

SOFTGENETICS[®]

GeneMarker[®]

The Biologist Friendly Software

Unique Tools to Increase Genotyping Speed and Accuracy

Compatible with all Major Sequencing Systems

Embedded Applications Eliminate Data Transfer



SOFTGENETICS[®]
Software PowerTools for Genetics Analysis

GeneMarker[®]

The Biologist Friendly Software

GeneMarker[®] is unique genotyping software that has been designed to provide genetic researchers with a “biologist friendly” genotyping tool. The program’s linked-navigation and intuitive layout make it easy to use, while its accuracy, speed, and extensive collection of post-genotypic applications make it a powerful research tool. These features are summarized below:

Features:

- Ease of Use
- Accurate Size/Allele Calls
- Rapid
- Linked Navigation
- Customizable Settings
- Low Cost

Clinical Research Applications:

- FragileX
- Trisomy/Aneuploidy
- MLPA
- MS-MLPA
- Cystic Fibrosis
- Microsatellite Instability

Basic Research Applications:

- AFLP
- T-RFLP
- Microsatellite
- SnapShot/SNPlex/SNPWave
- Clustering Analysis
- Kinship Analysis

GeneMarker can perform analysis on up to 1,000 lanes of four or five color data sets generated by either slab gel or capillary electrophoresis. GeneMarker is compatible with files from all major capillary and slab gel electrophoresis systems including ABI files (*.FSA, *.AB1, *.ABI, *.HID), SCF files, MegaBACE™ files (*.RSD, *.ESD), Beckman-Coulter files, and slab gel image files (TIFF, BIP, JPEG and TXT) from systems such as the LI-COR, DNA Analyzers and Kodak Image Station when used in combination with our JelMarker[®] software.

GeneMarker software is an excellent replacement for software packages such as SAGA from LI-COR, GeneMapper[®], Genotyper[®], and GeneScan[®] from Applied Biosystems, MegaBACE[®] Genetic Profiler and Fragment Profiler software.



**Be Wise,
Choose
GeneMarker!**

User Operation

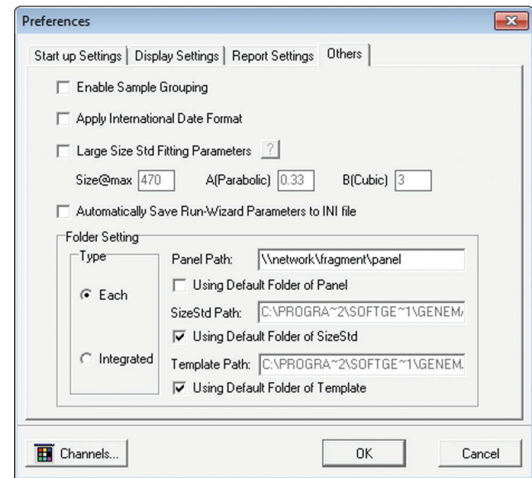
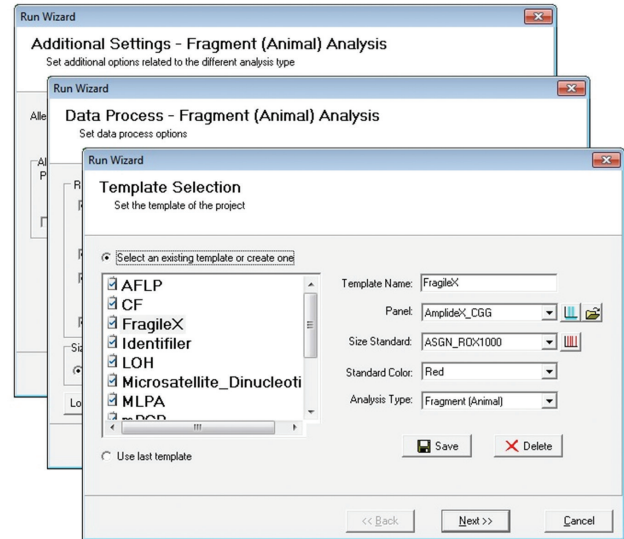
GeneMarker's "Run Wizard" simplifies parameter setting by guiding the user through 3 simple steps, thereby making repetitive analyses quick, easy, and accurate. The User may select one of several embedded templates, or create and save additional ones. GeneMarker's pattern recognition technology automatically corrects for most instrument and chemistry errors, such as saturated peaks, noisy data, wavelength bleed-through, instrument spikes, and stutter peaks.

