GeneMarker[®]HID Human Identity Software

- NDIS Approved Expert System
- Fast, Accurate, and User-friendly
 - ~ Documented time savings of up to 40%¹
 - ~ Up to 70% less analyst intervention²
- User Management with Audit Trail
- Mixture Analysis
- Relationship Testing
 - ~ Kinship

15 17 19

- ~ Paternity
- Database Search Tools:
 - ~ Missing Persons Search
 - ~ Mass Disasters
 - ~ State DNA Index System (SDIS) Search
- Compatible with all Major CE systems, Rapid instruments, and Human ID Chemistries.

You will see the difference!





GeneMarker®HID Software is an NDIS approved expert system designed for human identification and forensics applications. GeneMarkerHID Software offers several advantages including time savings¹, ease of use², and a suite of integrated post genotyping applications, thereby making it an excellent alternative to Genotyper[®], GeneMapper[®], GeneMa

Features:

NDIS Approved expert system Accurate Size/Allele Calls Linked Navigation Running Audit Trail All Major Chemistries

Project Review Tools:

All Color Browser Panel Editor Replicate Comparison Tool Project Comparison Tool CODIS Compatible Export

Human ID Applications:

Database Searching Mixture Deconvolution Percent Match Search Kinship Analysis Paternity Testing

GeneMarkerHID Software is compatible with all major commercially available Human Identity chemistries, as well as CE instruments, and Rapid Systems (Including 6 dye chemistries). The program's built-in panel editor tool also provides extensive support for custom chemistries. GeneMarkerHID Software can be run on Windows[®] Vista, 7, 8, and 10 Operating Systems.



User Operation

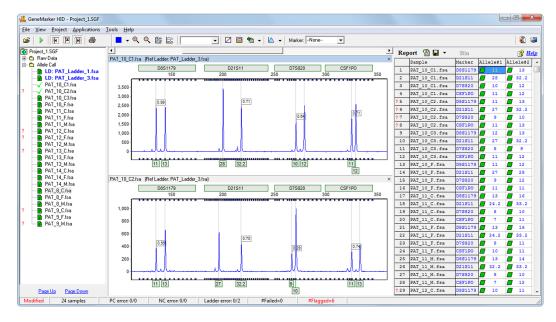
Thanks to its linked navigation and 'point and click' interface, GeneMarkerHID Software is easy to use for experts and beginners alike. The "Run Wizard" simplifies parameter setting by guiding the user through three simple steps, thereby making repetitive analyses quick, easy, and accurate. The user may select one of several preloaded templates, or create and save their own. GeneMarkerHID Software's patented pattern recognition technology automatically corrects for most chemistry errors, including peak saturation, noise, bleed-through, instrument spikes, and stutter, thereby reducing user intervention by up to 70%².

emplate Selection	Run Wizard			
Set the template of the project	Data Process - HID Analysis Set data process options		Run Wizard	
Select an existing template or create one Global Filler Jennithfiler PowerPlex_16 PowerPlex_18 PowerPlex_18 PowerPlex_18 PowerPlex_21 PowerPlex_6Color PowerPlex_ESI_17 PowerPlex_Fusion Use last template Use last template Cent	Raw Data Analysis Allel Image: Auto Range (Itame) Image: Auto Range (Itame) Start [0] End Image: Autor Range (Itame) Image: Autor Range (Itame) Image: Autor Range (Itame) Image: Autor Range (Itame)	le Cal Start [50 € End [500 €] In Internity, [50 €] Mas Internity, [30000 €] escentage > [20 €] Global Max	Additional Settings - HID Analysis Set additional option: related to the different analysis type Alfelic Ladder: NONE Positive Control Template: [PP16H5_3947A] Positive Control Template: [P16H5_3947A] Peter valuation Peter valuation Peter valuation Peter valuation Peter valuation Peter valuation Peter valuation Valid Moture Peter Percentage: [0] % Mm Mature Marker Number: [3]	Auto Salect Best Laddel Auto Panel Adjustment

The three pages of the Run Wizard allow the user to customize the analysis parameters for each project. Groups of settings can be saved as templates for future analyses.

