



# **Evaluation of GeneMapper<sup>®</sup> *ID-X* and GeneMarker<sup>®</sup> HID for use at the NYC OCME**

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September 29, 2011

# New York City Office of the Chief Medical Examiner



# Background

- DNA Analysis
  - Complicated, multi-step process
  - Will be focusing only on data analysis
- Evaluation a new software systems
  - 3500xl data compatibility
  - Implementation of a new LIMS
  - Reduction in analysis time
  - Availability of mixture assistant tools



# Background

- Data Analysis Software
  - Displays generated data
  - Allows for comparison
  - Tangible product for court testimony
- Sample Editing
  - Off-ladder allele
  - Drop-in/Drop-out
  - Pull-up
  - Spikes
  - Peak shoulders
  - Dye artifacts



# Topics

- Introduction to the Software
- Analysis and Edit Procedures
- Sample Concordance
- Ladder Assessment
- Pull-up Correction
- Stutter Filters
- Mixture Assistant Tools Examination
- Result Export Capabilities



# Introduction to the Systems

- GeneMapper<sup>®</sup> *ID-X* v1.1
  - Applied Biosystems- Foster City, California
  - GeneMapper<sup>®</sup> *ID* v.3.2.1 based
  
- GeneMarker<sup>®</sup> HID v1.95
  - SoftGenetics- State College, Pennsylvania
  - GeneMarker<sup>®</sup> based



# Introduction to the Systems

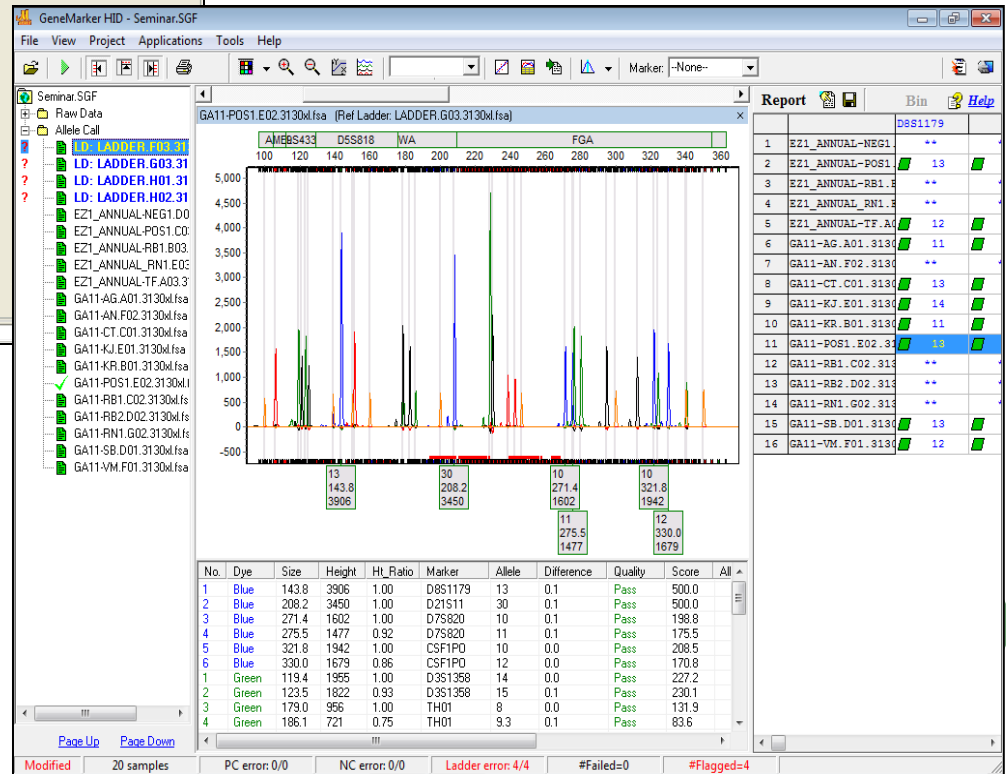
GeneMapper® ID-X - Ron presentation - gmidx Is Logged In Database fsc\_nest4

File Edit Analysis View Tools Admin Help

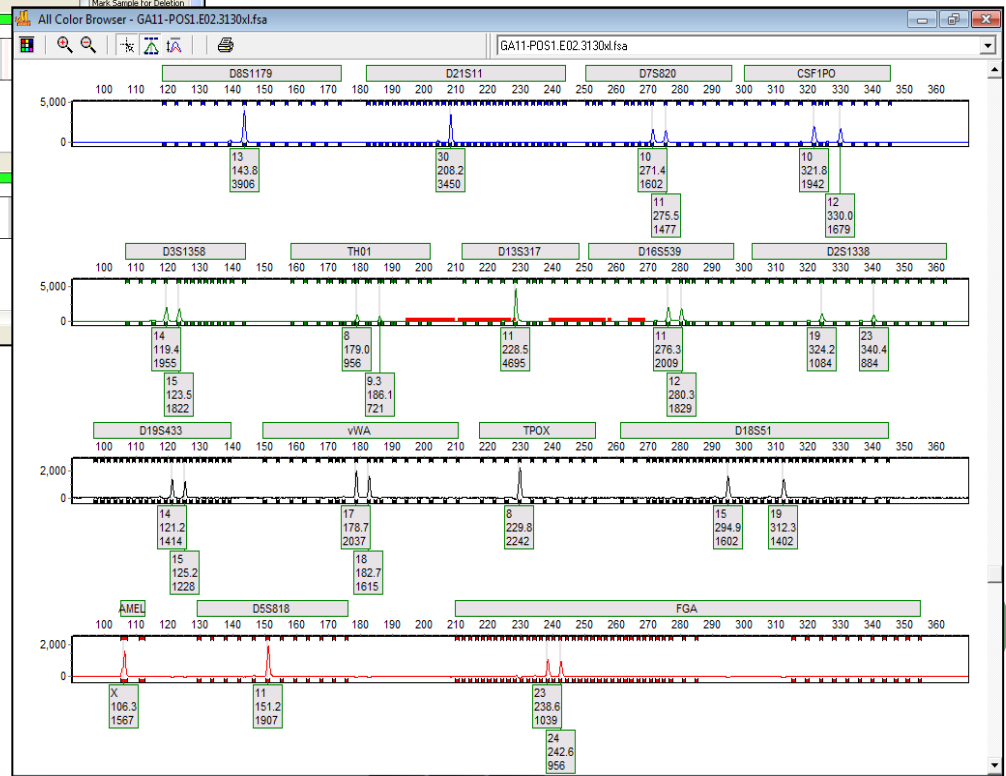
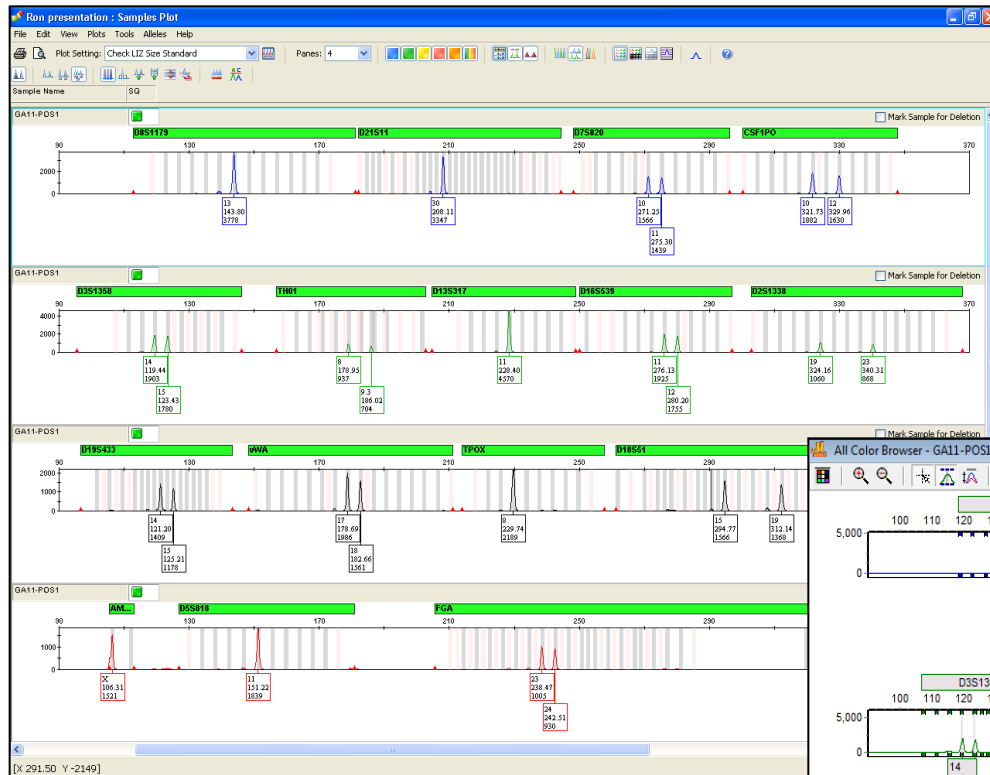
Table Settings: 31XX Data Analysis

