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

Promega Webinar
July 24, 2013

Promega

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®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

Next*GENe* ® analysis of NGS Sequencing

- Patent Pending Core Technology
  Condensation of "short reads"
- Analysis of mtDNA, STRs and mixtures

Croat Med J. 2011 Jun;52(3):299-313.
Second generation sequencing allows for mtDNA mixture deconvolution and high resolution detection of heteroplasmy.
Holland MM, McQuillan MR, O'Hanlon KA.

GeneMarker®/MTP Genotyping

AFLP®/TRFLP
Microsatellite, Trisomy
MLPA®, MS-MLPA
LOH/ MSI
SNaPshot®

ChimerMarker™

STR Analysis for Chimerism Testing

Single Donor Chimerism
Double Donor Chimerism
Long-Term Monitoring
MCC Analysis

GeneticistAssistant™

NGS Web-based visualization tool

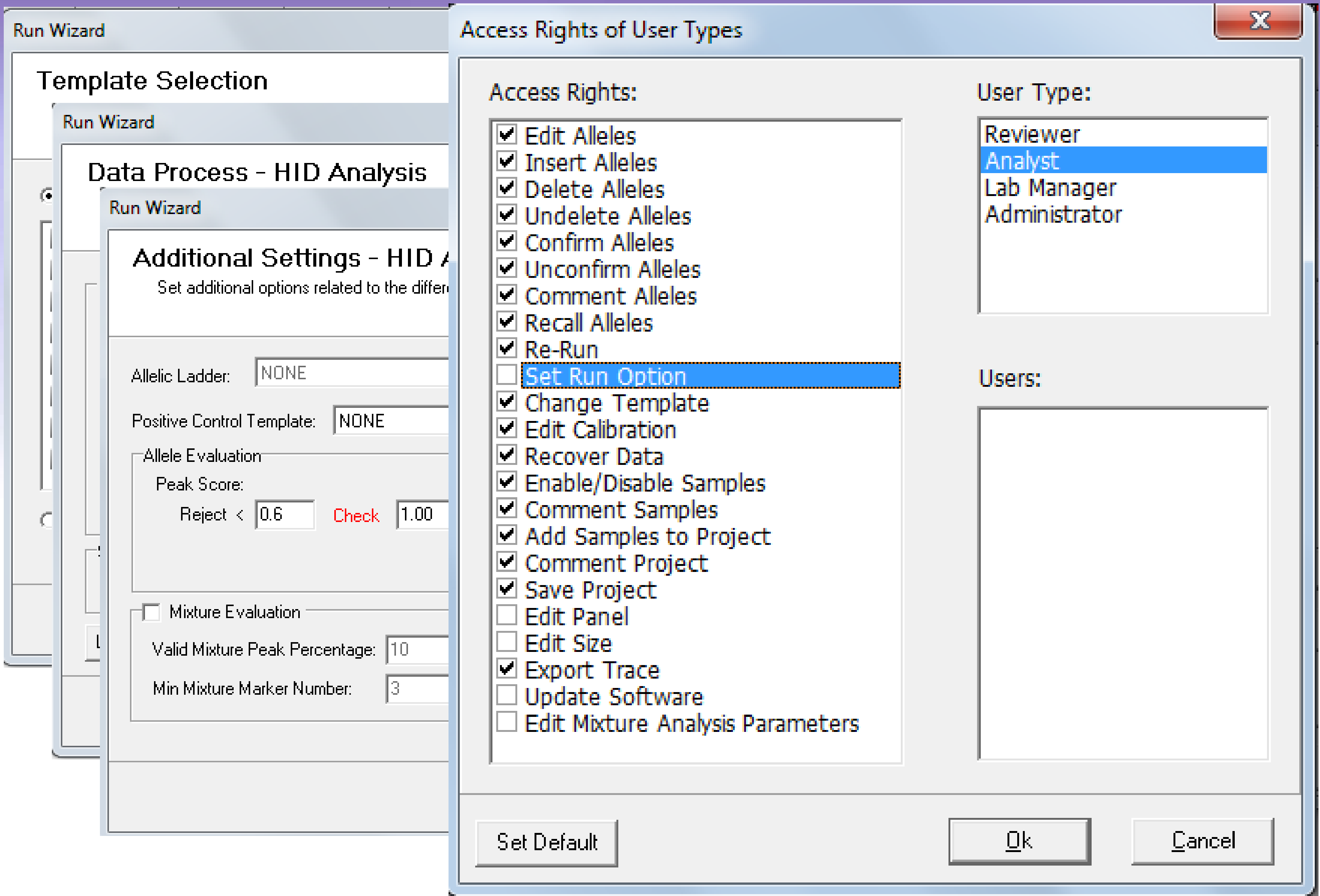
JelMarker®

Reading and converting gel images

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)



Run Wizard

Template Selection

Run Wizard

Data Process - HID Analysis

Run Wizard

Additional Settings - HID A

Set additional options related to the differ

Allelic Ladder: NONE

Positive Control Template: NONE

Allele Evaluation

Peak Score:

Reject < 0.6 Check 1.00

☐ Mixture Evaluation

Valid Mixture Peak Percentage: 10

Min Mixture Marker Number: 3

Access Rights of User Types

Access Rights:

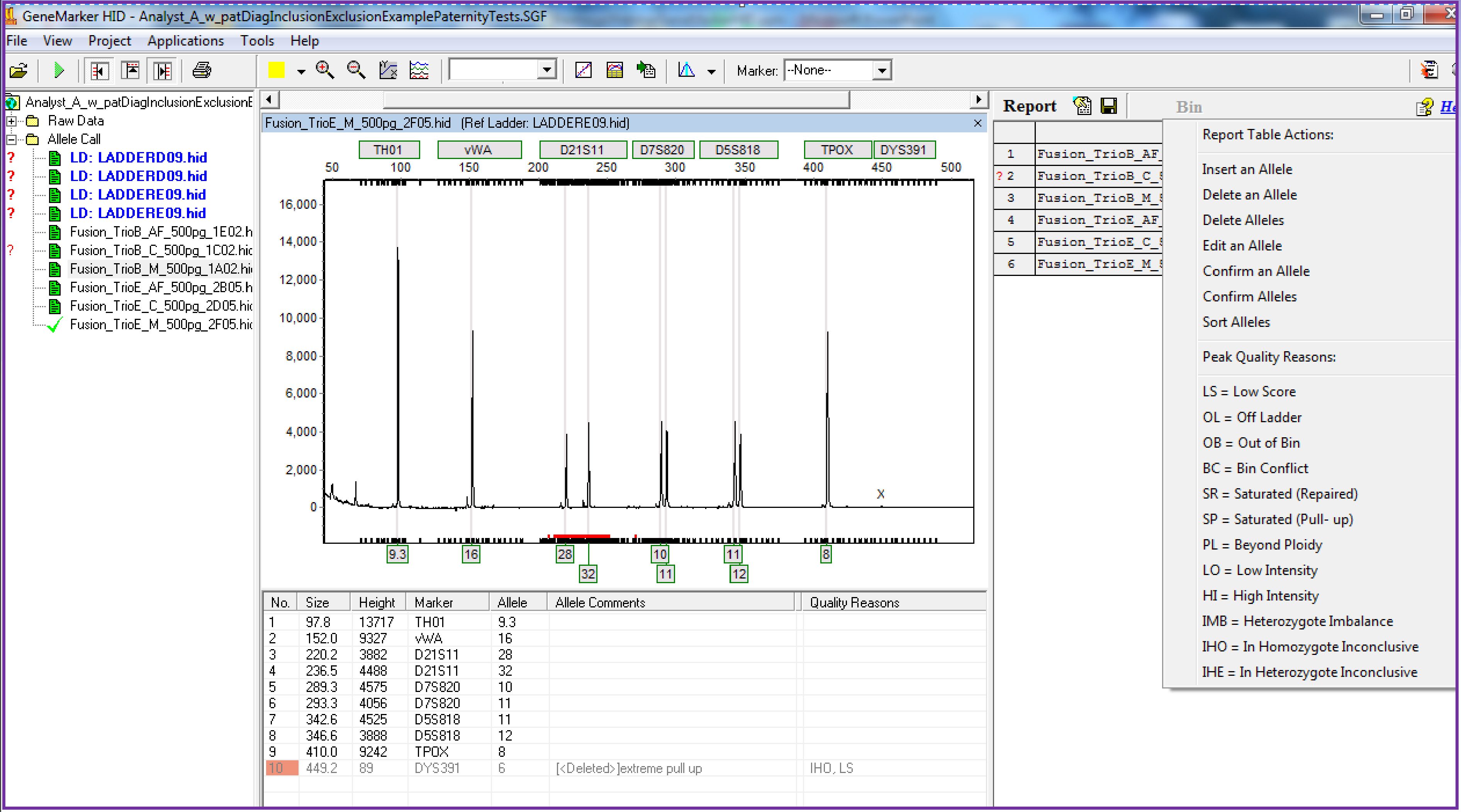
- ☑ Edit Alleles
- ☑ Insert Alleles
- ☑ Delete Alleles
- ☑ Undelete Alleles
- ☑ Confirm Alleles
- ☑ Unconfirm Alleles
- ☑ Comment Alleles
- ☑ Recall Alleles
- ☑ Re-Run
- ☐ Set Run Option
- ☑ Change Template
- ☑ Edit Calibration
- ☑ Recover Data
- ☑ Enable/Disable Samples
- ☑ Comment Samples
- ☑ Add Samples to Project
- ☑ Comment Project
- ☑ Save Project
- ☐ Edit Panel
- ☐ Edit Size
- ☑ Export Trace
- ☐ Update Software
- ☐ Edit Mixture Analysis Parameters

User Type:

- Reviewer
- Analyst
- Lab Manager
- Administrator

Users:

Review Allele Calls In Main Analysis Screen or All Color Browser
Any edits or comments are recorded as part of the audit trail