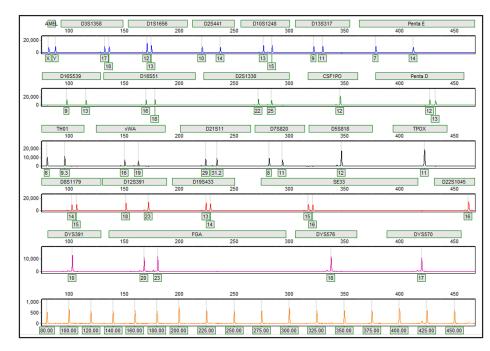
Main Analysis Window

After Data Processing, results are displayed in the Main Analysis Window. Colored icons on the left indicate the success of size calling, while colored icons in the report table indicate the strength of a given allele call. Flags alert the user to any fired rules.



The main analysis window: Colored icons indicate the success of size calling (left) and allele calls (right). The linked electropherogram in the center of the screen allows the user to edit, delete, and confirm calls.

Linked navigation allows the user to quickly and easily survey the results. A simple right-click allows the user to insert, delete, and edit alleles – actions that are all recorded by the software. Display options allow the user to customize what information is shown. The Main analysis window also serves as a portal to other tools and post-genotyping applications discussed below.

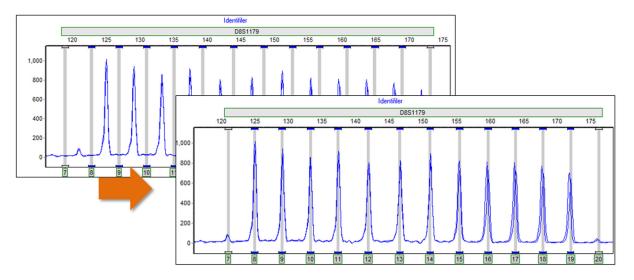


All Color Browser

The All Color Browser is another viewing option that allows the user to see every dye channel simultaneously. This view is particularly useful for detecting pull up and other artifacts, as well as making edits or comments.

Automated Panel Calibration

During data processing, GeneMarkerHID Software automatically calibrates the selected genotyping panel using Allelic Ladder samples that have been uploaded into the project. If multiple ladders have been uploaded, the program selects the best ladder for each sample.



The Panel Editor tool allows easy visualization and organization of genotyping panels. During panel calibration, panel bins are automatically aligned to ladder peaks, increasing accuracy.

Marker-Specific Parameters

In addition to the analysis settings discussed in the Run Wizard, GeneMarkerHID Software fully supports Marker-Specific analysis thresholds including:

- Homo and Heterozygote RFU thresholds
- Inconclusive (Stochastic) Ranges
- Heterozygous Imbalance thresholds
- Stutter Filters

These settings allow the user to fully customize their analysis parameters for each marker and save them for future analyses.

dit Marker	×
Marker Parameters	
Marker Name: D8S117	2
Nucleotide Repeats (x): 4	•
Boundary: 118.4	To 173.8
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 - ×	8 %
N - 2x	1 %
N + x	1 %
Apply Stutter Settings to All M	arkers
OK	Cancel

Marker specific settings for D8S1179

User Management and Audit Trail

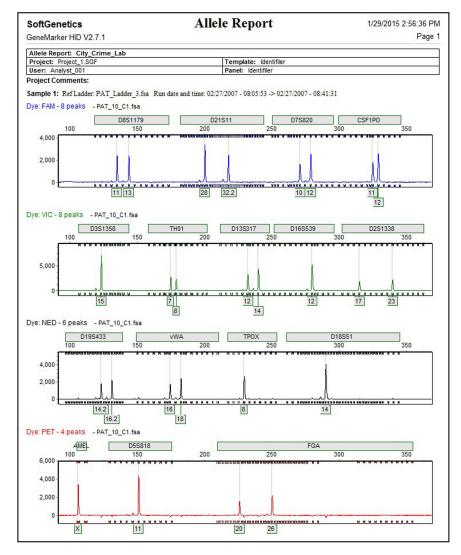
GeneMarkerHID Software is equipped with an extensive user management system and built-in audit trail. User management provides control of user access rights and generates an edit history for each sample profile in the project. Access rights for each user are set by the administrator – thereby providing assurance that unauthorized individuals don't accidentally modify or alter a project or settings. User Management is also linked with reports – report headers contain the analyst, date, institution name, and project information. Password control is also available, requiring users to log in with their password before opening the program.