Status	Sample Name	Sample Type	Analysis Method	Panel	Size Standard	Custom Control	AR1M	SOS	SG	SSPK	MX	OMR	COG
1	EZ1_ANNUAL-NE	Sample	RonMixture.TAP	Identifier_v1X	CE_G5_HD_G5500	None							
2	EZ1_ANNUAL-RC	Sample	RonMixture.TAP	Identifier_v1X	CE_G5_HD_G5500	None							
3	EZ1_ANNUAL-RE	Sample	RonMixture.TAP	Identifier_v1X	CE_G5_HD_G5500	None							
4	EZ1_ANNUAL-TF	Sample	RonMixture.TAP	Identifier_v1X	CE_G5_HD_G5500	None							
5	EZ1_ANNUAL-TR	Sample	RonMixture.TAP	Identifier_v1X	CE_G5_HD_G5500	None							
6	GA11-AG	Sample	RonMixture.TAP	Identifier_v1X	CE_G5_HD_G5500	None							
7	GA11-AN	Sample	RonMixture.TAP	Identifier_v1X	CE_G5_HD_G5500	None							
8	GA11-CT	Sample	RonMixture.TAP	Identifier_v1X	CE_G5_HD_G5500	None							
9	GA11-KJ	Sample	RonMixture.TAP	Identifier_v1X	CE_G5_HD_G5500	None							
10	GA11-KR	Sample	RonMixture.TAP	Identifier_v1X	CE_G5_HD_G5500	None							
11	GA11-POST1	Sample	RonMixture.TAP	Identifier_v1X	CE_G5_HD_G5500	None							
12	GA11-RB1	Sample	RonMixture.TAP	Identifier_v1X	CE_G5_HD_G5500	None							
13	GA11-RB2	Sample	RonMixture.TAP	Identifier_v1X	CE_G5_HD_G5500	None							
14	GA11-RN1	Sample	RonMixture.TAP	Identifier_v1X	CE_G5_HD_G5500	None							
15	GA11-SB	Sample	RonMixture.TAP	Identifier_v1X	CE_G5_HD_G5500	None							
16	GA11-VM	Sample	RonMixture.TAP	Identifier_v1X	CE_G5_HD_G5500	None							
17	LADDER	Allelic Ladder	RonMixture.TAP	Identifier_v1X	CE_G5_HD_G5500	None							
18	LADDER	Allelic Ladder	RonMixture.TAP	Identifier_v1X	CE_G5_HD_G5500	None							
19	LADDER	Allelic Ladder	RonMixture.TAP	Identifier_v1X	CE_G5_HD_G5500	None							
20	LADDER	Allelic Ladder	RonMixture.TAP	Identifier_v1X	CE_G5_HD_G5500	None							

Analysis Completed.

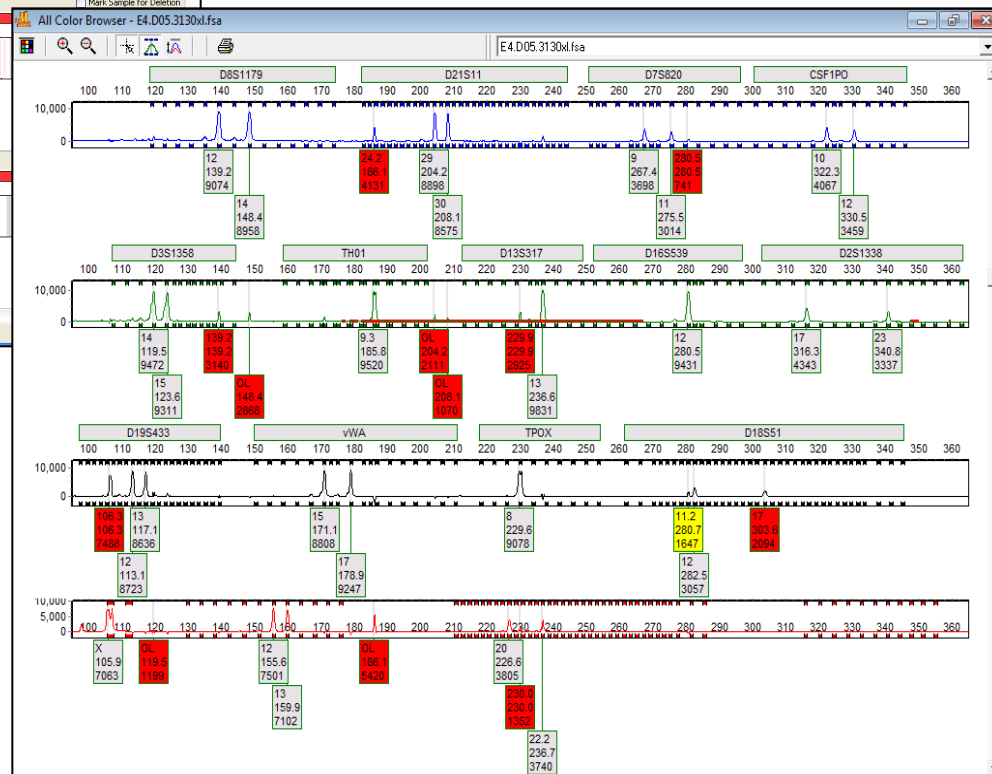
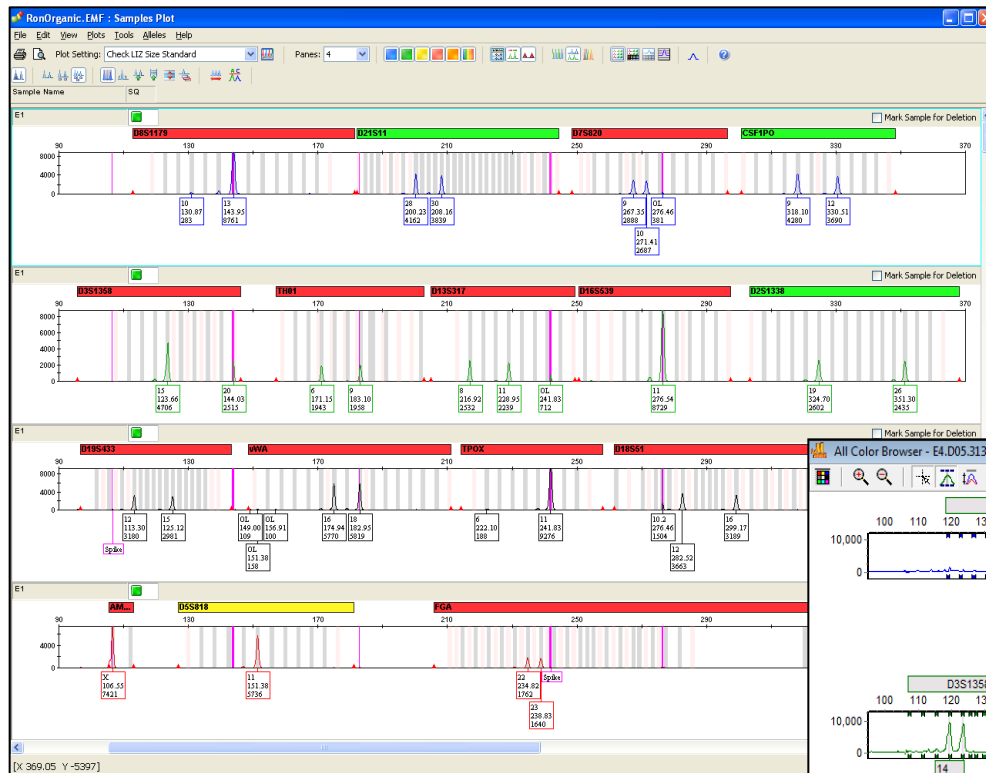


# Introduction to the Systems





# Introduction to the Systems



# Analysis of Raw Data

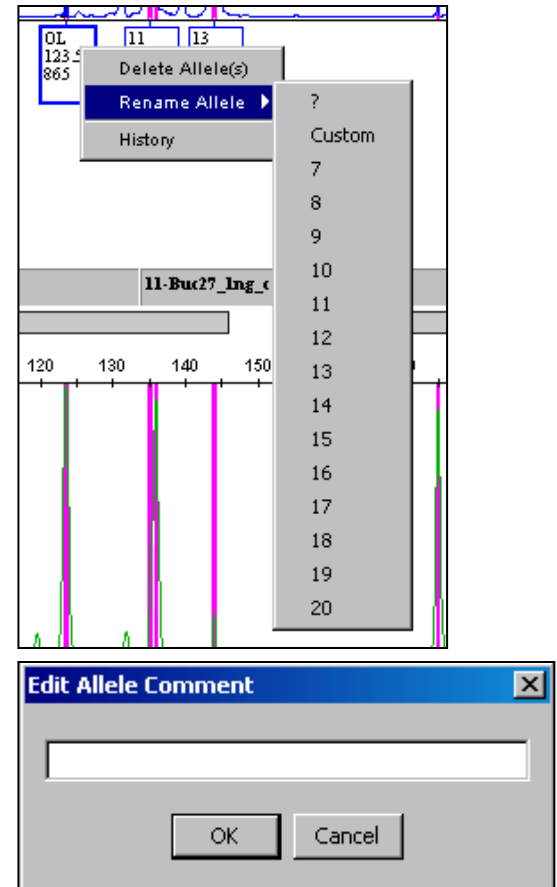
- Data is loaded into both programs in a similar fashion
- GeneMarker<sup>®</sup> HID is faster than GeneMapper<sup>®</sup> *ID-X*
- Sample type designation differs between programs
  - GeneMapper<sup>®</sup> *ID-X*
    - Sample types must be designated by the user prior to analysis
  - GeneMarker<sup>®</sup> HID
    - Identifiers automatically select sample types
    - The OCME used:
      - “LADDER” for the allelic ladders and “PE” for positive controls



