNP detection
- Detect Heteroplasmy
- Patented Core Technology

**NextGENe® analysis of NGS Sequencing**
- Patent Pending Core Technology Condensation of “short reads”
- Analysis of mtDNA, STRs and mixtures

**ChimerMarker™**
- STR Analysis for Chimerism Testing
  - Single Donor Chimerism
  - Double Donor Chimerism
  - Long-Term Monitoring
  - MCC Analysis

**GeneticistAssistant™**
- NGS Web-based visualization tool

**JelMarker®**
- Reading and converting gel images

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Second generation sequencing allows for mtDNA mixture deconvolution and high resolution detection of heteroplasmy. *Holland MM, McQuillan MR, O'Hanlon KA.*

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SoftGenetics Research Use Only (RUO) Genetics Analysis Software
GeneMarker® HID (Human Identity)

- **Concordant, Validated Expert System**
  - Alternative to Genotyper®, GeneMapper® IDX
  - Compatible with major chemistries, CE and Rapid Systems output; Windows® XP, Vista, 7 and 8

- **Single Source casework and databasing**
  - Streamlined Workflow with Audit Trail
  - CODIS compatible reporting

- **Reference Sample and Familial Search**
  - Mass Disaster, Crime Scene Response --- No Data Transfer Needed
  - Positive Matches Ranked by Likelihood Ratio

- **Kinship and Paternity Testing**
  - Identity by Descent (IBD) Calculations with pre-loaded or custom allele frequency tables
  - Automated Pedigree drawing and AABB trio/single parent PI calculations

- **Mixture Analysis**
  - Detects Mixtures and calculates Probability of Inclusion and Exclusion (PI, PE) for all mixtures
  - Performs Statistics two contributor including LR and searches database for deduced contributor (with or without reference sample)
Review Allele Calls In Main Analysis Screen or All Color Browser

Any edits or comments are recorded as part of the audit trail.
Project Panel .XML Recognizes Variant Alleles and Shifts Bins to Fit Allelic Ladders
Marker Specific Settings to Customize to the Lab Standard Operating Procedure
## Report Table Format Options:

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### Set Peak Table Columns

- **Style**: Allele List, Forensics, Ein Table, Peak Table, Allele Count, Grouped by Markers
- **Options**: Size Range (Base), Allele By Panel, Quality Score, Statt End, Allele Comments, Sample Comments, Quality Reasons
- **Columns**:
  - Allele
  - Marker
  - Size
  - Height
  - Ratio
  - Ct

### Allele Report Settings

- **Report Style**: Allele List, Forensics
- **Options**: Show All Alleles, Show Only Maximum Alleles, Show Reflected Low Score Alleles, Show Extra Sample Names
Expert System Summary

Accurate, Rapid Analysis of Forensic STR Data
Quality Flagging, Size Calibration Check, Automated Control Concordance

Compatible with all major file types
from CE and Rapid Instruments and Human Identity PCR Kits and Custom Chemistries

User Management, Access Rights and Audit Trail

Linked Navigation – Time Savings and Automated Edit History

Report Flexibility – Allele, Peak Table and CODIS formats
Selected Presentations and Journal Articles

- Expert System, Concordance with GeneMapper®
  - estimated analysis time savings 25%

- Autosomal and Y STR analysis of LCN data

“.GeneMarker HID to be a more efficient software system....results in a gain of information and a significant reduction in the number of edits needed per sample..making it the preferred software package...NYC OCME”

Evaluation of GeneMapper® ID-X and GeneMarker® HID for use in Forensic DNA Analysis

Ronald Schmidt BS¹, Justin Godby MSFS¹, Valerie Bostwick MSFS¹, Theresa Caragine PhD²

¹ Marshall University Forensic Science Center – 1401 Forensic Science Drive, Huntington, West Virginia 25701
² New York City – Office of the Chief Medical Examiner – 421 East 26th Street, New York City, New York 10016

GeneMarker HID better able to resolve alleles with Single-Base-Pair
Example of Single Base Pair Resolution

Samples amplified with PowerPlex® Fusion and separated on a Applied Biosystems® 3500 CE

Minor component allele 19 is automatically called in 2 out of 3 samples using default analysis settings

Manually insert allele 19 using a right mouse click to select insert allele
Searchable Database - Relationship Testing

Match Reference Samples or Family Members
Likelihood Ratios calculated using Identity by Descent

- Missing Persons, Mass Disasters
- Immigration / Human Trafficking
- Detect Bioterrorism – with the appropriate chemistry development
- Paternity/Kinship -- Three Generations (IBD)
- Pedigree Diagrams – Visualize Allele Conflicts
  PI Calculations  AABB Recommendations
Missing Person Example