Reporting Options

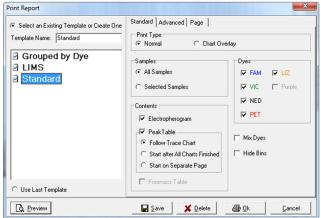
GeneMarkerHID Software supports numerous electronic and print reporting options, including:

- Complete PDF Reports
- Excel/Text table Reports
- CODIS Compatible Exports

GeneMarkerHID Software's extensive reporting customization options allow a vast array of different reporting formats and layouts. Print templates enable the user to save groups of settings under a single template name, enabling effortless printing of common reports.



Above is one of many possible print reports that may be generated in GeneMarkerHID Software. Detailed print settings (right) allow extensive customization of reports. Templates can be created and recalled for frequently used formats.



Linked Applications

Mixture Analysis Tool

GeneMarkerHID Software comes preloaded with a Mixture Analysis Tool. The tool can completely deconvolute mixtures with two contributors, and can calculate the probability of inclusion for mixtures with three contributors. In addition, GeneMarkerHID Software's mixture analysis tool automatically:

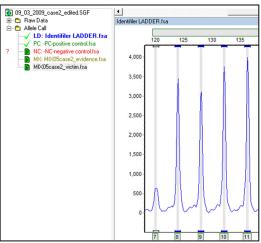
- Identifies the number of potential contributors in each mixture
- Displays all possible allele combinations at each locus
- Performs all PI, PE, LR, and RMNE calculations
- Ranks the most likely genotype combinations (with or without single source reference profiles)
- Deconvolutes major and minor contributors from mixture
- Searches database for consensus contributor genotypes

Using the recommendations of the DNA Commission of the international Society of Forensic Genetics (Gill et al. 2006) and methods of Clayton et al. 1998 and Gill et al. 1998, the mixture analysis application automatically determines the most likely genotype combination at each locus, and then determines the most probable contributor genotypes (major and minor).

If no files in the current project match a contributor genotype, the user may query GeneMarkerHID Software's built-in relationship testing database for possible matches. In addition, consensus contributor genotypes may be uploaded to the database for future searches.

In the main analysis window, sample names are colored according to the sample type. Mixture profiles are displayed in olive-green, negative controls in red, positive controls in green, and ladder profiles in blue.

In the Mixture Analysis Application, peaks are visually displayed in the trace comparison, while the peak table contains all data including PI, PE, LR, and RMNE. The peak table may be exported as .txt tab delimited or excel file. All results are presented in report tables that may be saved or copy/pasted into existing documents.