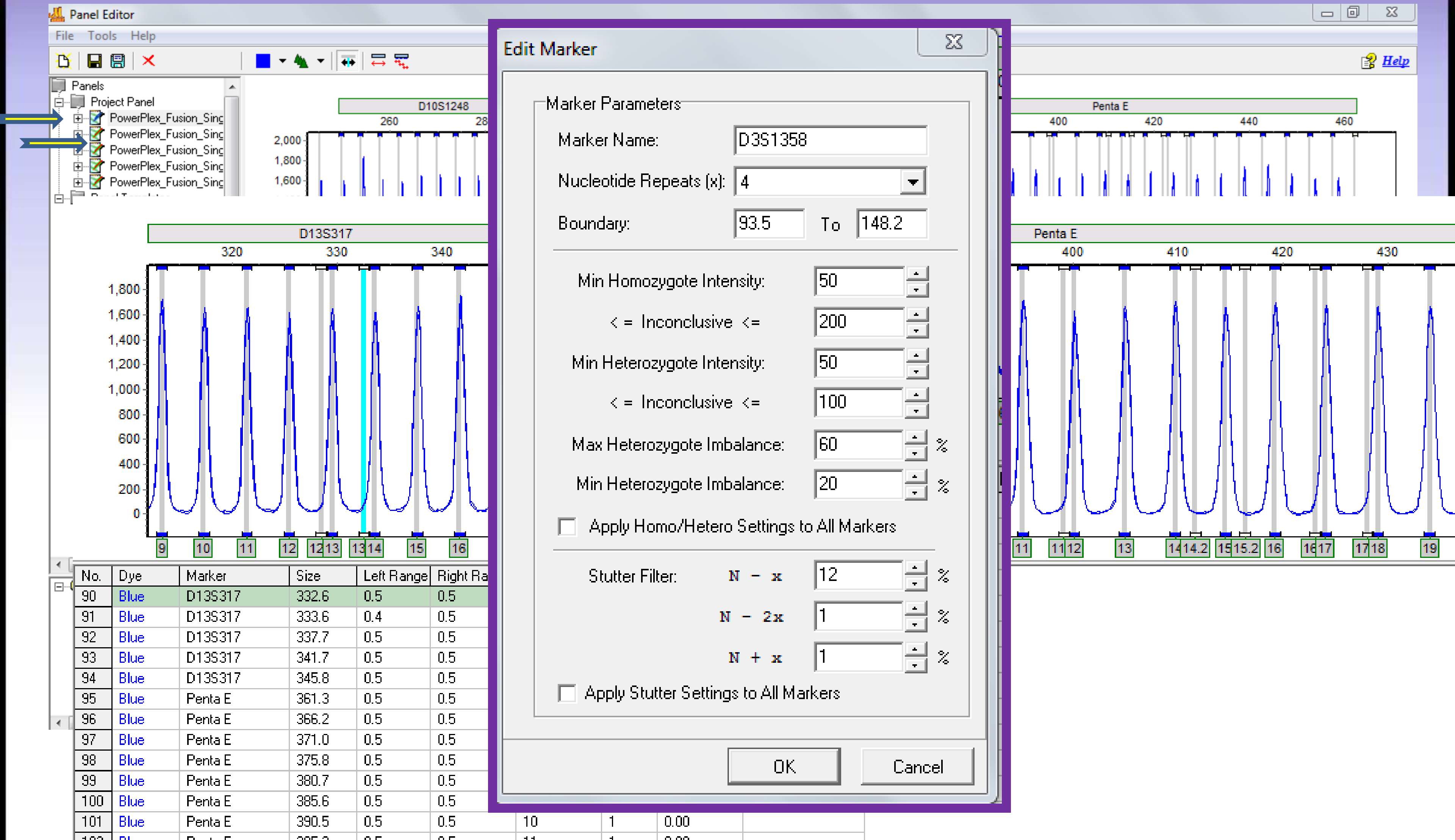


No.	Size	Height	Marker	Allele	Allele Comments	Quality Reasons
1	97.8	13717	TH01	9.3		
2	152.0	9327	vWA	16		
3	220.2	3882	D21S11	28		
4	236.5	4488	D21S11	32		
5	289.3	4575	D7S820	10		
6	293.3	4056	D7S820	11		
7	342.6	4525	D5S818	11		
8	346.6	3888	D5S818	12		
9	410.0	9242	TPOX	8		
10	449.2	89	DYS391	6	[<Deleted>]extreme pull up	IHO, LS

Peak Quality Reasons:

LS = Low Score
OL = Off Ladder
OB = Out of Bin
BC = Bin Conflict
SR = Saturated (Repaired)
SP = Saturated (Pull- up)
PL = Beyond Ploidy
LO = Low Intensity
HI = High Intensity
IMB = Heterozygote Imbalance
IHO = In Homozygote Inconclusive
IHE = In Heterozygote Inconclusive

Project Panel .XML Recognizes Variant Alleles and Shifts Bins to Fit Allelic Ladders Marker Specific Settings to Customize to the Lab Standard Operating Procedure



Edit Marker

Marker Parameters

Marker Name: D3S1358

Nucleotide Repeats (x): 4

Boundary: 93.5 To 148.2

Min Homozygote Intensity: 50

< = Inconclusive <= 200

Min Heterozygote Intensity: 50

< = Inconclusive <= 100

Max Heterozygote Imbalance: 60 %

Min Heterozygote Imbalance: 20 %

☐ Apply Homo/Hetero Settings to All Markers

Stutter Filter: N − x 12 %

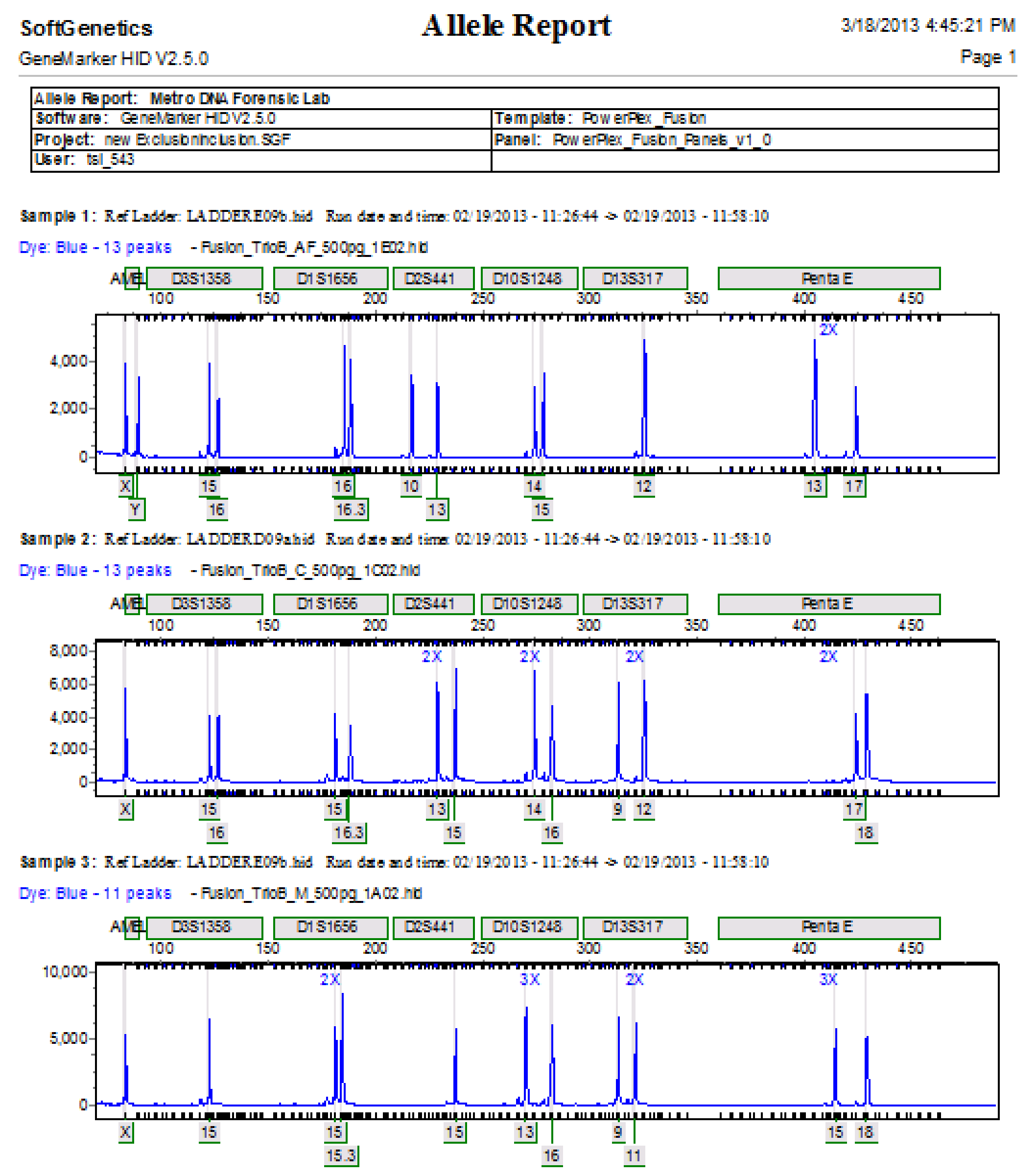
N − 2x 1 %

N + x 1 %

☐ Apply Stutter Settings to All Markers

No.	Dye	Marker	Size	Left Range	Right Ra			
90	Blue	D13S317	332.6	0.5	0.5			
91	Blue	D13S317	333.6	0.4	0.5			
92	Blue	D13S317	337.7	0.5	0.5			
93	Blue	D13S317	341.7	0.5	0.5			
94	Blue	D13S317	345.8	0.5	0.5			
95	Blue	Penta E	361.3	0.5	0.5			
96	Blue	Penta E	366.2	0.5	0.5			
97	Blue	Penta E	371.0	0.5	0.5			
98	Blue	Penta E	375.8	0.5	0.5			
99	Blue	Penta E	380.7	0.5	0.5			
100	Blue	Penta E	385.6	0.5	0.5			
101	Blue	Penta E	390.5	0.5	0.5	10	1	0.00

Reporting Options -- Print/Save Allele Reports



SoftGenetics **Allele Report** 3/18/2013 4:45:21 PM

GeneMarker HID V2.5.0 Page 1

Allele Report: Metro DNA Forensic Lab	
Software: GeneMarker HID V2.5.0	Template: PowerPlex_Fusion
Project: new Exclusioninclusion.SGF	Panel: PowerPlex_Fusion_Panels_v1_0
User: tsl_543	

Sample 1: Ref Ladder: LADDERE09b.hid Run date and time: 02/19/2013 - 11:26:44 -> 02/19/2013 - 11:58:10