# Sample Editing

- GeneMapper® *ID*

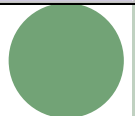
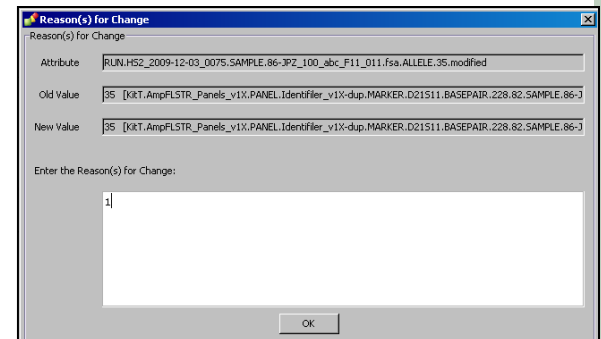
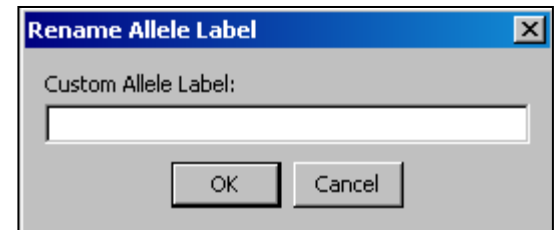
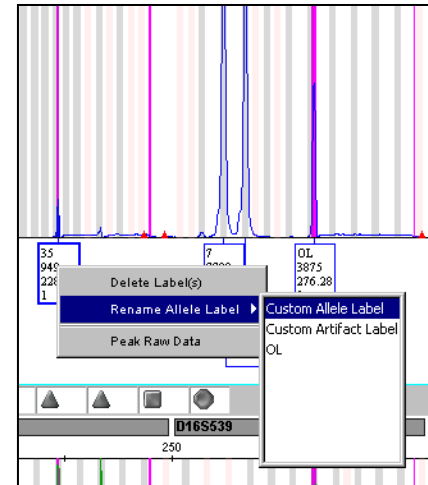
- Initial Analysis Requirements
  - Any allele call can be reassigned
  - Edit code must be assigned
- Technical Review Requirements
  - Edit code must be typed prior to deletion



# Sample Editing

- GeneMapper® *ID-X*

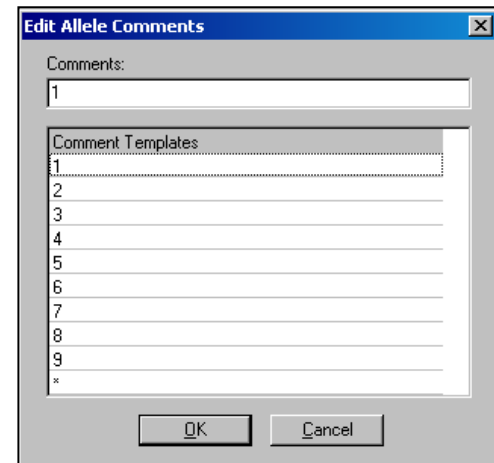
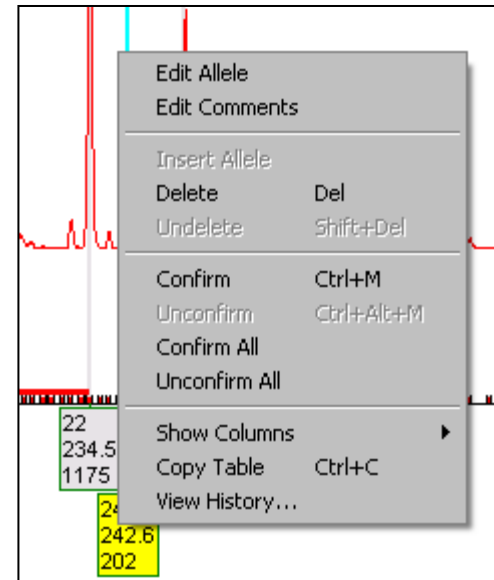
- Initial Analysis Requirements
  - Any allele call can be reassigned
  - Edit code must be assigned
- Technical Review Requirements
  - Edit code must be typed prior to deletion



# Sample Editing

- GeneMarker<sup>®</sup> HID

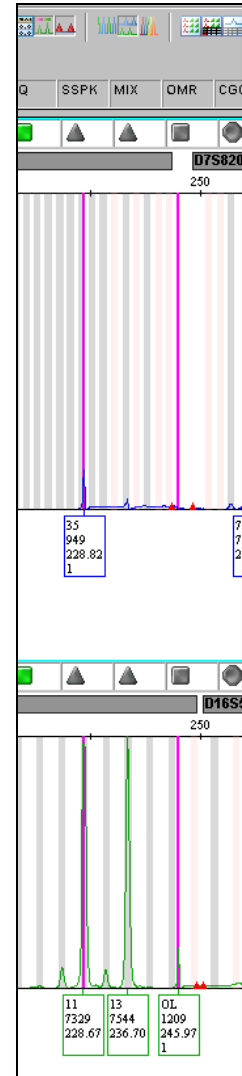
- Initial Analysis Requirements
  - No allele call reassignment
  - Edit code is not necessary
- Technical Review Requirements
  - Call can be deleted without analyst input



# Oversaturated Peaks

## GeneMapper® ID

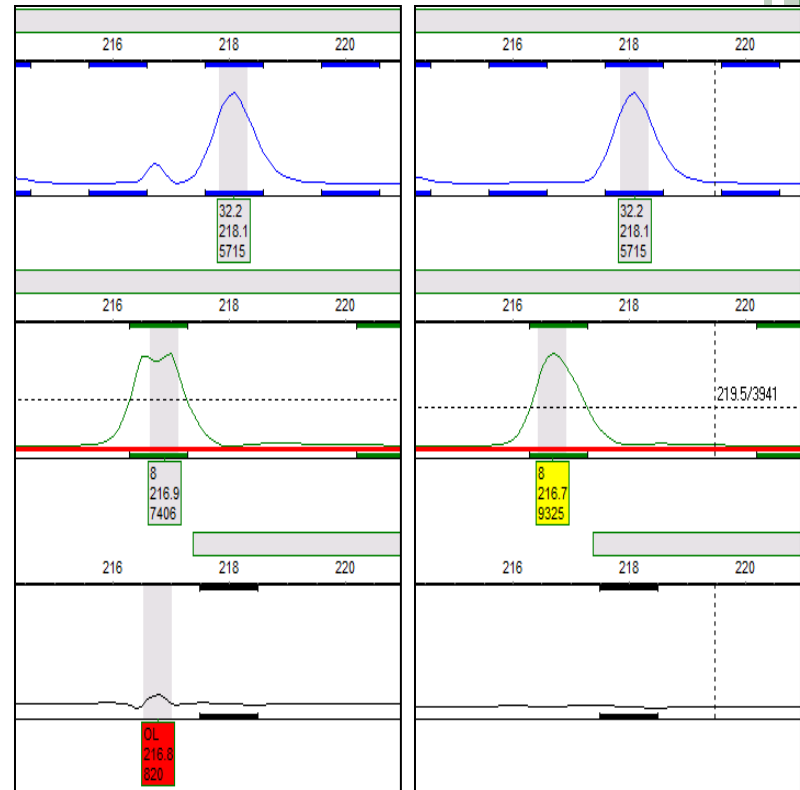
- Camera oversaturation is displayed by the presence of a pink indicator line
  - This line will cross all color channels
  - Provides possible reason for OL allele calls
  - Warns analysts when peak height ratios might be misrepresented
- GeneMapper® ID-X retains this feature



# Oversaturated Peaks

## GeneMarker<sup>®</sup> HID

- Saturated Peak Repair
  - Identification of oversaturation
    - Removes pull-up peaks
    - Augments offending peak
  - In/Out of bin peaks removed
  - Some pull-up peaks remain
  - All allele calls are maintained
    - n= 30 single source samples



# Oversaturated Peaks

## GeneMarker<sup>®</sup> HID

- Goal- determine if repair affects peak height ratios
  - n = 6 single source samples, 100 pg, ID31
  - Normal injection with option enabled
  - Low injection without the option enabled