😼 🍓 භ 📸 🐔 Ma	rker: All M	arkers	-											
- One Contributor	No.	Marker	Major	Minor		Residual 🔺	Trac	e Data Repo	ort					
- Two Contributors	1-1	D8S1179	11,13	11,14	0.87	0.0000	No.	Marker	Mixture	PI	PE	Contributor 1	Contributor 2	LB
MIX05case2_evidence	1-2	D8S1179	11,13	14,14	0.93	0.0085	1	D8S1179	11,13,14	0.31586	0.68414	11,14	11,13	208.906
- MIX05case2_victim	1-3	D8S1179	11,13	13,14	0.88	0.0091	2	D21S11	28,30,31,32.2	0.32685	0.67315	30,31	28,32.2	1175.021
- Three or more Contributors	2-1	D21S11	28,32.2	30,31	0.83	0.0098	3	D7S820	8,9,10,11	0.77414	0.22586	9,11	8,10	144.475
	3-1	D7S820	8,10	9,11	0.87	0.0011	4	CSF1P0	7,10,12,13	0.41470	0.58530	12,13	7,10	1684.433
	4 1	CSF1P0	7,10	12,13	0.90	0.0033	5	D3S1358	15,16	0.40653	0.59347	15,16	15	34.717
	5-1	D3S1358	15,15	15,16	0.88	0.0000	6	TH01	7.8.9.3.10	0.53097	0.46903	8,10	7.9.3	3024.934
	5-2	D3S1358	15,15	16,16	0.94	0.0092	7	D13S317	8,9,12,14	0.27584	0.72416	8,9	12.14	15554.529
	6 - 1	TH01	7,9.3	8,10	0.89	0.0092	8	D16S539	9.10.11.12	0.68160	0.31840	9,12	10,11	176.555
	7-1	D13S317	12,14	8,9	0.85	0.0032	9	D2S1338	16,17,21,24	0.14541	0.85459	16.24	17,21	3758.716
	8 1	D16S539	10,11	9,12	0.85	0.0010	10	D195433	12,13,14	0.34027	0.65973	12.14	13	126.352
	9-1	D2S1338	17,21	16,24	0.90	0.0023	11	WA	15.16.19	0.24614	0.75386	16,19	15	938.906
	10 - 1	D19S433	13,13	13,14	0.84	0.0003	12	TPOX	8,9,10,11	0.73706	0.26294	8,11	9.10	193.028
	10 - 2	D19S433	13,13	12,14	0.82	0.0053	13	D18551	12.15.17.18	0.26381	0.73619	12.15	17,18	1069.590
	10 - 3	D19S433	13,13	14,14	0.92	0.0054	14	AMEL	X.Y	0.20301	0.73013	12,15 X	XY	
	11 1	vw/A	15,15	16,19	0.89	0.0006	15	D5S818	8.11.12.13	0.76054	0.23946	11.12	8.13	263.901
	12-1	TPOX	9,10	8,11	0.86	0.0062	16	FGA	20,23,24	0.12168	0.23348	23,24	20,24	362.640
	13-1	D18S51	17,18	12,15	0.84	0.0012	10	гая	Cumulative:	2.95E-07	1-2.95E-07	23,24	20,24	1.52E+41
	4	1 44471	~~	00	0.74	0.0070			Cumulative:	2.30E-07	1-2.35E-07			1.526+41
	Q	-A -				,								
	-	.0												
	Contri	butor 1: MD	(05case2_vic	tim (Minor)		- Contes	Com	ment:						
	Contri	butor 2: Ind	ivid_XYZ (Maj	jor)	·	 Contes 								
	A	ge Major Mx	0.970	_										

The mixture analysis application: Possible genotype combinations are shown on the left, and PI, PE, and LR values are calculated on the right. The dialogs in the bottom left allow the user to change which individuals are being contested.

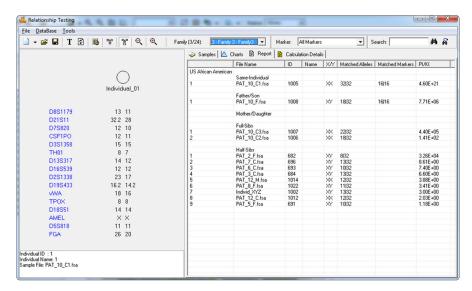
Database Searching

GeneMarkerHID Software's linked, searchable database enables hundreds of thousands of profiles to be imported into the program and saved for future searches. Genotypes can be imported from the current project, from a text file, or from CODIS .cmf files. This lends itself to numerous applications, including:

- Missing Persons Search
- SDIS Search
- Mass Disaster Identification
- Crime Scene Suspect or Familial Search
- Kinship Analysis Statistics for Three Generations

Using the Relationship Testing application, the user may query a sample against the database for potential matches across different populations at varying relationship levels. For each potential match, an Identity by Descent (IBD) calculation is used to determine the likelihood ratio of each possible relation to the test sample. Calculation details and the electropherogram are easily accessible for analyst review.

Genotypes can be imported into the database directly from the current project, limiting data transfer; they may also be imported from CODIS or text files.



The Database Search Tool: Profiles may be tested against a database of known genotypes. Likelihood Ratios (LR = PI/ KI) are displayed for the most likely candidate at each relationship level.

The linked kinship analysis tool allows direct comparison of two individual profiles on a variety of different relationship levels: siblings, parent-child, uncle-nephew, and so on. Likelihood ratios and probabilities are available for analyst review and can be saved as a text file, an excel file, or may be copy/pasted.

The built-in Kinship Analysis tool allows the user to calculate probabilities and likelihood ratios across three generations. Report tables may be saved as .txt tab-delimited files, or may be copy/pasted into an existing document.