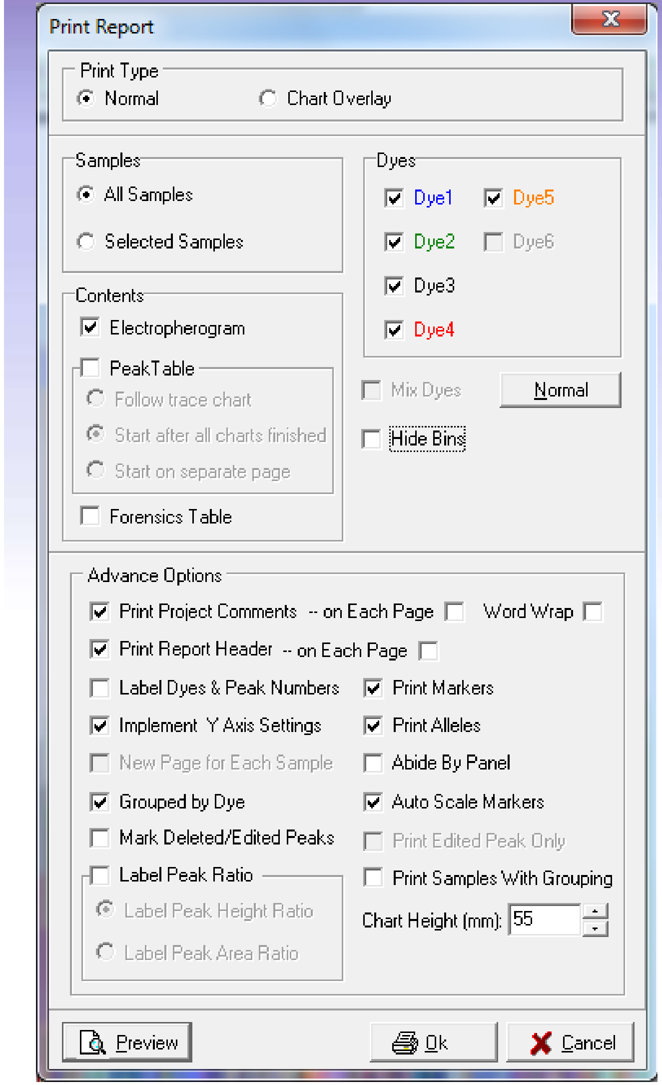
Dye: Blue - 13 peaks - Fusion_TrioB_AF_500pg_1E02.hid

Sample 2: Ref Ladder: LADDERD09a.hid Run date and time: 02/19/2013 - 11:26:44 -> 02/19/2013 - 11:58:10

Dye: Blue - 13 peaks - Fusion_TrioB_C_500pg_1C02.hid

Sample 3: Ref Ladder: LADDERE09b.hid Run date and time: 02/19/2013 - 11:26:44 -> 02/19/2013 - 11:58:10

Dye: Blue - 11 peaks - Fusion_TrioB_M_500pg_1A02.hid



Print Report

Print Type
● Normal ○ Chart Overlay

Samples
● All Samples
○ Selected Samples

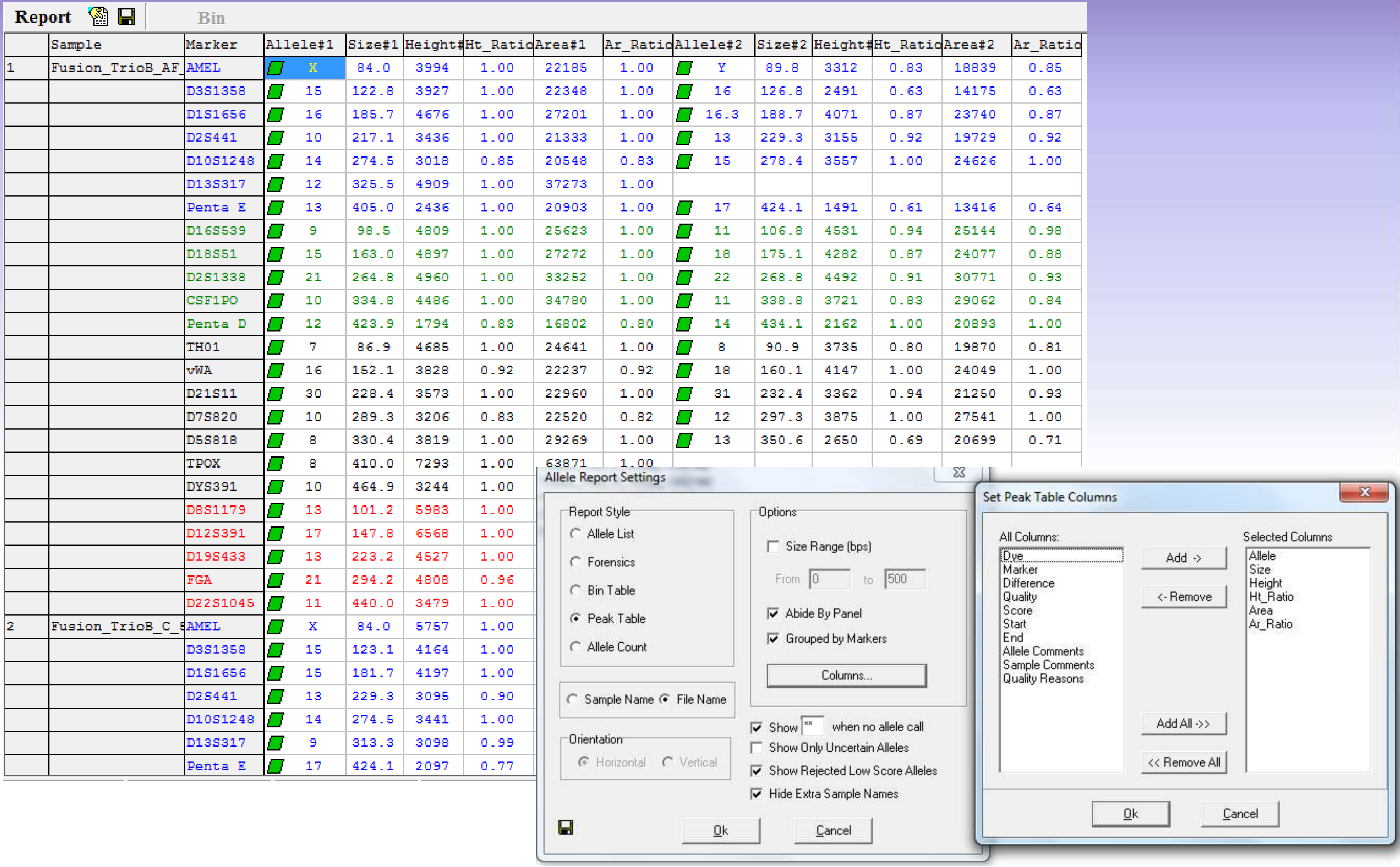
Dyes
☑ Dye1 ☑ Dye5
☑ Dye2 ☐ Dye6
☑ Dye3
☑ Dye4

Contents
☑ Electropherogram
☐ PeakTable
○ Follow trace chart
● Start after all charts finished
○ Start on separate page
☐ Forensics Table

☐ Mix Dyes
☐ Hide Bins

Advance Options
☑ Print Project Comments -- on Each Page ☐ Word Wrap ☐
☑ Print Report Header -- on Each Page ☐
☐ Label Dyes & Peak Numbers ☑ Print Markers
☑ Implement Y Axis Settings ☑ Print Alleles
☐ New Page for Each Sample ☐ Abide By Panel
☑ Grouped by Dye ☑ Auto Scale Markers
☐ Mark Deleted/Edited Peaks ☐ Print Edited Peak Only
☐ Label Peak Ratio ☐ Print Samples With Grouping
● Label Peak Height Ratio
○ Label Peak Area Ratio
Chart Height (mm): 55

Report Table Format Options:



Report Bin

	Sample	Marker	Allele#1	Size#1	Height#	Ht_Ratio	Area#1	Ar_Ratio	Allele#2	Size#2	Height#	Ht_Ratio	Area#2	Ar_Ratio
1	Fusion_TrioB_AF_	AMEL	🟩 X	84.0	3994	1.00	22185	1.00	🟩 Y	89.8	3312	0.83	18839	0.85
		D3S1358	🟩 15	122.8	3927	1.00	22348	1.00	🟩 16	126.8	2491	0.63	14175	0.63
		D1S1656	🟩 16	185.7	4676	1.00	27201	1.00	🟩 16.3	188.7	4071	0.87	23740	0.87
		D2S441	🟩 10	217.1	3436	1.00	21333	1.00	🟩 13	229.3	3155	0.92	19729	0.92
		D10S1248	🟩 14	274.5	3018	0.85	20548	0.83	🟩 15	278.4	3557	1.00	24626	1.00
		D13S317	🟩 12	325.5	4909	1.00	37273	1.00						
		Penta E	🟩 13	405.0	2436	1.00	20903	1.00	🟩 17	424.1	1491	0.61	13416	0.64
		D16S539	🟩 9	98.5	4809	1.00	25623	1.00	🟩 11	106.8	4531	0.94	25144	0.98
		D18S51	🟩 15	163.0	4897	1.00	27272	1.00	🟩 18	175.1	4282	0.87	24077	0.88
		D2S1338	🟩 21	264.8	4960	1.00	33252	1.00	🟩 22	268.8	4492	0.91	30771	0.93
		CSF1PO	🟩 10	334.8	4486	1.00	34780	1.00	🟩 11	338.8	3721	0.83	29062	0.84
		Penta D	🟩 12	423.9	1794	0.83	16802	0.80	🟩 14	434.1	2162	1.00	20893	1.00
		TH01	🟩 7	86.9	4685	1.00	24641	1.00	🟩 8	90.9	3735	0.80	19870	0.81
		vWA	🟩 16	152.1	3828	0.92	22237	0.92	🟩 18	160.1	4147	1.00	24049	1.00
		D21S11	🟩 30	228.4	3573	1.00	22960	1.00	🟩 31	232.4	3362	0.94	21250	0.93
		D7S820	🟩 10	289.3	3206	0.83	22520	0.82	🟩 12	297.3	3875	1.00	27541	1.00
		D5S818	🟩 8	330.4	3819	1.00	29269	1.00	🟩 13	350.6	2650	0.69	20699	0.71
		TPOX	🟩 8	410.0	7293	1.00	63871	1.00						
		DYS391	🟩 10	464.9	3244	1.00								
		D8S1179	🟩 13	101.2	5983	1.00								
		D12S391	🟩 17	147.8	6568	1.00								
		D19S433	🟩 13	223.2	4527	1.00								
		FGA	🟩 21	294.2	4808	0.96								
		D22S1045	🟩 11	440.0	3479	1.00								
2	Fusion_TrioB_C_5	AMEL	🟩 X	84.0	5757	1.00								
		D3S1358	🟩 15	123.1	4164	1.00								
		D1S1656	🟩 15	181.7	4197	1.00								
		D2S441	🟩 13	229.3	3095	0.90								
		D10S1248	🟩 14	274.5	3441	1.00								
		D13S317	🟩 9	313.3	3098	0.99								
		Penta E	🟩 17	424.1	2097	0.77								

Allele Report Settings

Report Style
- ○ Allele List
- ○ Forensics
- ○ Bin Table
- ● Peak Table
- ○ Allele Count

○ Sample Name ● File Name

Orientation
● Horizontal ○ Vertical

Options
- ☐ Size Range (bps)
- From 0 to 500
- ☑ Abide By Panel
- ☑ Grouped by Markers

Columns...