<b>No Peaks Repaired</b>	<b>Average Ratio Difference</b>	0.03
n=35	<b>Standard Deviation</b>	0.05
<b>One Peak Repaired</b>	<b>Average Ratio Difference</b>	0.12
n=22	<b>Standard Deviation</b>	0.12
<b>Both Peaks Repaired</b>	<b>Average Ratio Difference</b>	0.06
n=16	<b>Standard Deviation</b>	0.05

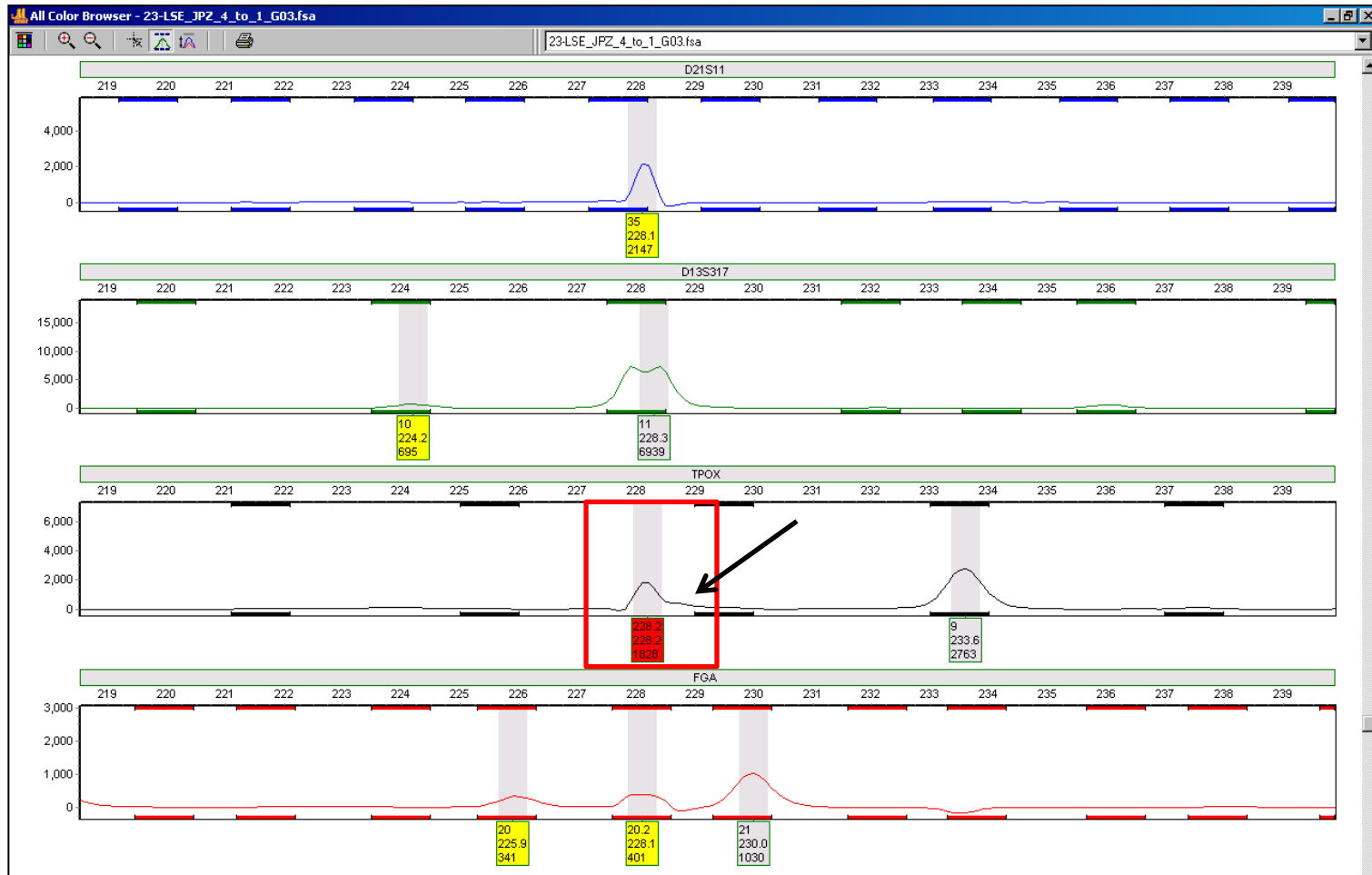
- 10 of 82 loci, the major peak flipped with the repair option – 12.1%
- 4 of 82 loci, the peak height ratio was decreased past 0.50 – 4.8%

**Saturated repair does not significantly affect peak height ratios**

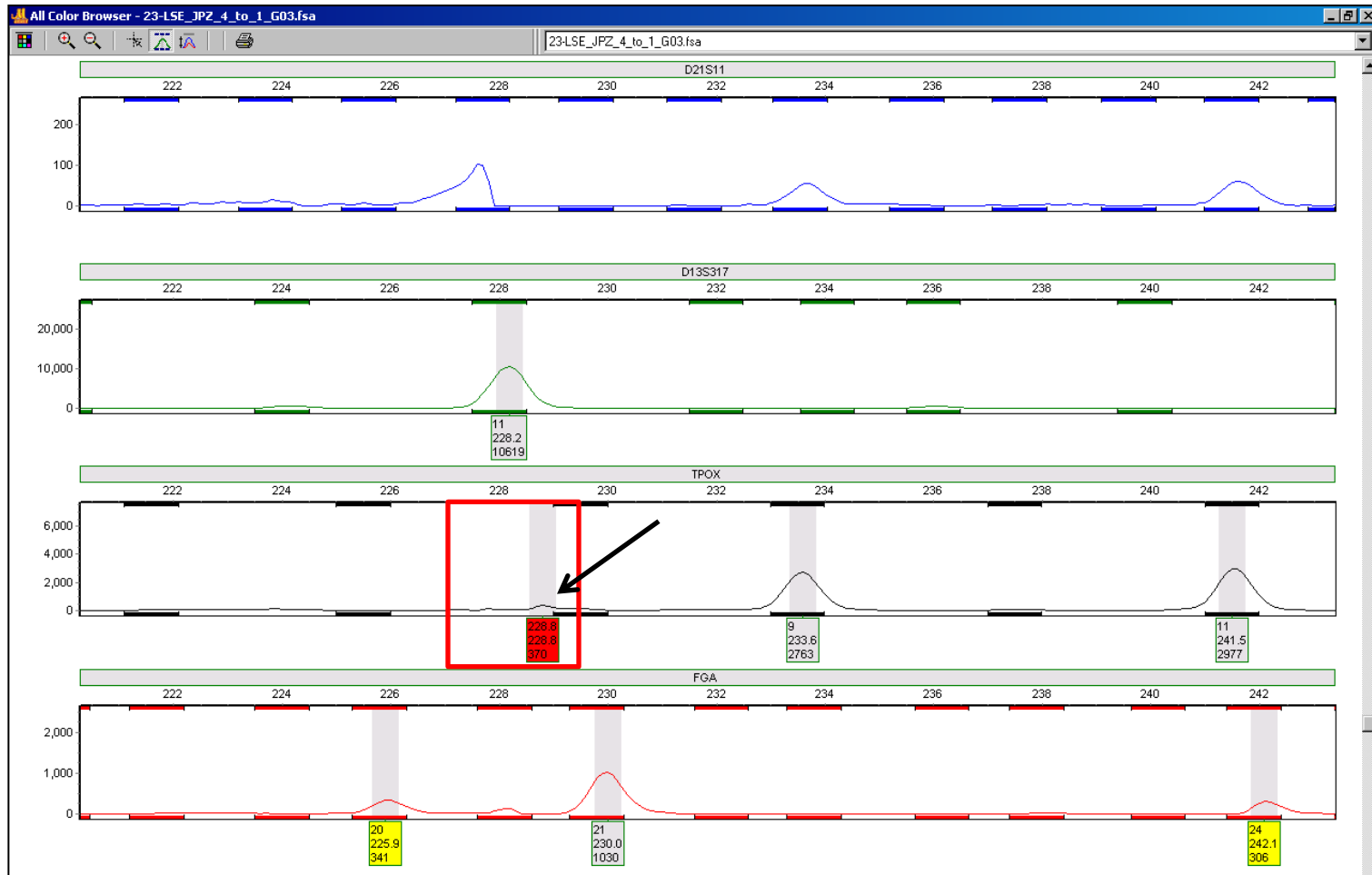




# Movement of pull-up edits



# Movement of pull-up edits



# Movement of pull-up edits

- Goal- assess frequency of occurrence
  - n = 49 samples, 253 total repaired peaks examined
  - 13 peaks left a residual peak after repair- 5.1%
  - The residual peak moved out of a bin 3 times
  - The residual peak never moved into a bin

## Difference Between Peaks (bp)

<b>Average Before Repair</b>	0.1
<b>Standard Deviation</b>	0.1
<b>Average After Repair</b>	0.6
<b>Standard Deviation</b>	0.1



# Change in Peak Heights

- Low Copy Number typing relies on peak height levels
- Original protocols were designed using old systems
  - AB GeneScan<sup>®</sup>/Genotyper<sup>®</sup>
- Goal- determine how peak height levels compared
  - Average height levels were averaged across samples
  - Compared with GeneScan<sup>®</sup>/Genotyper<sup>®</sup> values
  - n = 24 single source samples