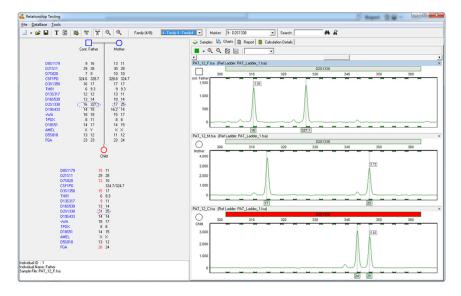
	Individ	hual A		Individ	tual R							
Marker		10_C1.fsa	•	_	10_C2.fsa	-	Parent/Child (LR)	Full-Sibs (LR)	Half-Sibs (LR)	Uncle-Nephew (LR)	Cousins (LR)	Grandpa
CSF1P0	11	12	Π	11	13		1.00390	0.75195	1.00195	1.00195	1.00097	1.00195
TPOX	8			8			2.68752	3.39945	1.84376	1.84376	1.42188	1.84376
TH01	7	8		7	8		1.88446	2.72596	1.44223	1.44223	1.22112	1.44223
ww.	16	18		15	18		1.61249	1.05624	1.30624	1.30624	1.15312	1.30624
D16S539	12			10	13		0.00201	0.25000	0.50000	0.50000	0.75000	0.50000
D7S820	10	12		9	10		0.75438	0.62719	0.87719	0.87719	0.93859	0.87719
D13S317	12	14		12	14		7.75647	12.57203	4.37824	4.37824	2.68912	4.37824
D5S818	11			11	12		2.14998	1.32499	1.57499	1.57499	1.28750	1.57499
FGA	20	26		22	25		0.00390	0.25000	0.50000	0.50000	0.75000	0.50000
D8S1179	11	13		11	13		6.76096	16.55182	3.88048	3.88048	2.44024	3.88048
D18S51	14			13	16		0.00302	0.25000	0.50000	0.50000	0.75000	0.50000
D21S11	28	32.2		27	32.2		4.29997	2.39998	2.64998	2.64998	1.82499	2.64998
D3S1358	15			15	17		1.65382	1.07691	1.32691	1.32691	1.16346	1.32691
D2S1338	17	23		17	20		2.51965	1.50983	1.75983	1.75983	1.37991	1.75983
D19S433	14.2	16.2	- 1	14	14.2		3.14624	1.82312	2.07312	2.07312	1.53656	2.07312

Paternity Testing

GeneMarkerHID Software's paternity testing application is a versatile tool with many capabilities, including:

- Automated (and manual) Pedigree Drawing
- Identity By Descent (IBD) Calculations
- AABB paternity trio and motherless case calculations
- Reverse Parentage Testing
- Easy visualization of results
- Separate Kinship Analysis tool

The relationship testing module is embedded within GeneMarkerHID Software, allowing the user to proceed directly from data processing to paternity and/or kinship analysis. Profiles from the current project are used to automatically construct pedigrees and family trees. Conflicting genotypes are automatically flagged and linked to an electropherogram comparison for user review.



An example pedigree demonstrating exclusion: Conflicting alleles are highlighted in red. The pedigree is linked to an electropherogram, allowing the user to review potential conflicts in more detail.

The user may select one of several preloaded allele frequency tables or create their own to contest individuals within their pedigrees. Paternity Index (PI), Probability of Exclusion (PE), and Probability of Paternity (POP) calculations are available for this purpose. A wide variety of settings allow the user to customize the procedure, and take into account variables such as mutation rates and step-wise mutations.