- ☑ Show ** when no allele call
- ☐ Show Only Uncertain Alleles
- ☑ Show Rejected Low Score Alleles
- ☑ Hide Extra Sample Names

Ok Cancel

Set Peak Table Columns

All Columns:		Selected Columns
Dye Marker Difference Quality Score Start End Allele Comments Sample Comments Quality Reasons	Add -> <- Remove Add All ->> << Remove All	Allele Size Height Ht_Ratio Area Ar_Ratio

Ok Cancel

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

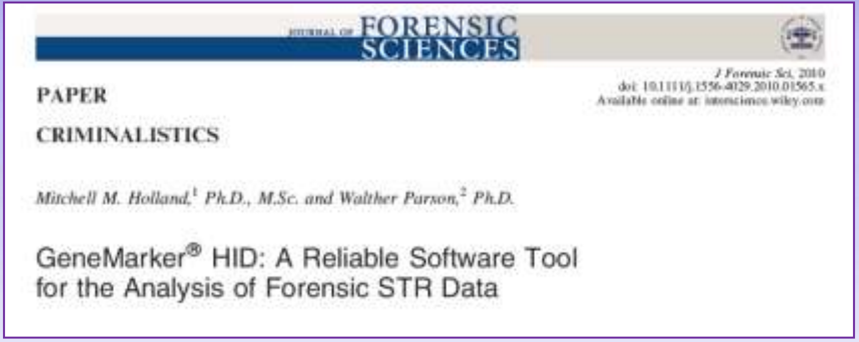
User Management, Access Rights and Audit Trail

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%



JOURNAL OF FORENSIC SCIENCES

J Forensic Sci, 2010
doi: 10.1111/j.1556-4029.2010.01565.x
Available online at: interscience.wiley.com

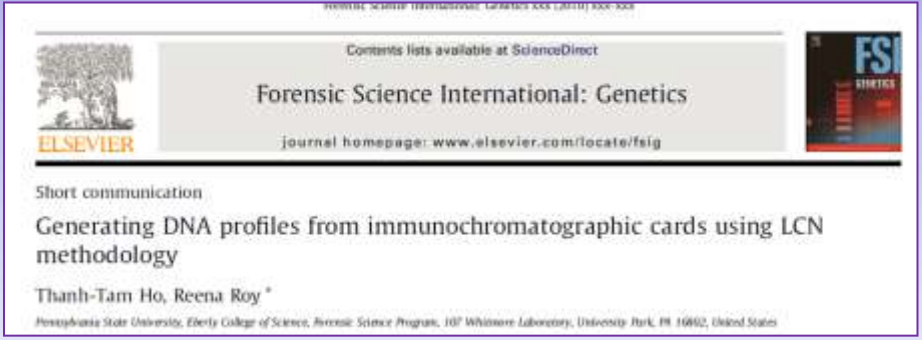
PAPER

CRIMINALISTICS

Mitchell M. Holland,[1] Ph.D., M.Sc. and Walther Parson,[2] Ph.D.

GeneMarker® HID: A Reliable Software Tool
for the Analysis of Forensic STR Data

Autosomal and Y STR analysis of LCN data



Contents lists available at ScienceDirect

Forensic Science International: Genetics

journal homepage: www.elsevier.com/locate/fsig

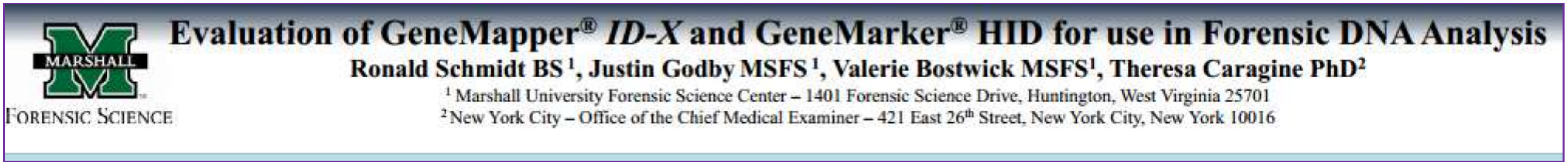
Short communication

Generating DNA profiles from immunochromatographic cards using LCN methodology

Thanh-Tam Ho, Reena Roy *

Pennsylvania State University, Eberly College of Science, Forensic Science Program, 107 Whitmore Laboratory, University Park, PA 16802, United States

"..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 BS[1], Justin Godby MSFS[1], Valerie Bostwick MSFS[1], Theresa Caragine PhD[2]

[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

MARSHALL
FORENSIC SCIENCE

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

Promega

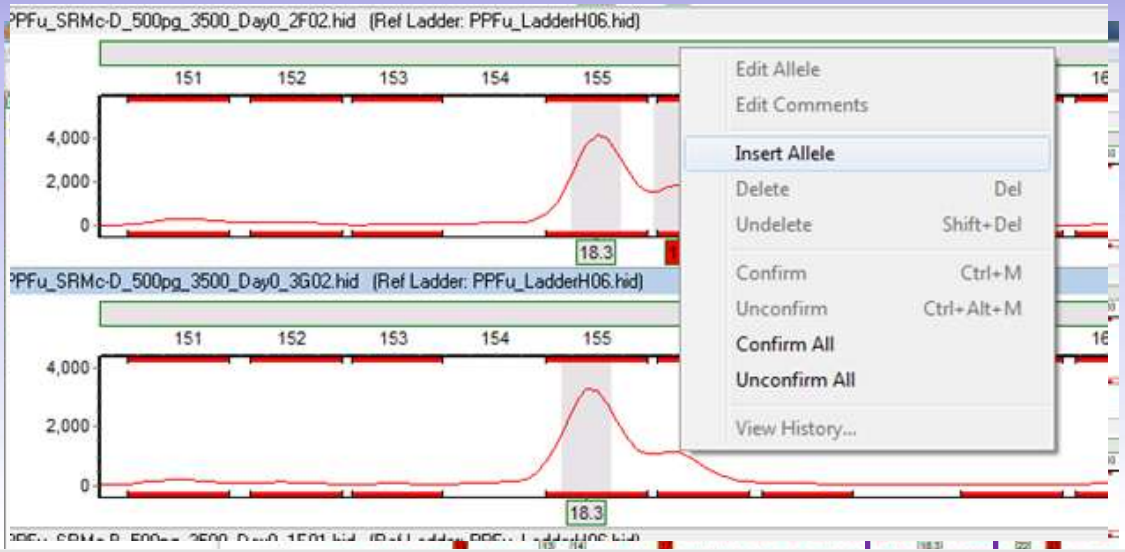
PowerPlex® ESI Systems: D12S391 and D2S441. Resolving Alleles With Single-Base-Pair...

Example of Single Base Pair Resolution

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

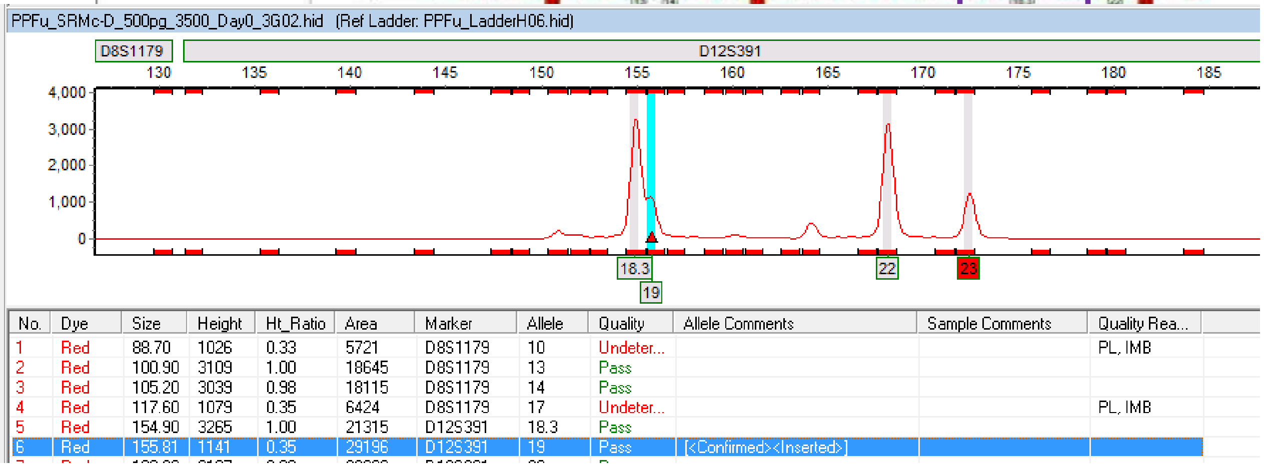
Minor component allele 19 is automatically called in 2 out of 3 samples using default analysis settings

Manually insert allele 19 using a right mouse click to select insert allele



PPFu_SRMc-D_500pg_3500_Day0_2F02.hid (Ref Ladder: PPFu_LadderH06.hid)

PPFu_SRMc-D_500pg_3500_Day0_3G02.hid (Ref Ladder: PPFu_LadderH06.hid)



PPFu_SRMc-D_500pg_3500_Day0_3G02.hid (Ref Ladder: PPFu_LadderH06.hid)

No.	Dye	Size	Height	Ht_Ratio	Area	Marker	Allele	Quality	Allele Comments	Sample Comments	Quality Rea...
1	Red	88.70	1026	0.33	5721	D8S1179	10	Undeter...			PL, IMB
2	Red	100.90	3109	1.00	18645	D8S1179	13	Pass			
3	Red	105.20	3039	0.98	18115	D8S1179	14	Pass			
4	Red	117.60	1079	0.35	6424	D8S1179	17	Undeter...			PL, IMB
5	Red	154.90	3265	1.00	21315	D12S391	18.3	Pass			
6	Red	155.81	1141	0.35	29196	D12S391	19	Pass	[<Confirmed><Inserted>]		