System	% change (RFUs)
GeneMapper <sup>®</sup> ID v3.2.1	-3.00%
GeneMapper <sup>®</sup> ID-X v1.1	-2.80%
GeneMarker <sup>®</sup> HID v1.95	-0.38%



# Sample Edits

- Goal- determine optimal system and parameters for reduction of inaccurate allele calls and analysis time
  - n = 24 (n = 12, 50pg; n = 12 100pg) ID31

	<u>50pg samples</u>		<u>100pg samples</u>	
	GMID-X # of edits	GeneMarker HID (w/o repair) # of edits	GMID-X # of edits	GeneMarker HID (w/o repair) # of edits
<b>Average</b>	5.4	8.8	17.5	23.4
<b>Standard Deviation</b>	3.1	3.5	3.8	5.6
<b>Differences in edits</b>	<b>3.4</b>		<b>5.9</b>	
<b>% Difference</b>	<b>63.1</b>		<b>33.8</b>	

	<u>50pg samples</u>		<u>100pg samples</u>	
	GMID-X # of edits	GeneMarker HID (w/ repair) # of edits	GMID-X # of edits	GeneMarker HID (w/ repair) # of edits
<b>Average</b>	5.4	4.4	17.5	4.8
<b>Standard Deviation</b>	3.1	1.8	3.8	2.8
<b>Differences in edits</b>	<b>-1.0</b>		<b>-12.7</b>	
<b>% Difference</b>	<b>-18.5</b>		<b>-72.9</b>	



# Allele Calls in Mixtures

- Goal- test peak calling algorithms at extreme input
  - Allele calls in mixtures were analyzed
  - Compared with GeneScan<sup>®</sup>/Genotyper<sup>®</sup> values
  - n = 22, 25 pg 1:5:5, 5:1:1 mixtures, ID31
- GeneMapper<sup>®</sup> *ID-X*
  - Gained 1 peak that was drop-out in GeneScan<sup>®</sup>/Genotyper<sup>®</sup>
- GeneMarker<sup>®</sup> HID
  - Gained 3 peaks that were previously drop-out
  - Lost 2 peaks that were previously drop-in
  - Lost 1 accurate allele call- new drop-out

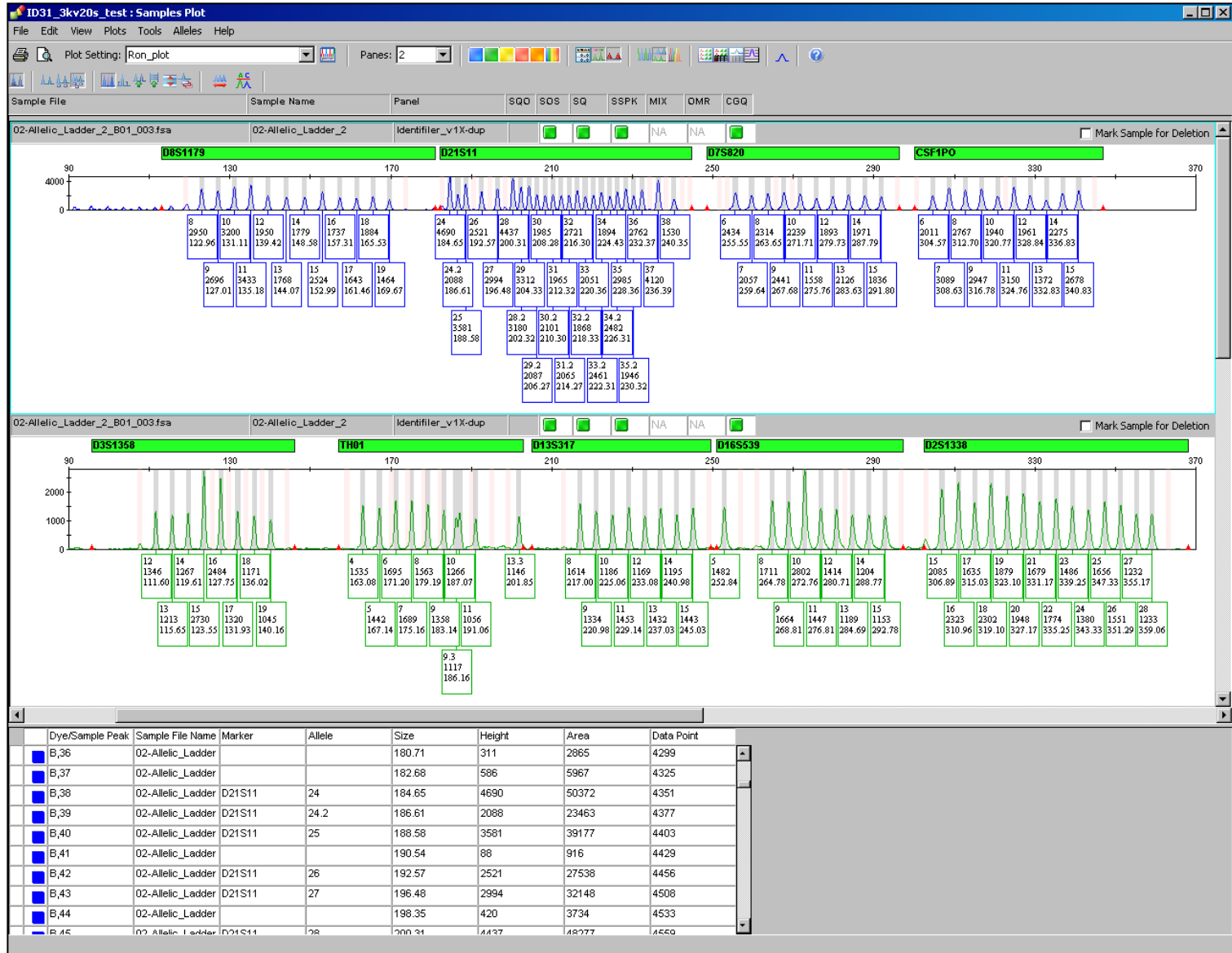


# Allelic Ladders

- Low Copy Number typing will stress any system
- Ladder samples are a necessary control for DNA typing
- The systems deal with ladders in different manners
- GeneMapper<sup>®</sup>*ID*
  - All ladders examined passed requirements
- GeneMapper<sup>®</sup>*ID-X*
  - All ladders examined passed requirements



# GeneMapper® ID-X Ladders





# Allelic Ladders

- Low Copy Number typing will stress any system
- Ladder samples are a necessary control for DNA typing
- The systems deal with ladders in different manners
  
- GeneMapper<sup>®</sup> *ID*
  - All ladders examined passed requirements
  
- GeneMapper<sup>®</sup> *ID-X*
  - All ladders examined passed requirements
  
- GeneMarker<sup>®</sup> HID
  - Several issues arose with the initial version of the software