		-0	🧼 Sample	s 🛛 🔼 Chart:	🛿 🖹 Repor	t 🗎 Calcu	lation Details				
	Cont. Father	Mother		PI	POP	PE	Cont. Father	Mother	Child	Numerator	Denominator
			Total	8.64E+08	99.99%	1.0000000					
D8S1179	13 10	13 13	CSF1P0	2.01E+00	66.75%	1.00E+00	11:12	11:12	11	3.71E-02	1.85E-02
D21511 D75820	32.2 31 9 11	30 28 11 8	TPOX	2.69E+00	72.88%	1.00E+00	8	8	8	1.38E-01	5.15E-02
CSF1P0	12 11	12 11	TH01	6.62E+00	86.86%	1.00E+00	9	7	7:9	2.28E-02	3.45E-03
D3S1358 FH01	17 17 9 9	15 16 7 7	WA	2.06E+00	67.36%	1.00E+00	15:17	17	17	4.51E-02	2.18E-02
D135317	14 11	8 8	D16S539	1.29E+01	92.80%	1.00				3.79E-03	2.94E-04
D165539 D251338	12 8 24 18	11 9 24 25	D75820	1.14E+00	53.19%	1.00	Copy Table Export Calcu			1.10E-02	9.72E-03
D195433	16.2 15	13 14	D13S317	1.63E+00	62.01%	1.00E+00	111:14	ation Detail	811	1.07E-02	6.54E-03
VWA TPOX	15 17 8 8	17 17 8 8	D55818	8.46E-01	45.82%	1.00E+00	11:12	12:13	12:13	4.10E-02	4.85E-02
D18551	15 20	14 14	FGA	1.74E+00	63.54%	1.00E+00	23:24	21:23	21:23	1.04E-02	5.97E-03
MEL 055818	X Y 11 12	X X 12 13	D8S1179	1.72E+01	94.50%	1.00E+00	10:13	13	10:13	6.31E-03	3.67E-04
GA	24 23	23 21	D18551	7.79E+00	88.62%	1.00E+00	15:20	14	14:20	1.04E-02	1.33E-03
	Ľ	1	D21S11	6.14E+00	85.99%	1.00E+00	31:32.2	28:30	28:31	2.37E-03	3.85E-04
	Ch	ld	D3S1358	4.87E+00	82.95%	1.00E+00	17	15:16	16:17	2.11E-02	4.33E-03
			D351338	4.07E+00	92.77%	1.00E+00	18:24	24:25	18:25	1.55E-03	4.33E-03
D8S1179 D21S11	10 31										
D7S820	11	8	D195433	6.45E+00	86.57%	1.00E+00	15:16.2	13:14	14:15	1.05E-03	1.63E-04
CSF1P0 D3S1358	11 17										
TH01	9										
D13S317	11										
D16S539	8										
D2S1338	18										
D195433	15										
VWA .	17										
TPOX	8										
D18551	20										
AMEL	Y										
D5S818	12										
FGA	23	21									

An example pedigree demonstrating inclusion: PI, POP, and PE scores are calculated for each marker. The table can be copied directly or exported as a text file.

Percent Match and Cell Line Authentication Tool

The percent match tool uses a percent-similarity calculation to compare profiles in the current project to a database of known genotypes. This lends itself to several applications including the following:

- Cell Line Authentication
- Y-STR Comparison
- Database Search when allele frequencies are not available

Results are ranked in order of their percent match score, with differences highlighted in yellow. Results can be exported in the form of an excel or text file.

🕌 Cell Line Authenticat	ion	ALC: N	100	1	Contract 1	1	14	No. of Concession, Name			
<u>F</u> ile											
	Sample			Show Matches Greater Than 30 % Sex Marker AMEL							
Marker	Y23_Mixture_01.SG 💌 🗃	Y23_Suspect_01.SG1		No	File Name	ID	X/Y	Matched / Total	Percent Match	Matched Marker DYS391, DYS5	
		T125_Suspect_01.Su1	Shared Number	2	Y23_Suspect_01.SG1 Y23_Suspect_02.SG1	2	Unkn	25 / (38, 38) 27 / (42, 42)	65.8% 64.3%	DYS391, DYS5	
DYS576	<mark>16 17</mark>	16	1								
DYS448	<mark>19 20 -</mark>	20	1								
DYS389 II	<mark>28 30 </mark>	28	1								
DYS19	<mark>13 14</mark>	14	1								
DYS391	10	10	2								
DYS481	<mark>26 27</mark>	26	1								
DYS549	13	13	2								
DYS533	11	11	2								
DYS438	<mark>10 11 </mark>	10	1								
DYS437	14 16	16	1								
DYS570	19	19	2								
DYS635	<mark>21 22 </mark>	21	1								
DYS390	23 24	23	1								
DYS439	<mark>10 12 </mark>	10	1								
DYS392	<mark>11 16</mark>	11	1								
DYS393	13	13	2								
DYS458	<mark>15 19</mark>	15	1								
DYS385	13 14	13 14	2								
YGATAH4	<mark>11 12</mark>	11	1								
	Sum: 38	Sum: 38	Sum: 25								

The Percent Match Tool: After querying a sample against the database, results will be ranked on the right according to their percent match score. Here the tool is being used to demonstrate that all peaks from a Y-STR profile are included in a mixture sample.