Nearly every function of GeneMarker has been automated so that once the template is selected from the menu, or created by the user, the software automatically performs the analysis, providing a myriad of display and reporting options. Once the analysis has been completed and confirmed, the software saves all of the analysis parameters, raw data, and results for easy archiving and call back at a future date.

GeneMarker software includes many tools designed to assist the quality and speed of analysis. Results are clearly displayed and linked to electropherograms, navigation is simple and allows for efficient, detailed review of projects. GeneMarker software includes a user control function that password protects and limits program access, while also providing an audit trail of edits and comments.

In addition, GeneMarker allows users to change the default directories to which panel XML files, size standards, and template files are saved. This feature allows easy synchronization of computers that are connected via a network.

The View Preferences window, which enables users to change display settings and default working directories.



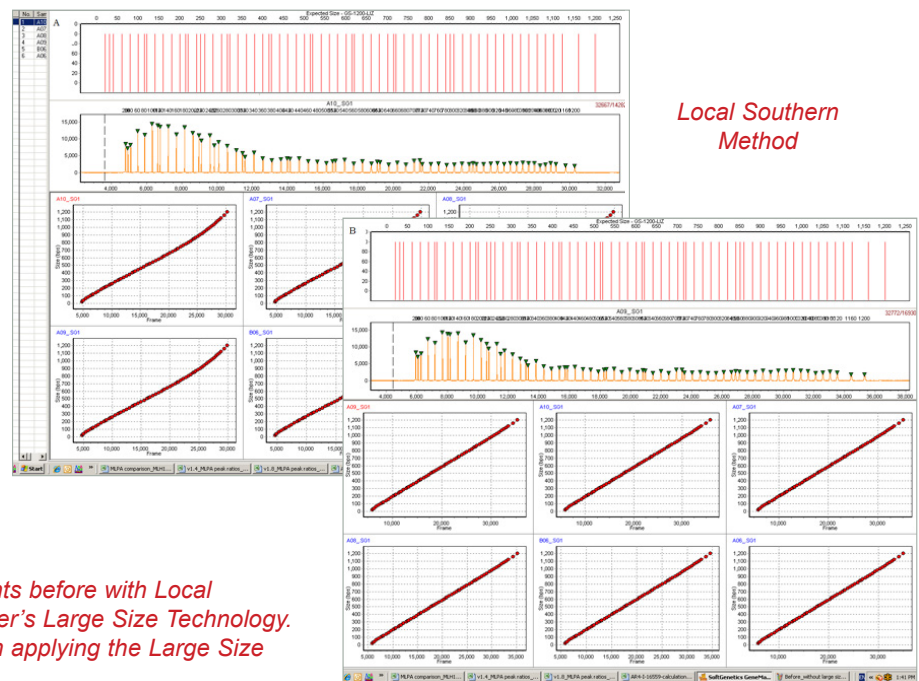
Sizing Technologies

GeneMarker's advanced sizing technology rapidly and accurately performs size calling for a variety of sizing chemistries. Size calling options include:

- Local Southern
- Cubic Spline
- Large Size Range (1000bp+)
- Custom Size Ranges

This flexibility affords many time and cost reducing multiplexing opportunities, such as the addition of 4x probes to clinical assays (such as MLPA, Allele Specific Amplification ARMs™, ASA, OLA, etc.) forensic profiling, and ecology analyses including TILLING®, STR/microsatellite profiles, AFLP, T-RFLP, VNTR, and BAC fingerprinting.

Comparison of the linear range for DNA fragments before with Local Southern (80-800 bps) and after with GeneMarker's Large Size Technology. Note the linear calibration curve that results from applying the Large Size call setting which is unique to GeneMarker.