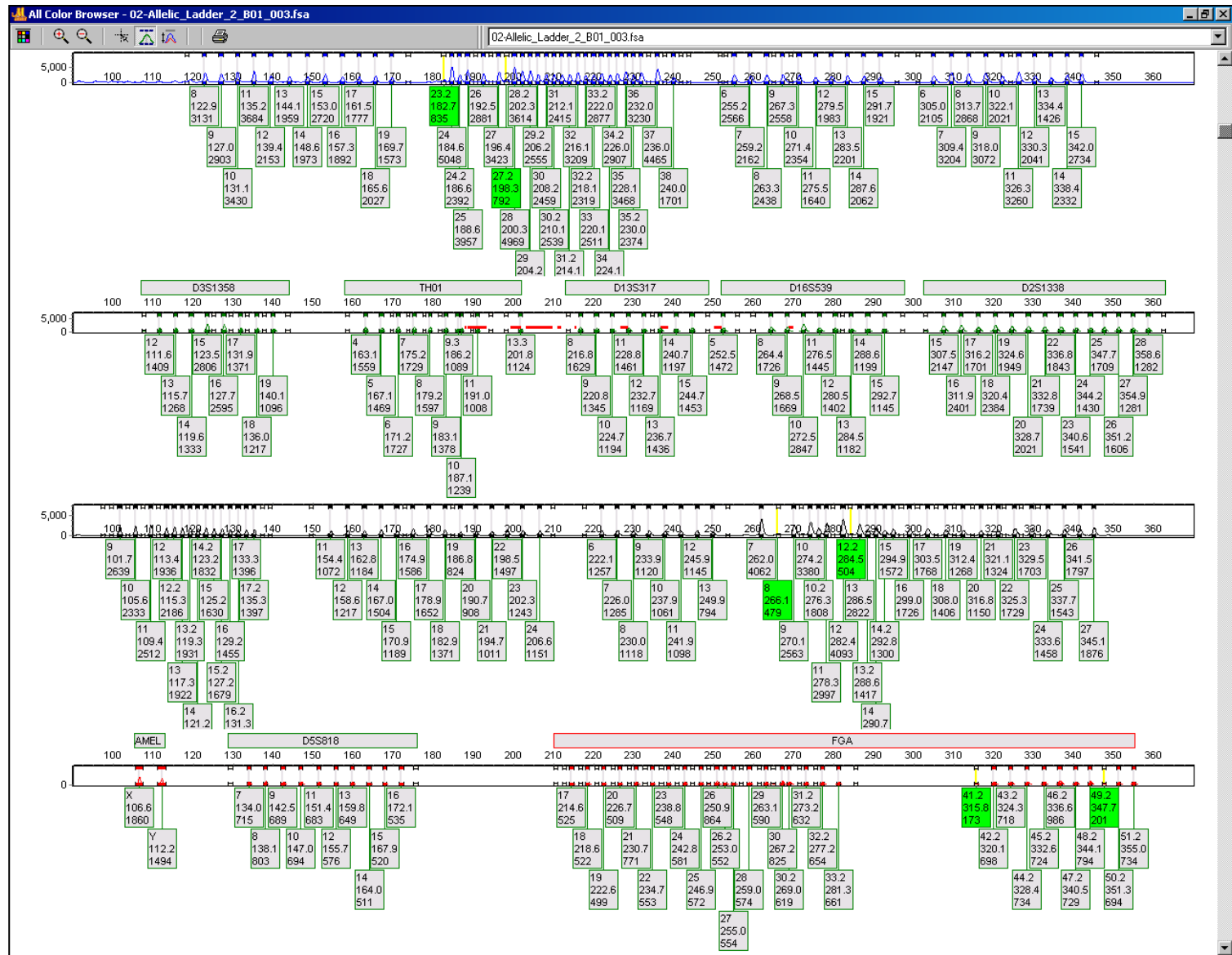


# Allelic Ladders

- GeneMarker<sup>®</sup> HID
- ID31 normal injection parameter
  - All published peaks are present
  - Peaks are present in virtual allele bins



# GeneMarker® HID Ladders

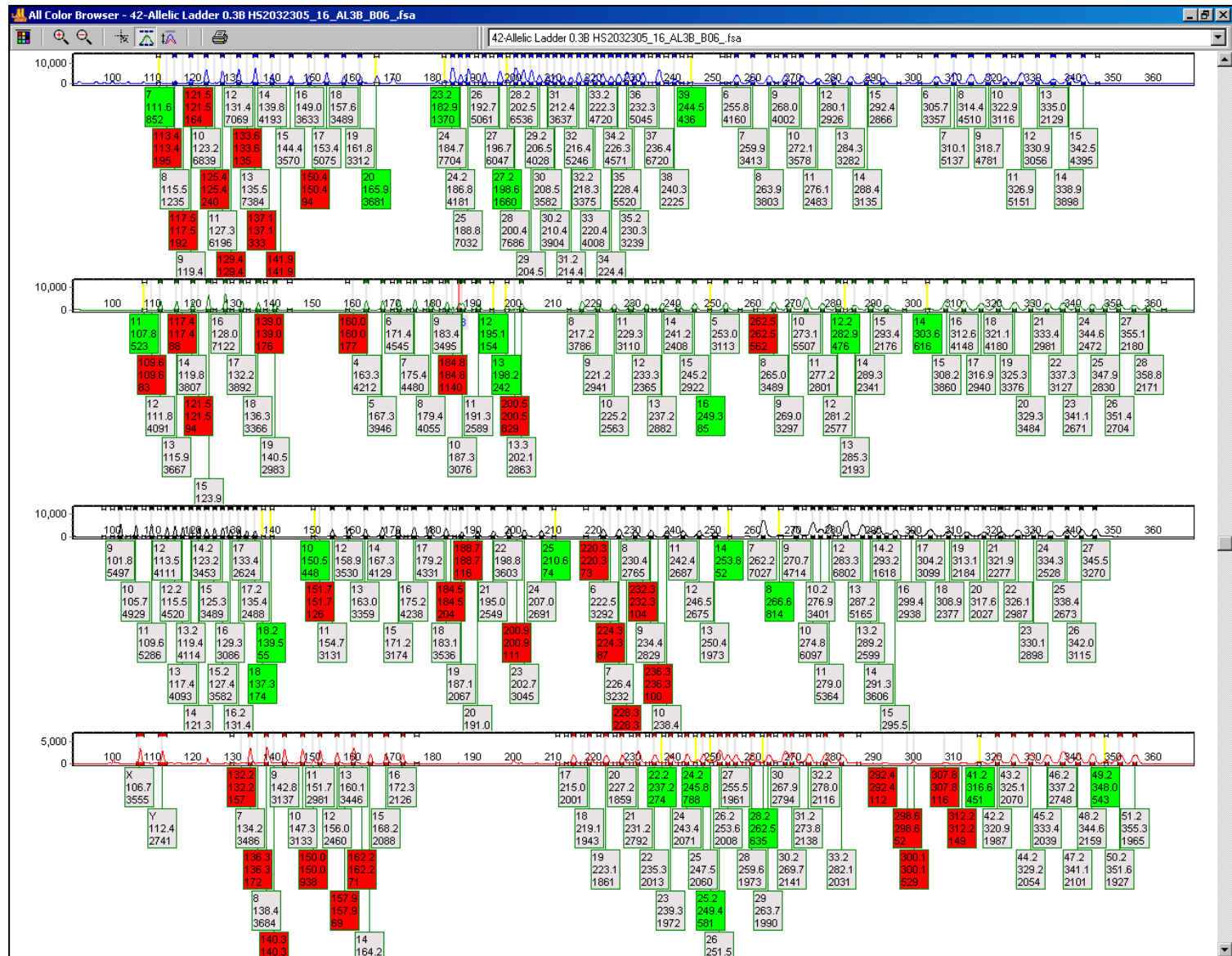


# Allelic Ladders

- GeneMarker<sup>®</sup> HID
- ID31 normal injection parameter
  - All published peaks are present
  - Peaks are present in virtual allele bins
- ID31 high injection parameter
  - 12 ladders failed- n = 63, 19%
  - Not all published peaks are present
  - Peaks are present in virtual allele bins
  - Peaks were present outside of bins



# GeneMarker® HID Ladders



# GeneMarker<sup>®</sup> HID Ladders

- GeneMarker<sup>®</sup> HID
- ID31 high injection parameter
  - Programmers provided the OCME with a new version
  - All ladders passed- used to generate bin offsets
- Other system high injection ladders were tested
  - ID28 ladders passed
  - Yfiler<sup>®</sup> ladders passed