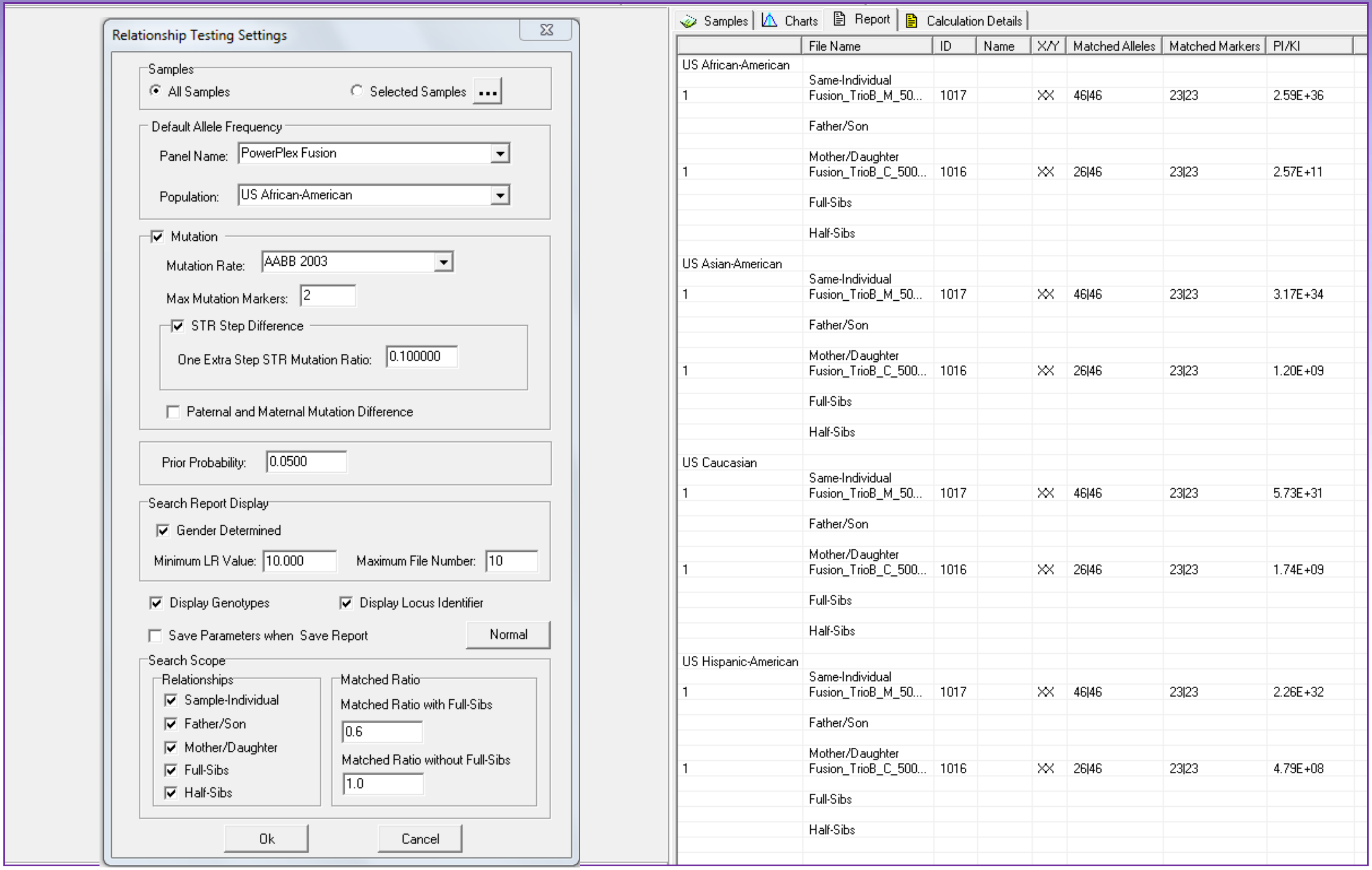
Searchable Database - Relationship Testing

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



Relationship Testing Settings

Samples
● All Samples ○ Selected Samples

Default Allele Frequency
Panel Name: PowerPlex Fusion
Population: US African-American

☑ Mutation
Mutation Rate: AABB 2003
Max Mutation Markers: 2
☑ STR Step Difference
One Extra Step STR Mutation Ratio: 0.100000
☐ Paternal and Maternal Mutation Difference

Prior Probability: 0.0500

Search Report Display
☑ Gender Determined
Minimum LR Value: 10.000 Maximum File Number: 10

☑ Display Genotypes ☑ Display Locus Identifier
☐ Save Parameters when Save Report

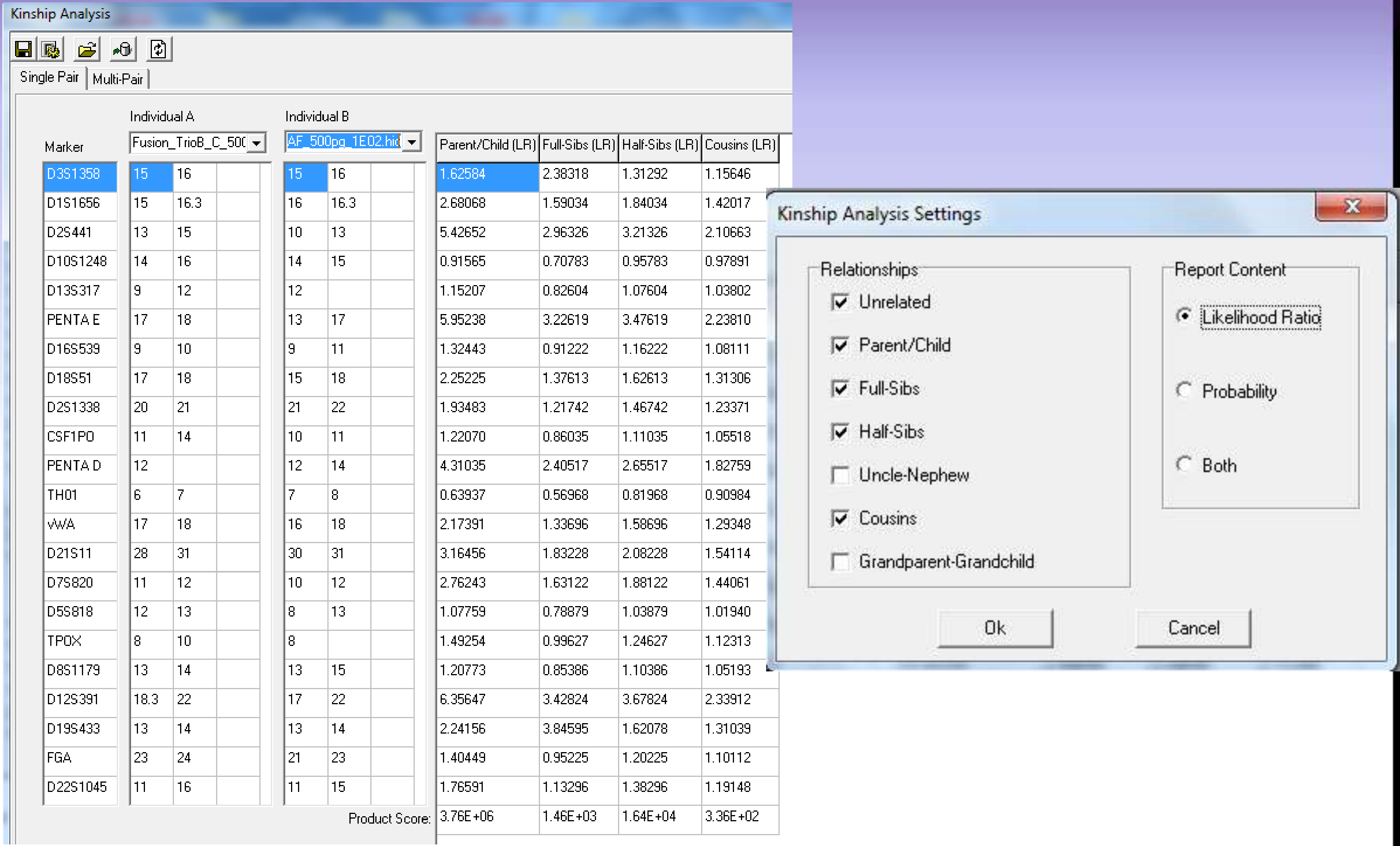
Search Scope
Relationships
☑ Sample-Individual
☑ Father/Son
☑ Mother/Daughter
☑ Full-Sibs
☑ Half-Sibs

Matched Ratio
Matched Ratio with Full-Sibs: 0.6
Matched Ratio without Full-Sibs: 1.0

	File Name	ID	Name	X/Y	Matched Alleles	Matched Markers	PI/KI
US African-American							
1	Same-Individual Fusion_TrioB_M_50...	1017		XX	46|46	23|23	2.59E+36
	Father/Son						
1	Mother/Daughter Fusion_TrioB_C_500...	1016		XX	26|46	23|23	2.57E+11
	Full-Sibs						
	Half-Sibs						
US Asian-American							
1	Same-Individual Fusion_TrioB_M_50...	1017		XX	46|46	23|23	3.17E+34
	Father/Son						
1	Mother/Daughter Fusion_TrioB_C_500...	1016		XX	26|46	23|23	1.20E+09
	Full-Sibs						
	Half-Sibs						
US Caucasian							
1	Same-Individual Fusion_TrioB_M_50...	1017		XX	46|46	23|23	5.73E+31
	Father/Son						
1	Mother/Daughter Fusion_TrioB_C_500...	1016		XX	26|46	23|23	1.74E+09
	Full-Sibs						
	Half-Sibs						
US Hispanic-American							
1	Same-Individual Fusion_TrioB_M_50...	1017		XX	46|46	23|23	2.26E+32
	Father/Son						
1	Mother/Daughter Fusion_TrioB_C_500...	1016		XX	26|46	23|23	4.79E+08
	Full-Sibs						
	Half-Sibs						