Likelihood Ratios of Relationship Level versus Random Person From the Population Calculated using Identity by Descent Equations and Allele Frequency Tables. Highest scoring relationship level displayed in exportable table.
## Kinship Analysis Across Three Generations

#### Kinship Analysis Table

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#### Kinship Analysis Settings

- **Relationships:**
  - **Unrelated**
  - **Parent/Child**
  - **Full-Sibs**
  - **Half-Sibs**
  - **Cousins**
  - **Uncle-Nephew**
  - **Grandparent-Grandchild**

- **Report Content:**
  - **Likelihood Ratio**
  - **Probability**
  - **Both**

---

**Note:** The table and settings are designed for genetic analysis, allowing for the examination of kinship across three generations. The relationships and likelihood ratios are used to determine the genetic similarity and potential familial connections.
## Automated Pedigree Drawing – Inclusion Example

![Pedigree Diagram](image)

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Automated Pedigree Drawing – Exclusion Example

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- **AMEL**: Y
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- **D18S156**: 15
- **D25S441**: 11
- **D10S1248**: 17
- **D13S317**: 17
- **PENTA E**: 16
- **D16S539**: 9
- **D18S51**: 13
- **D21S330**: 17
- **CSF1PO**: 12
- **PENTA D**: 13
- **TH01**: 33
- **vWA**: 18
- **D21S11**: 28
- **D7S820**: 12
- **D5S818**: 13
- **TPCX**: 8
- **DYS389I**: 10/10
- **D8S1179**: 15/15
- **D12S391**: 24/15

Individual ID: 3
Individual Name: 3
Sample File: Fusion_Tiec_E_500bp_2005.hiw
Applications Include:
- Missing Persons, Mass Disaster, Paternity, Immigration, Human Trafficking
- Linked to Main Analysis – No Data Transfer
- Search Database for Exact Matches and Familial Search
- Automates Repetitive Calculations LR using Identity by Descent
- Kinship Analysis Across Three Generations
- Immediate Visualization of Allele Conflicts in Pedigree Drawings
- Paternity Index Calculations using Standards for Relationship Testing, AABB
Mixture Analysis Application – Assists Forensic Experts

- No Data Transfer – Linked to Results of Genotyping Analysis
- ISFG and SWGDAM Guidelines and Recommendations
- Customize Analysis Parameters to Lab Standards
- Automates Repetitive Calculations: PI, PE, CPI, CPE, RMNE, LR
### Mixture Analysis

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### PowerFlex 16: US Asian American

- **Marker**: D8S1858
  - **Major Mix**: 16.17
  - **Minor Mix**: 0.97
- **Contributor 1**: Female_F07
- **Contributor 2**: Male_G04

### PowerFlex 16: US Caucasian

- **Marker**: D8S1858
  - **Major Mix**: 14.15
  - **Minor Mix**: 0.97
- **Contributor 1**: Male_G04
- **Contributor 2**: Female_F07

**Comment:**
- Contesting minor contributor (male sample) LR displayed for results using each of the population allele frequencies.
Mixture Analysis Assistant Summary

- Developed using recommendations of the DNA Commission of the International Society of Forensic Genetics, compatible with SWGDAM Guidelines

- Linked to Main Analysis – No Data Transfer Needed

- Automates Repetitive Calculations

- Calculates PI, PE, LR, RMNE

- Database search for contributor; with or without a reference sample
Quality Control and Validation Tools:

- Project Comparison
- Replicate Comparison

Highlights any Differences – Table is Linked to Electropherograms
IN CONCLUSION, GeneMarker HID:

**Expert System**
- Documented Time Savings
- Decrease Backlogs
- Concordant / Validated

**Searchable Database / Relationship Testing**
- Linked to Main Analysis – No Manual Data Transfer
- Missing Persons, Mass Disaster Applications
- Locate Same Individual or Family Members (IBD)
- Kinship Across Three Generations (IBD)
- Paternity/Immigration Pedigrees and Paternity Index (AABB relationship testing guidelines appendix 8, trios and single parent cases)

**Mixture Analysis**
- Linked to Main Analysis – No Manual Data Transfer
- Customize Analysis Parameters
- Automate Repetitive Calculations, PE, PI, RMNE, LR
- Search Database – Calculate LR
Thank you for attending this presentation on GeneMarker HID!

Acknowledgments: we are grateful to the following people for sharing their thoughts and experience to assist us during development

Dr. Mitchell Holland, Penn State University Forensics Program
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Drs. John Butler, Kristen O’Connor, Pete Valone and Michael Coble, NIST
Drs. Rhonda Roby, John Planz, Arthur Eisenburg, and Nicole Philips, UNT CHI

Drs. Andy Hopwood and Bob McLaren, Promega, for providing data