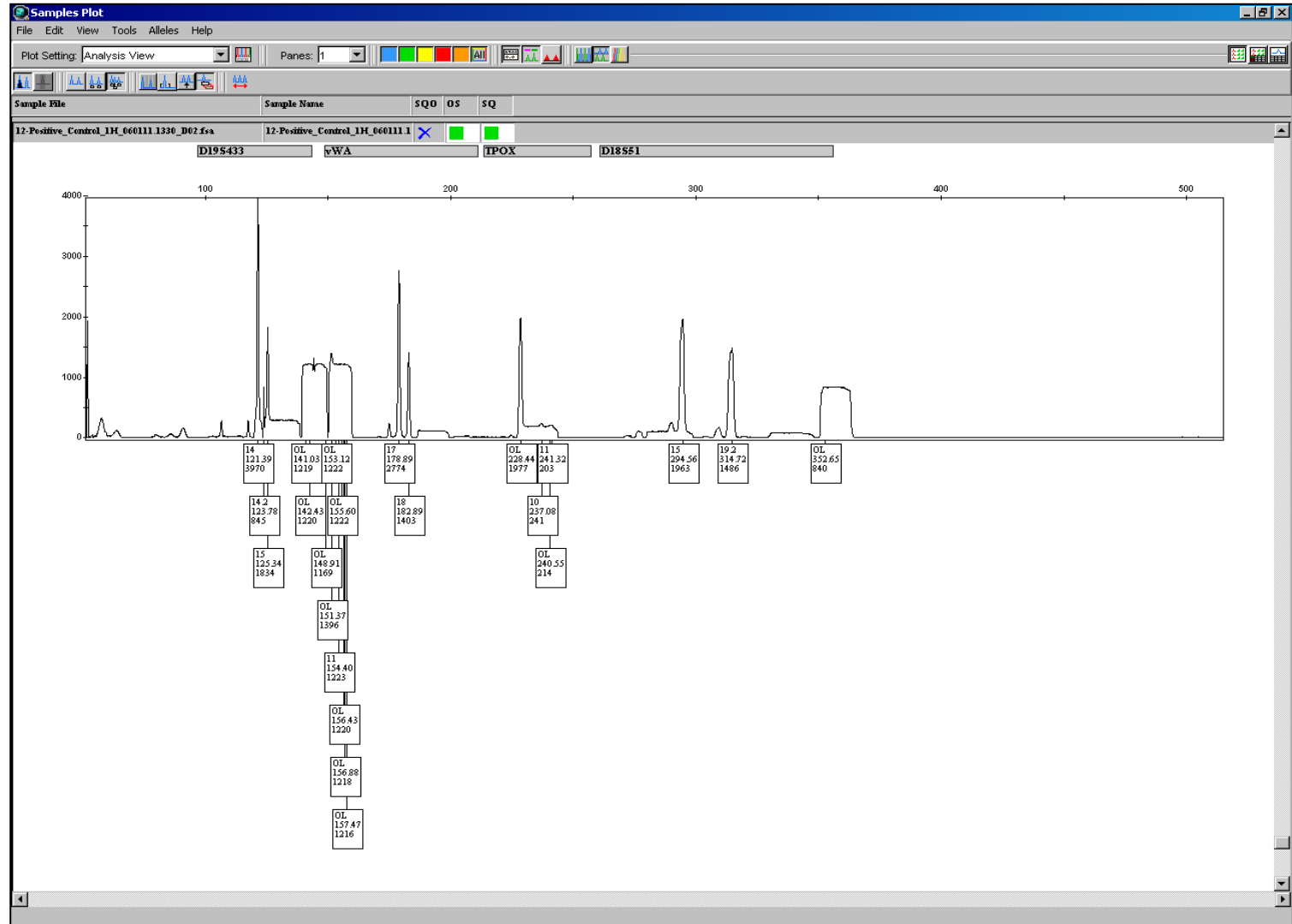


# Pull-up Correction in Positive Control

- Positive control- high injection parameters
  - Contain distorted peak shapes and extra allele calls
  - Results from pull-up of the internal size standard
  - Documented in GeneMapper® *ID*
  - Maintained in GeneMapper® *ID-X*
  - Resolved in GeneMarker® HID
    - Pull-up correction algorithm applied when data analysis is run



# Pull-up Correction in Positive Control





# Pull-up Correction in Positive Control



# Stutter Filters

- GeneMapper<sup>®</sup> *ID* stutter filters are fixed
  - 1 filter allowed per marker
  - Position determined by bp value of repeat
- GeneMapper<sup>®</sup> *ID-X* stutter filters
  - 8 filters can be programmed into each marker
  - Position set independently for each marker
- GeneMarker<sup>®</sup> *HID* stutter filters
  - 3 filters can be set at each marker
  - $N - x$ ,  $N - 2x$ , and  $N + x$  positions



# Stutter Filters

- AB's Yfiler<sup>®</sup> kit suffers with GeneMapper<sup>®</sup>*ID* filters
  - DYS19 is a tetranucleotide repeat
  - DYS19 has a known minus 2 stutter artifact
- GeneMapper<sup>®</sup>*ID-X*
  - Allows a minus 2 bp filter to be programmed at DYS19
- GeneMarker<sup>®</sup> HID
  - Requires that the x value be set at 2 bps at DYS19



# Mixture Assistant Tools

- Mixture Assistant Tools use edited data
  - GeneMapper<sup>®</sup> *ID-X*- ignores samples with OL peaks
  - GeneMarker<sup>®</sup> *HID*- assigns OL peaks to contributors
- Data for systems is exportable for use in MS Excel
- Systems provide multiple allele combinations
- Systems calculate most likely allele combinations
  - User defined settings
  - Residual score
  - A low score makes the combination more likely



# GeneMapper® ID-X

Mixture Analysis Results Viewer: Minimum Number of Contributors = 2

Sample File : 35-DH\_JR\_ACB\_511\_a\_C05\_005.fsa Panel : Identifier\_v1X-dup  
 Run Folder : HS3\_2009-09-22\_0088  
 Sample Name : 35-DH\_JR\_ACB\_511\_a

Mixture Analysis Results | RMP Statistics: C1 (Major) | RMP Statistics: C2 (Minor) | CPI/CPE Statistics | LR Statistics

Average Mx: 0.276 Residual Threshold: 0.040 Filter by Marker: All Known: None  Sample Review Complete

**Selected Genotype Combinations**

Marker	C1 (Major)	Inconclusiv	C2 (Minor)	Inconclusiv	Known Genotyp	ADBI	Mx	Residual	PHR1	PHR2	Residual Status	PHR Status	IQ
1	D8S1179	13,13	10,13				N/A	0.005	N/A	N/A		NA	
2	D8S1179	13,13	10,10				0.188	0.015	N/A	N/A		NA	
3	D21511	28,32	30,33.2				0.352	0.026	0.525	0.968			
4	D7S820	8,10	8,11				0.387	0.021	0.696	0.696			
5	D7S820	8,8	10,11				0.410	0.031	N/A	0.631			
6	CSF1PO	10,10	13,13				0.290	0.000	N/A	N/A		NA	
7	CSF1PO	10,13	10,10				N/A	0.010	N/A	N/A		NA	
8	CSF1PO	10,13	10,F1				0.207	0.039	0.515	0.515			
9	D3S1358	15,16	15,16				N/A	0.002	0.869	0.869			
10	D3S1358	15,16	16,16				N/A	0.021	N/A	N/A		NA	
11	D3S1358	15,16	16,F1				0.045	0.023	0.910	0.910			
12	TH01	7,10	10,10				N/A	0.000	N/A	N/A		NA	
13	TH01	10,10	7,7				0.369	0.017	N/A	N/A		NA	
14	TH01	7,10	10,F1				0.024	0.032	0.598	0.598			
15	TH01	7,10	7,10				N/A	0.034	0.584	0.584			

Sort...  
Unselect  
Missing Markers...

**Unselected Genotype Combinations**

Filter on IQ:     Filter on Known Match:

Marker	C1 (Major)	Inconclusiv	C2 (Minor)	Inconclusiv	Known Genotyp	ADBI	Mx	Residual	PHR1	PHR2	Residual Status	PHR Status	IQ
1	D8S1179	13,13	10,F1				N/A	0.023	N/A	0.078			
2	D8S1179	10,13	13,13				N/A	0.060	N/A	N/A		NA	
3	D8S1179	10,13	13,F1				N/A	0.136	0.250	0.250			
4	D8S1179	10,13	10,13				N/A	0.194	0.232	0.232			
5	D8S1179	13,F1	10,13				N/A	0.213	0.250	0.250			
6	D8S1179	10,13	F1,F2				0.028	0.245	0.232	N/A			
7	D8S1179	10,13	F1,F1				N/A	0.291	0.232	N/A			
8	D8S1179	10,13	10,F1				N/A	0.306	0.228	0.228			
9	D8S1179	13,F2	10,F1				0.197	0.320	0.015	0.015			
10	D8S1179	13,F1	10,10				N/A	0.321	0.018	N/A			
11	D8S1179	10,13	10,10				N/A	0.404	N/A	N/A		NA	

Sort...  
Select

Notes... Calculate Statistics... Export... Mixture Analysis Parameters... Samples Plot... Cancel < Back Next > Finish ?

# GeneMarker® HID

Mixture Analysis

Marker: All Markers

One Contributor  
 Two Contributors  
 42-091709.1600PE3H\_B06\_004  
 35-DH\_JR\_ACB\_511\_a\_C05\_005  
 36-DH\_JR\_ACB\_511\_b\_D05\_007  
 37-DH\_JR\_ACB\_511\_c\_E05\_009  
 39-MLD\_AJT\_ACB\_155\_a\_G05\_013  
 40-MLD\_AJT\_ACB\_155\_b\_H05\_015  
 43-MLD\_AJT\_ACB\_155\_c\_C06\_006  
 45-MLD\_AJT\_ACB\_511\_a\_E06\_010  
 46-MLD\_AJT\_ACB\_511\_b\_F06\_012  
 47-MLD\_AJT\_ACB\_511\_c\_G06\_014  
 Three or more Contributors