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

Individual A: Fusion_TrioB_C_50(
Individual B: AF_500pg_1E02.hid

Marker	Individual A		Individual B		Parent/Child (LR)	Full-Sibs (LR)	Half-Sibs (LR)	Cousins (LR)
D3S1358	15	16	15	16	1.62584	2.38318	1.31292	1.15646
D1S1656	15	16.3	16	16.3	2.68068	1.59034	1.84034	1.42017
D2S441	13	15	10	13	5.42652	2.96326	3.21326	2.10663
D10S1248	14	16	14	15	0.91565	0.70783	0.95783	0.97891
D13S317	9	12	12		1.15207	0.82604	1.07604	1.03802
PENTA E	17	18	13	17	5.95238	3.22619	3.47619	2.23810
D16S539	9	10	9	11	1.32443	0.91222	1.16222	1.08111
D18S51	17	18	15	18	2.25225	1.37613	1.62613	1.31306
D2S1338	20	21	21	22	1.93483	1.21742	1.46742	1.23371
CSF1PO	11	14	10	11	1.22070	0.86035	1.11035	1.05518
PENTA D	12		12	14	4.31035	2.40517	2.65517	1.82759
TH01	6	7	7	8	0.63937	0.56968	0.81968	0.90984
vWA	17	18	16	18	2.17391	1.33696	1.58696	1.29348
D21S11	28	31	30	31	3.16456	1.83228	2.08228	1.54114
D7S820	11	12	10	12	2.76243	1.63122	1.88122	1.44061
D5S818	12	13	8	13	1.07759	0.78879	1.03879	1.01940
TPOX	8	10	8		1.49254	0.99627	1.24627	1.12313
D8S1179	13	14	13	15	1.20773	0.85386	1.10386	1.05193
D12S391	18.3	22	17	22	6.35647	3.42824	3.67824	2.33912
D19S433	13	14	13	14	2.24156	3.84595	1.62078	1.31039
FGA	23	24	21	23	1.40449	0.95225	1.20225	1.10112
D22S1045	11	16	11	15	1.76591	1.13296	1.38296	1.19148
				Product Score:	3.76E+06	1.46E+03	1.64E+04	3.36E+02

Kinship Analysis Settings

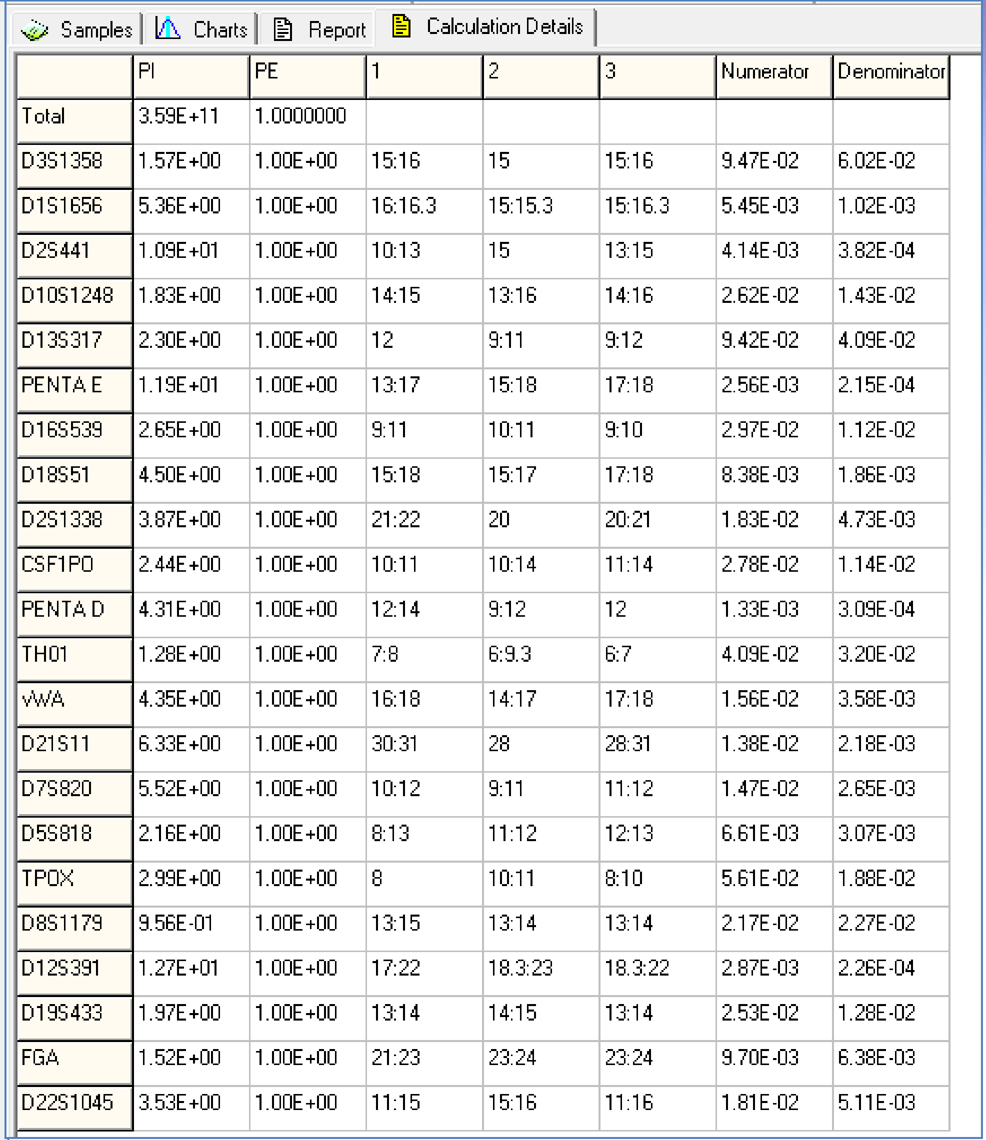
Relationships
- ☑ Unrelated
- ☑ Parent/Child
- ☑ Full-Sibs
- ☑ Half-Sibs
- ☐ Uncle-Nephew
- ☑ Cousins
- ☐ Grandparent-Grandchild

Report Content
- ● Likelihood Ratio
- ○ Probability
- ○ Both

Automated Pedigree Drawing – Inclusion Example

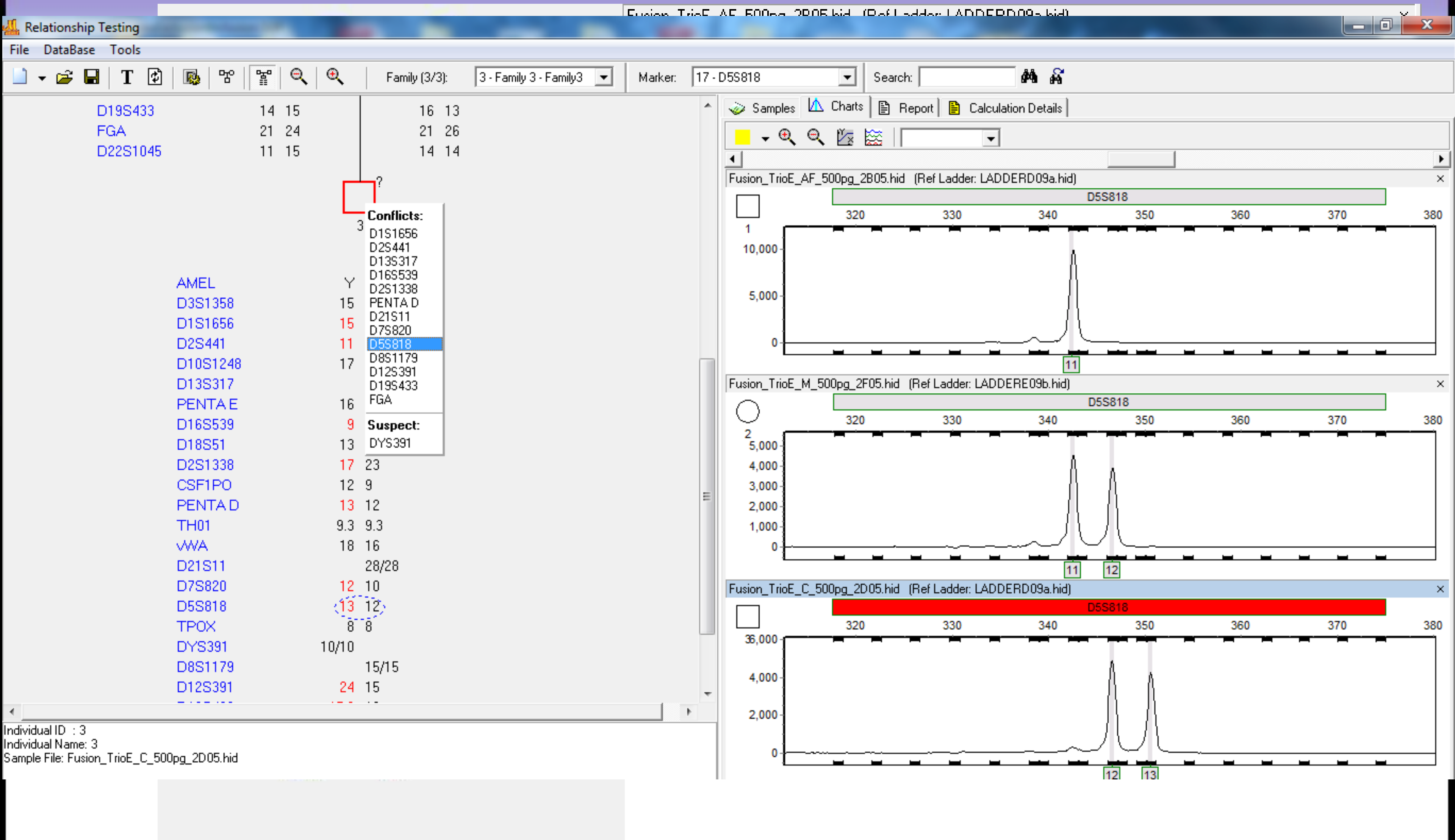
AMEL	Y	X	X	X
D3S1358	15	16	15	15
D1S1656	16	16.3	15.3	15
D2S441	10	13	15	15
D10S1248	15	14	13	16
D13S317	12	12	11	9
PENTA E	13	17	15	18
D16S539	11	9	11	10
D18S51	15	18	15	17
D2S1338	22	21	20	20
CSF1PO	10	11	10	14
PENTA D	14	12	9	12
TH01	8	7	9.3	6
vWA	16	18	14	17
D21S11	30	31	28	28
D7S820	10	12	9	11
D5S818	8	13	11	12
TPOX	8	8	11	10
DYS391	10	10	?	?
D8S1179	15	13	13	14
D12S391	17	22	23	18.3
D19S433	14	13	15	14
FGA	21	23	23	24
D22S1045	15	11	15	16

AMEL	X	X
D3S1358	16	15
D1S1656	16.3	15
D2S441	13	15
D10S1248	14	16
D13S317	12	9
PENTA E	17	18
D16S539	9	10
D18S51	18	17
D2S1338	21	20
CSF1PO	11	14
PENTA D	12	12
TH01	7	6
vWA	18	17
D21S11	31	28
D7S820	12	11
D5S818	13	12
TPOX	8	10
DYS391	?	?
D8S1179	13	14
D12S391	22	18.3
D19S433	13	14
FGA	23	24
D22S1045	11	16