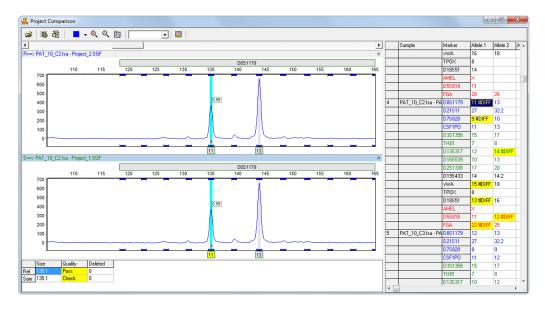
Quality Control and Validation Tools

Project Comparison Tool

The project comparison tool compares two completed projects to one another, enabling easy identification of any inconsistencies. This can provide significant time savings when comparing projects completed by two different analysts, or when performing a validation between two different procedures.

Related projects are opened in a single screen; differences detected in the parameters of interest are highlighted in the report table and linked to the electropherogram. The tool reduces time required for SOP and analysis system validation.

The Project Comparison Tool: Projects are displayed side by side with differences flagged in yellow.

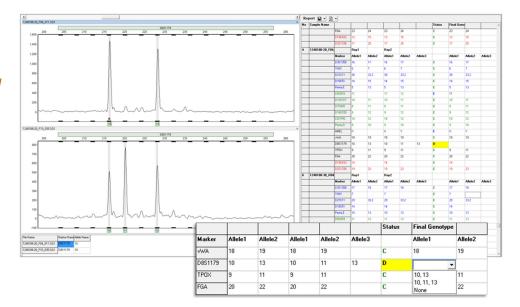


Replicate Comparison Tool

The Replicate Comparison Tool automates replicate comparison and determination of consensus genotypes. This tool is a valuable companion to genotyping applications, and any chemistry that has elevated risk of allele dropout and other artifacts. The tool groups replicates together, and automatically compares each replicate based on a number of factors, including peak call, size, height, and score.

Flagging alerts the user to any discrepancies, at which point the user may manually enter the desired allele call, or select none if a consensus cannot be determined. Otherwise, if each replicate has the same call, the program automatically selects that call as the consensus genotype. Linked navigation and sorting features allow the user to quickly and efficiently navigate through the results, which can be exported as a text or excel file. The replicate comparison tool can also accommodate larger projects with up to 5 replicates per sample.

The replicate comparison tool: A peak comparison is displayed on the left, while sample genotypes are displayed on the right. Flagging indicates whether or not the calls are concordant based on user defined parameters, and warns of possible mixtures or contaminants.



Please open disc to review your applications of interest and to install a 30-day trial of GeneMarkerHID Software.

Disk Contains:

Applications Notes 30-day free GeneMarkerHID Trial User Manual Demonstration Data

Minimum Hardware Requirements

PC

32 or 64 bit Windows® Operating System: Vista, 7, or 8, 10 Processor: Pentium III, 1GHZ RAM: 128MB 20GB Minimum available hard disk space.

Intel Powered Macintosh Computer

Virtual Windows Machine running on OS: 10.4.6 or higher. RAM: 2GB 20GB Minimum available on hard disk

References

- 1) Holland, Mitchell and Walther Parson. GeneMarkerHID: A Reliable Software Tool for the Analysis of Forensic STR Data. J Forensic Sci. 2011. 56:29-35.
- 2) Schmidt, Ronald. Evaluation of GeneMapper ID-X and GeneMarker HID for use in Forensic DNA Analysis. 2012; http://www.marshall.edu/forensics/files/2012/09/Schmidt-Ron-Poster-New.pdf.





Software PowerTools for Genetics Analysis

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