No.	Marker	Major	Minor	Major Mx	Residual	Major HIM	Minor HIM
1 - 1	D8S1179	13,13	10,13	0.63	0.0026		
1 - 2	D8S1179	13,13	10,10	0.81	0.0261		
1 - 3	D8S1179	13,13	10,0	0.80	0.0302		0.08
1 - 4	D8S1179	10,13	13,13	0.37	0.0540		
1 - 5	D8S1179	10,13	13,0	0.93	0.1381	0.25	0.25
1 - 6	D8S1179	10,13	10,13		0.1975	0.23	0.23
1 - 7	D8S1179	10,13	0,0	0.99	0.3148	0.23	
1 - 8	D8S1179	10,13	10,0	0.98	0.3241	0.22	0.22
1 - 9	D8S1179	10,13	10,10		0.4311	0.23	
1 - 10	D8S1179	10,10	13,13	0.19	0.5289		
1 - 11	D8S1179	10,10	13,0	0.18	0.7120		0.02
1 - 12	D8S1179	10,10	10,13		0.8825		0.23
2 - 1	D21S11	28,32	30,33.2	0.65	0.0231	0.53	0.95
2 - 2	D21S11	28,30	32,33.2	0.60	0.0399	0.43	0.77
2 - 3	D21S11	28,33.2	30,32	0.60	0.0434	0.41	0.81
2 - 4	D21S11	30,32	28,33.2	0.40	0.1204	0.81	0.41
2 - 5	D21S11	32,33.2	28,30	0.40	0.1238	0.77	0.43
2 - 6	D21S11	30,33.2	28,32	0.35	0.1406	0.95	0.53
3 - 1	D7S820	8,10	8,11	0.61	0.0177	0.70	0.70
3 - 2	D7S820	8,8	10,11	0.59	0.0227		0.64
3 - 3	D7S820	8,11	8,10	0.39	0.0543	0.70	0.70
3 - 4	D7S820	8,10	11,0	0.82	0.0775	0.43	0.15
3 - 5	D7S820	8,10	11,11	0.84	0.0864	0.43	
3 - 6	D7S820	8,11	10,10	0.75	0.0956	0.27	
3 - 7	D7S820	8,11	10,0	0.73	0.1132	0.27	0.10
3 - 8	D7S820	8,10	10,11	0.79	0.1189	0.34	0.34
3 - 9	D7S820	10,11	8,8	0.41	0.1293	0.64	
3 - 10	D7S820	8,11	10,11	0.70	0.1831	0.19	0.19
3 - 11	D7S820	10,11	8,0	0.40	0.2450	0.64	0.04
3 - 12	D7S820	10,11	8,10	0.21	0.2906	0.34	0.34
3 - 13	D7S820	10,11	8,11	0.30	0.3180	0.19	0.19
3 - 14	D7S820	10,10	8,11	0.25	0.3939		0.27
3 - 15	D7S820	11,11	8,10	0.16	0.4947		0.43
4 - 1	CSF1PO	10,10	13,13	0.71	0.0001		
4 - 2	CSF1PO	10,13	10,10	0.58	0.0068		
4 - 3	CSF1PO	10,13	10,0	0.80	0.0382	0.52	0.52
4 - 4	CSF1PO	10,10	13,0	0.66	0.0229		0.25
4 - 5	CSF1PO	10,10	10,13	0.42	0.0400		
4 - 6	CSF1PO	10,13	10,13		0.0869	0.41	0.41
4 - 7	CSF1PO	10,13	13,0	0.91	0.1547	0.37	0.37
4 - 8	CSF1PO	10,13	0,0	0.93	0.1554	0.41	
4 - 9	CSF1PO	10,13	13,13		0.2570	0.41	
4 - 10	CSF1PO	13,13	10,10	0.29	0.3337		
4 - 11	CSF1PO	13,13	10,0	0.27	0.4500		0.10
4 - 12	CSF1PO	13,13	10,13		0.6238		0.41

Trace Data Report

No.	Marker	Mixture	PI	PE	Contributor 1	Contributor 2	LR
1	D8S1179	10,13	0.13532	0.86468			--
2	D21S11	28,30,32,33.2	0.15716	0.84284			--
3	D7S820	8,10,11	0.45082	0.54918			--
4	CSF1PO	10,13	0.08576	0.91424			--
5	D3S1358	15,16	0.33474	0.66526			--
6	TH01	7,10	0.08787	0.91213			--
7	D13S317	8,10	0.04903	0.95097			--
8	D16S539	8,10,11,13	0.34725	0.65275			--
9	D2S1338	17,19,23	0.22225	0.77775			--
10	D19S433	13,13.2,14.2,15.2	0.13011	0.86989			--
11	vWA	14,19	0.02699	0.97301			--
12	TPDX	10,11	0.09433	0.90567			--
13	D18S51	14,15	0.10796	0.89204			--
14	AMEL	X	--	--			--
15	D5S818	9,12	0.15434	0.84566			--
16	FGA	22,24,27	0.11755	0.88245			--
		Cumulative:	5.94E-14	1.5.94E-14			--

Contributor 1: None  Contested

Contributor 2: None  Contested

Average Major Mx: 0.700

Cumulative LR:  RMNE: 1.5.94E-14

Comment:

# Mixture Assistant Tools

- Goal- determine the accuracy of assistant tools
  - n = 4, ID28, 4:1

<u>GeneMarker<sup>®</sup>HID</u>		<u>GeneMapper<sup>®</sup>ID-X</u>	
Major Donor	Minor w/ major	Major Donor	Minor w/ major
16/16	13/16* <sup>^</sup>	16/16	10/16* <sup>^~</sup>
16/16	12/16* <sup>~</sup>	16/16	8/16* <sup>^~</sup>
15/16*	11/16*	15/16*	5/16* <sup>~</sup>
16/16	14/16* <sup>~</sup>	15/16* <sup>^</sup>	9/16* <sup>~</sup>
<b>98.4%</b>	<b>78.1%</b>	<b>96.9%</b>	<b>50.0%</b>

\* At least once the correct combination was not the first valid choice

<sup>^</sup> At least once the correct combination was not a valid choice

<sup>~</sup> At least once the correct combination was not a choice



# Electropherogram Export

- New system- long term integration with LIMS
- Manner of electropherogram export is integral
- GeneMapper<sup>®</sup>*ID*- ID31 samples
  - Sorted by color channel and exported separately
  - Results in five documents per sample set
- GeneMapper<sup>®</sup>*ID-X* retains this problem
- GeneMarker<sup>®</sup> HID offers two solutions
  - Selected samples can be sorted by dye before printing
  - The sorted document can be exported as a .png or .jpeg
  - The sorted document can also be exported as a PDF





# Exported Data Tables

- Generation of exported data
  - GeneMapper<sup>®</sup> *ID&ID-X* saves export table formats
  - GeneMarker<sup>®</sup> HID does not save export table formats
- Report sheets- generated with excel macros
  - GeneMapper<sup>®</sup> *ID&ID-X* table data share export layouts
  - GeneMarker<sup>®</sup> HID table data has a different layout
- Available columns
  - GeneMapper<sup>®</sup> *ID&ID-X* allows user defined columns
  - GeneMarker<sup>®</sup> HID has no sample comment column



	GeneMapper®ID-X	GeneMarker® HID (w/Repair)
Sample Editing	More time consuming	Less time consuming
Oversaturated Peaks	Indicated	Repaired - fewer reruns
Peak Heights as Compared to Genotyper	3% lower	0.4% lower Gained information
Number of Sample Edits		Significantly fewer edits
Allele Calls in Mixtures		Gained more information
Ladders	All Passing Ladders	All Passing Ladders
Pull-up Correction	N/A	Removes pull-up peaks
Stutter Filters	Slightly more customizable	Useful - but less customizable
Mixture Assistant Tool		Easier to use More accurate
Exported Data	Separate PDFs per color channel (ID31)	One PDF or Separate .png or .jpeg file per print page (ID31)
Exported Tables	No additional work required	Additional work required

# Recommendation: GeneMarker<sup>®</sup> HID

- Simple analysis and fewer edits provide a reduction in analyst time requirements
- Saturated peak repair prevents re-running of samples- fewer reagents consumed
- Peak recognition algorithms result in a gain of accurate information
- Better functionality with soon to be implemented LIMS

# Stutter Filters

- Both systems allow for increased flexibility in stutter filters
  - Allows for the removal of the 10% global filter that is currently applied
  - Extensive additional validation required
- Goal- Assess a possible gain of useful information
  - n = 12 mixture samples for each condition
  - Information gained was examined with respect to known profiles

<b><u>GeneMarker<sup>®</sup>HID</u></b>	
<u>ID28 mixtures</u>	
Lost drop-out	16
New drop-in	4

<b><u>GeneMarker<sup>®</sup>HID</u></b>	
<u>ID31 touched items</u>	
Lost drop-out	8
New drop-in	18

<b><u>GeneMapper<sup>®</sup>ID-X</u></b>	
<u>ID28 mixtures</u>	
Lost drop-out	14
New drop-in	2

<b><u>GeneMapper<sup>®</sup>ID-X</u></b>	
<u>ID31 touched items</u>	
Lost drop-out	8
New drop-in	19



# Mixture Assistant Tools

- Goal- a more extensive study on tool accuracy
  - n = 10 samples per mixture ratio

<u>GeneMarker®HID</u>		<u>GeneMarker®HID</u>	
4:1 - 500pg - ID28		2:1 - 500pg - ID28	
Major Donor	Minor w/ major	Major Donor	Minor w/ major
16/16	13/16*^	13/16*	13/16*
16/16	12/16*~	13/16*^	13/16*^
15/16*	11/16*	12/16*	13/16*^
16/16	14/16*~	14/16*	12/16*~
16/16	12/16*	14/16*	13/16*^
16/16	8/16*	12/16*	10/16*
16/16	11/16*^	11/16*^	11/16*^
16/16	8/16*	15/16*	12/16*
15/16*	12/16*	13/16*	13/16*
15/16*	11/16*^	13/16*	13/16*^
<b>98.1%</b>	<b>70.0%</b>	<b>81.3%</b>	<b>76.9%</b>

\* At least once correct combination was not the first valid choice

^ At least once the correct combination was not a valid choice

~ At least once the correct combination was not a choice



# Acknowledgements

- Dr. Theresa Caragine
- Dr. Mechthild Prinz and the OCME Research team
  - Cindy Rodriguez, Troy Holder, and Kathleen O'Connell
- Dr. Teresa Snyder-Lieby and staff of SoftGenetics LLC
- Justin Godby and Valerie Bostwick
- Dr. Pamela Staton
- Alyssa Strohbusch
- Emily Fete



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