	PI	PE	1	2	3	Numerator	Denominator
Total	3.59E+11	1.0000000					
D3S1358	1.57E+00	1.00E+00	15:16	15	15:16	9.47E-02	6.02E-02
D1S1656	5.36E+00	1.00E+00	16:16.3	15:15.3	15:16.3	5.45E-03	1.02E-03
D2S441	1.09E+01	1.00E+00	10:13	15	13:15	4.14E-03	3.82E-04
D10S1248	1.83E+00	1.00E+00	14:15	13:16	14:16	2.62E-02	1.43E-02
D13S317	2.30E+00	1.00E+00	12	9:11	9:12	9.42E-02	4.09E-02
PENTA E	1.19E+01	1.00E+00	13:17	15:18	17:18	2.56E-03	2.15E-04
D16S539	2.65E+00	1.00E+00	9:11	10:11	9:10	2.97E-02	1.12E-02
D18S51	4.50E+00	1.00E+00	15:18	15:17	17:18	8.38E-03	1.86E-03
D2S1338	3.87E+00	1.00E+00	21:22	20	20:21	1.83E-02	4.73E-03
CSF1PO	2.44E+00	1.00E+00	10:11	10:14	11:14	2.78E-02	1.14E-02
PENTA D	4.31E+00	1.00E+00	12:14	9:12	12	1.33E-03	3.09E-04
TH01	1.28E+00	1.00E+00	7:8	6:9.3	6:7	4.09E-02	3.20E-02
vWA	4.35E+00	1.00E+00	16:18	14:17	17:18	1.56E-02	3.58E-03
D21S11	6.33E+00	1.00E+00	30:31	28	28:31	1.38E-02	2.18E-03
D7S820	5.52E+00	1.00E+00	10:12	9:11	11:12	1.47E-02	2.65E-03
D5S818	2.16E+00	1.00E+00	8:13	11:12	12:13	6.61E-03	3.07E-03
TPOX	2.99E+00	1.00E+00	8	10:11	8:10	5.61E-02	1.88E-02
D8S1179	9.56E-01	1.00E+00	13:15	13:14	13:14	2.17E-02	2.27E-02
D12S391	1.27E+01	1.00E+00	17:22	18.3:23	18.3:22	2.87E-03	2.26E-04
D19S433	1.97E+00	1.00E+00	13:14	14:15	13:14	2.53E-02	1.28E-02
FGA	1.52E+00	1.00E+00	21:23	23:24	23:24	9.70E-03	6.38E-03
D22S1045	3.53E+00	1.00E+00	11:15	15:16	11:16	1.81E-02	5.11E-03

Automated Pedigree Drawing – Exclusion Example



Fusion_TrioE_AF_500pg_2B05.hid (Ref Ladder: LADDERD09a.hid)

Family (3/3): 3 - Family 3 - Family3

Marker: 17 - D5S818

Search:

D19S433	14 15	16 13
FGA	21 24	21 26
D22S1045	11 15	14 14

Conflicts:
- D1S1656
- D2S441
- D13S317
- D16S539
- D2S1338
- PENTA D
- D21S11
- D7S820
- D5S818
- D8S1179
- D12S391
- D19S433
- FGA

Suspect:
- DYS391

AMEL	Y	
D3S1358	15	
D1S1656	15	
D2S441	11	
D10S1248	17	
D13S317		
PENTA E	16	
D16S539	9	
D18S51	13	
D2S1338	17	23
CSF1PO	12	9
PENTA D	13	12
TH01	9.3	9.3
vWA	18	16
D21S11		28/28
D7S820	12	10
D5S818	13	12
TPOX	8	8
DYS391	10/10	
D8S1179		15/15
D12S391	24	15

Individual ID : 3
Individual Name: 3
Sample File: Fusion_TrioE_C_500pg_2D05.hid

Fusion_TrioE_AF_500pg_2B05.hid (Ref Ladder: LADDERD09a.hid)

Fusion_TrioE_M_500pg_2F05.hid (Ref Ladder: LADDERE09b.hid)

Fusion_TrioE_C_500pg_2D05.hid (Ref Ladder: LADDERD09a.hid)

Searchable Database - Relationship Testing Summary

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

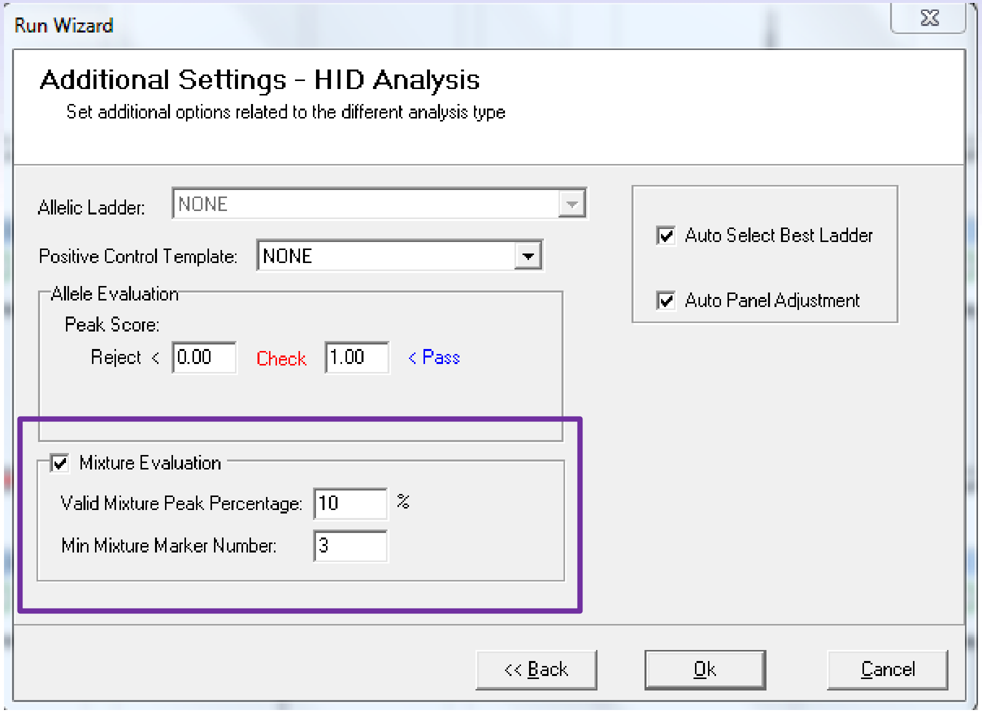
- Kinship Analysis Across Three Generations

- 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



Run Wizard

Additional Settings - HID Analysis

Set additional options related to the different analysis type

Allelic Ladder: NONE

Positive Control Template: NONE

Allele Evaluation

Peak Score:

Reject < 0.00 Check 1.00 < Pass

☑ Mixture Evaluation

Valid Mixture Peak Percentage: 10 %

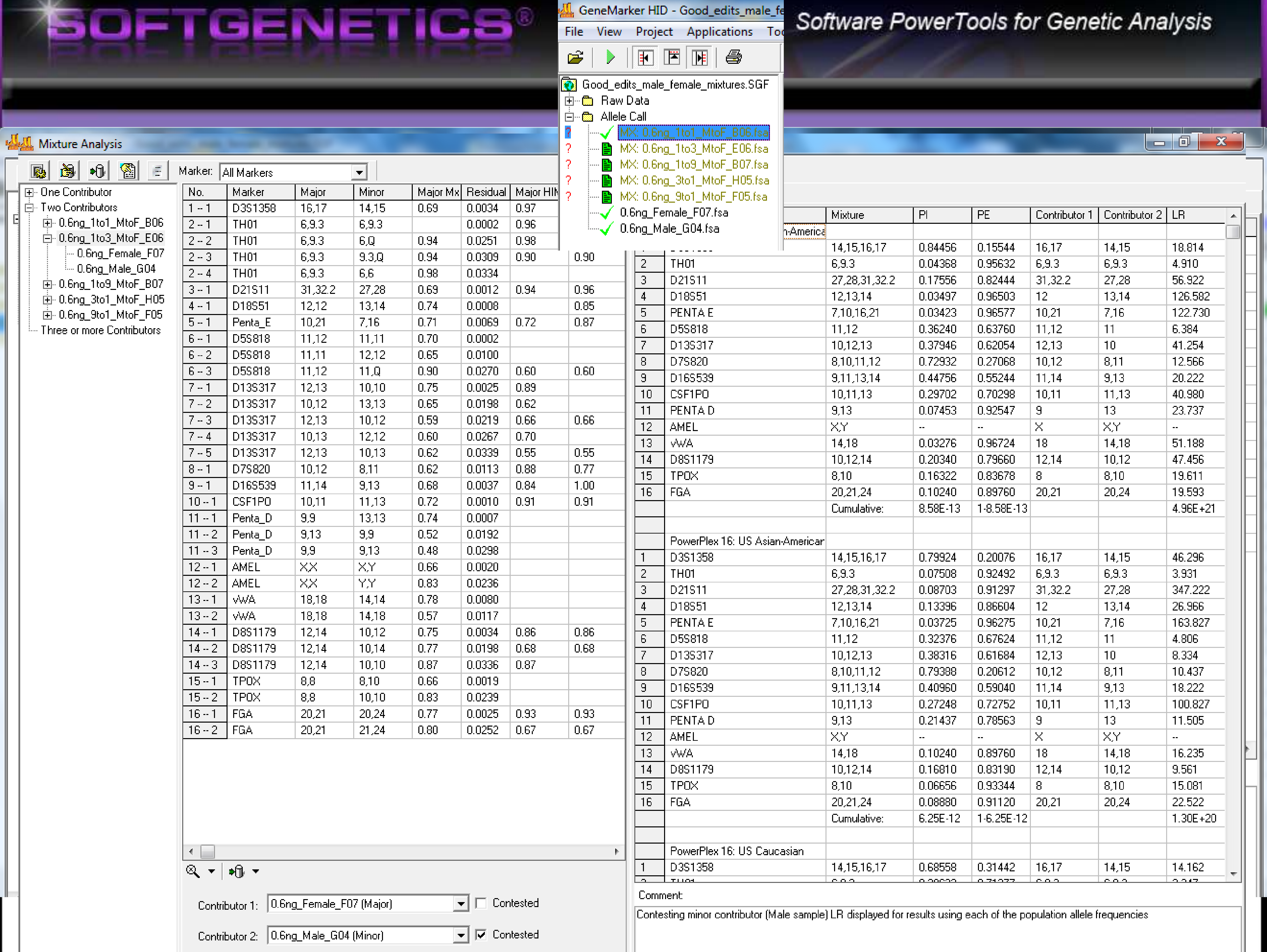
Min Mixture Marker Number: 3

☑ Auto Select Best Ladder

☑ Auto Panel Adjustment

SOFTGENETICS®

Software PowerTools for Genetic Analysis



Marker: All Markers

No.	Marker	Major	Minor	Major Mx	Residual	Major HIM	
1 -- 1	D3S1358	16,17	14,15	0.69	0.0034	0.97	
2 -- 1	TH01	6,9.3	6,9.3		0.0002	0.96	
2 -- 2	TH01	6,9.3	6,Q	0.94	0.0251	0.98	
2 -- 3	TH01	6,9.3	9.3,Q	0.94	0.0309	0.90	0.90
2 -- 4	TH01	6,9.3	6,6	0.98	0.0334		
3 -- 1	D21S11	31,32.2	27,28	0.69	0.0012	0.94	0.96
4 -- 1	D18S51	12,12	13,14	0.74	0.0008		0.85
5 -- 1	Penta_E	10,21	7,16	0.71	0.0069	0.72	0.87
6 -- 1	D5S818	11,12	11,11	0.70	0.0002		
6 -- 2	D5S818	11,11	12,12	0.65	0.0100		
6 -- 3	D5S818	11,12	11,Q	0.90	0.0270	0.60	0.60
7 -- 1	D13S317	12,13	10,10	0.75	0.0025	0.89	
7 -- 2	D13S317	10,12	13,13	0.65	0.0198	0.62	
7 -- 3	D13S317	12,13	10,12	0.59	0.0219	0.66	0.66
7 -- 4	D13S317	10,13	12,12	0.60	0.0267	0.70	
7 -- 5	D13S317	12,13	10,13	0.62	0.0339	0.55	0.55
8 -- 1	D7S820	10,12	8,11	0.62	0.0113	0.88	0.77
9 -- 1	D16S539	11,14	9,13	0.68	0.0037	0.84	1.00
10 -- 1	CSF1PO	10,11	11,13	0.72	0.0010	0.91	0.91
11 -- 1	Penta_D	9,9	13,13	0.74	0.0007		
11 -- 2	Penta_D	9,13	9,9	0.52	0.0192		
11 -- 3	Penta_D	9,9	9,13	0.48	0.0298		
12 -- 1	AMEL	X,X	X,Y	0.66	0.0020		
12 -- 2	AMEL	X,X	Y,Y	0.83	0.0236		
13 -- 1	vWA	18,18	14,14	0.78	0.0080		
13 -- 2	vWA	18,18	14,18	0.57	0.0117		
14 -- 1	D8S1179	12,14	10,12	0.75	0.0034	0.86	0.86
14 -- 2	D8S1179	12,14	10,14	0.77	0.0198	0.68	0.68
14 -- 3	D8S1179	12,14	10,10	0.87	0.0336	0.87	
15 -- 1	TPOX	8,8	8,10	0.66	0.0019		
15 -- 2	TPOX	8,8	10,10	0.83	0.0239		
16 -- 1	FGA	20,21	20,24	0.77	0.0025	0.93	0.93
16 -- 2	FGA	20,21	21,24	0.80	0.0252	0.67	0.67

Contributor 1: 0.6ng_Female_F07 (Major) ☐ Contested

Contributor 2: 0.6ng_Male_G04 (Minor) ☑ Contested

		Mixture	PI	PE	Contributor 1	Contributor 2	LR
	n-America						
		14,15,16,17	0.84456	0.15544	16,17	14,15	18.814
2	TH01	6,9.3	0.04368	0.95632	6,9.3	6,9.3	4.910
3	D21S11	27,28,31,32.2	0.17556	0.82444	31,32.2	27,28	56.922
4	D18S51	12,13,14	0.03497	0.96503	12	13,14	126.582
5	PENTA E	7,10,16,21	0.03423	0.96577	10,21	7,16	122.730
6	D5S818	11,12	0.36240	0.63760	11,12	11	6.384
7	D13S317	10,12,13	0.37946	0.62054	12,13	10	41.254
8	D7S820	8,10,11,12	0.72932	0.27068	10,12	8,11	12.566
9	D16S539	9,11,13,14	0.44756	0.55244	11,14	9,13	20.222
10	CSF1PO	10,11,13	0.29702	0.70298	10,11	11,13	40.980
11	PENTA D	9,13	0.07453	0.92547	9	13	23.737
12	AMEL	X,Y	--	--	X	X,Y	--
13	vWA	14,18	0.03276	0.96724	18	14,18	51.188
14	D8S1179	10,12,14	0.20340	0.79660	12,14	10,12	47.456
15	TPOX	8,10	0.16322	0.83678	8	8,10	19.611
16	FGA	20,21,24	0.10240	0.89760	20,21	20,24	19.593
		Cumulative:	8.58E-13	1-8.58E-13			4.96E+21
	PowerPlex 16: US Asian-Americar						
1	D3S1358	14,15,16,17	0.79924	0.20076	16,17	14,15	46.296
2	TH01	6,9.3	0.07508	0.92492	6,9.3	6,9.3	3.931
3	D21S11	27,28,31,32.2	0.08703	0.91297	31,32.2	27,28	347.222
4	D18S51	12,13,14	0.13396	0.86604	12	13,14	26.966
5	PENTA E	7,10,16,21	0.03725	0.96275	10,21	7,16	163.827
6	D5S818	11,12	0.32376	0.67624	11,12	11	4.806
7	D13S317	10,12,13	0.38316	0.61684	12,13	10	8.334
8	D7S820	8,10,11,12	0.79388	0.20612	10,12	8,11	10.437
9	D16S539	9,11,13,14	0.40960	0.59040	11,14	9,13	18.222
10	CSF1PO	10,11,13	0.27248	0.72752	10,11	11,13	100.827
11	PENTA D	9,13	0.21437	0.78563	9	13	11.505
12	AMEL	X,Y	--	--	X	X,Y	--
13	vWA	14,18	0.10240	0.89760	18	14,18	16.235
14	D8S1179	10,12,14	0.16810	0.83190	12,14	10,12	9.561
15	TPOX	8,10	0.06656	0.93344	8	8,10	15.081
16	FGA	20,21,24	0.08880	0.91120	20,21	20,24	22.522
		Cumulative:	6.25E-12	1-6.25E-12			1.30E+20
	PowerPlex 16: US Caucasian						
1	D3S1358	14,15,16,17	0.68558	0.31442	16,17	14,15	14.162

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

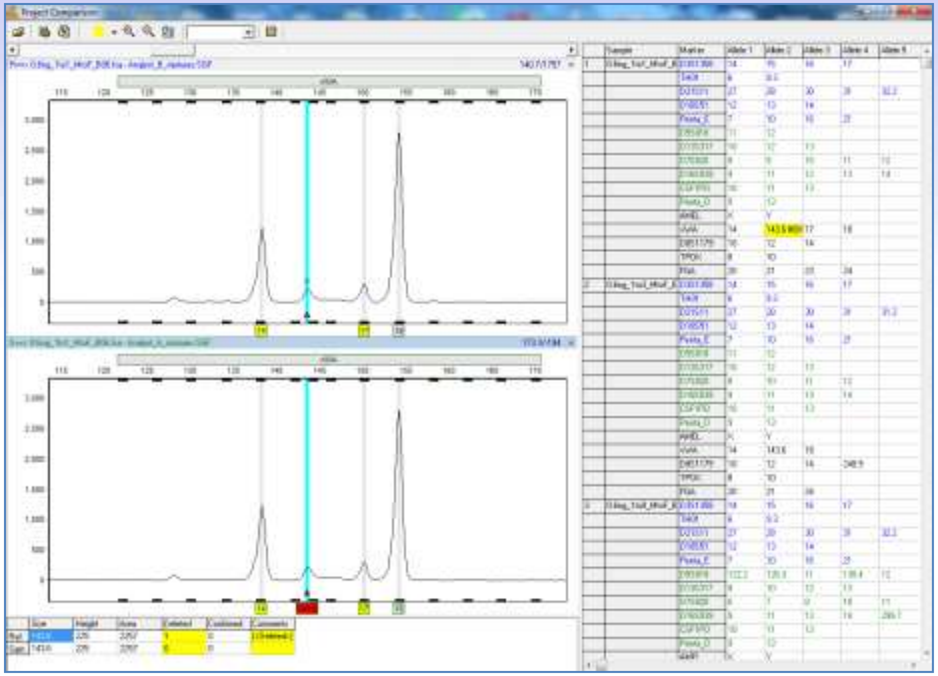
- Automates Repetitive Calculations

- Calculates PI, PE, LR, RMNE

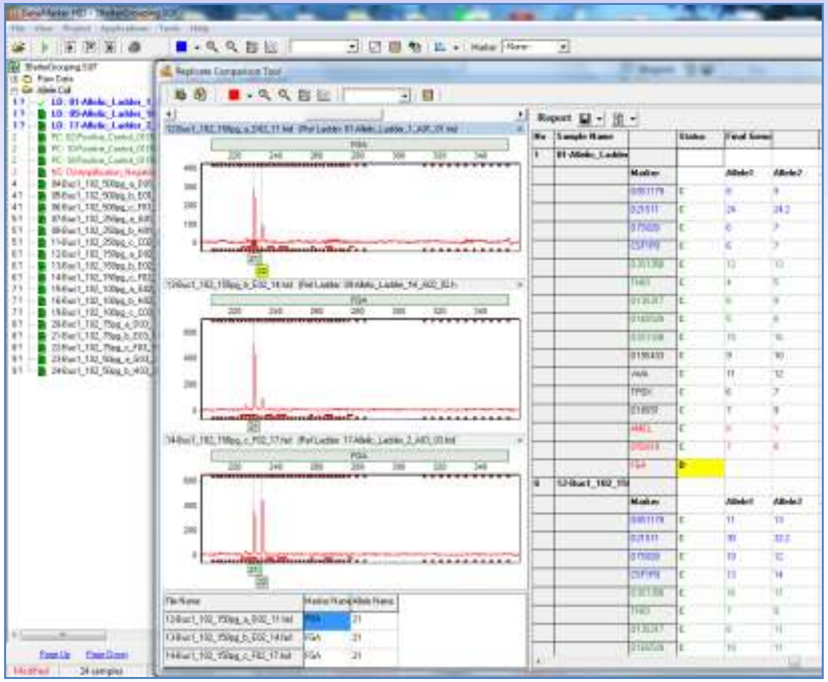
- Database search for contributor; with or without a reference sample

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!

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