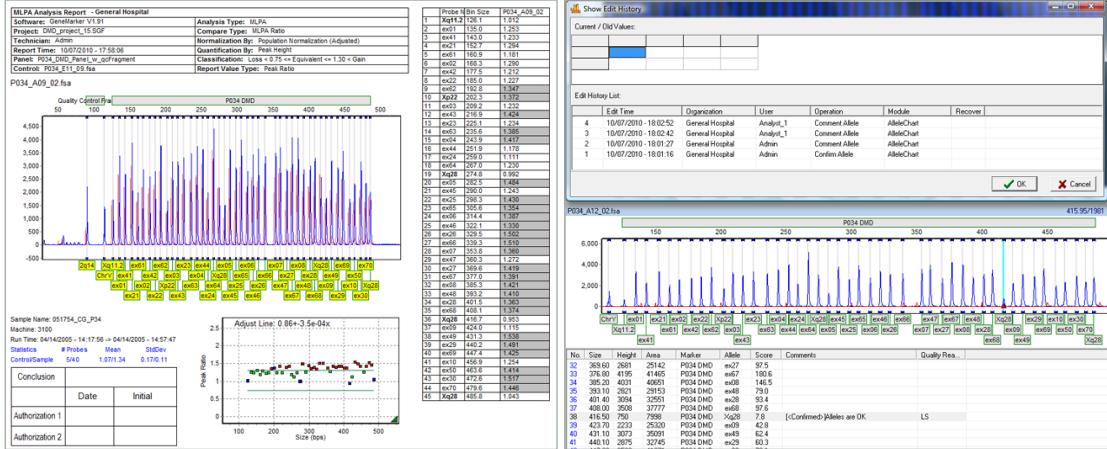


New Large Size Method

Useful Tools

User Management and Audit Trail

User management provides control of user access rights and generates an edit history for each project. Access rights for each user may be selected by the administrator – providing assurance that unauthorized individuals do not accidentally modify or alter a project. User management also provides a user ID and Organization name to the header of the final clinical research reports.

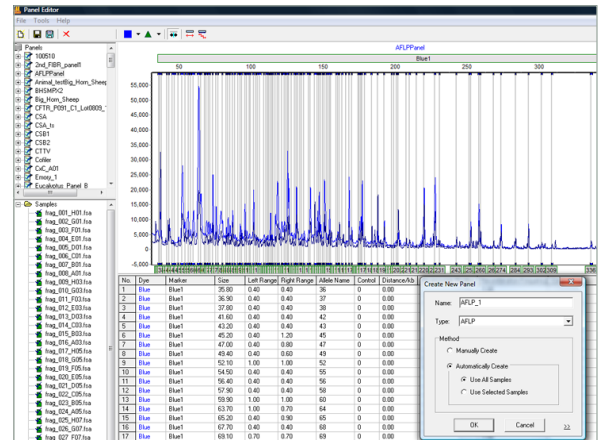


Print Report Header (left) has Institution and User ID. User Management (right) records and saves edit history and comments.

Easy-to-use Panel Editor

Panels can be selected from several embedded templates, downloaded and imported from <http://www.softgenetics.com/downloads.html>, or the user can create and save custom templates. Previously saved ABI panels can also be imported into GeneMarker. Custom panels are easily created in the panel editor and can be generated automatically using the signal information from data files. They may also be created manually, by simply inserting the desired alleles using the mouse.

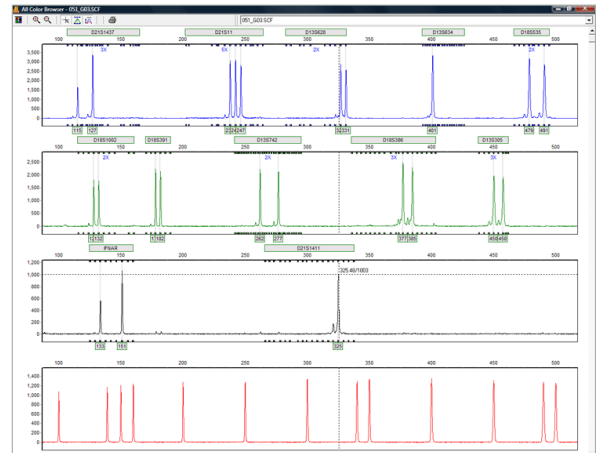
Custom Panels are easily created for applications such as AFLP or microsatellite analysis.



All Color Browser

The All Color Browser provides a complete 4 or 5 color review of an analysis. Edits or comments made in this view are automatically recorded in the electropherogram, allele report and peak table of the main analysis window and are saved when the project is saved.

The All Color Browser



Project Comparison Tool

The Project Comparison tool provides two useful functions. First, it can be used to compare two independent analysts' projects. Second, it can be used as a validation tool to determine differences in allele calls based on analysis parameters or instrument runs. Related projects are opened in one screen; any differences in the selected parameters are highlighted in the report table and linked to the electropherogram. The tool reduces time required for SOP and analysis system validation.



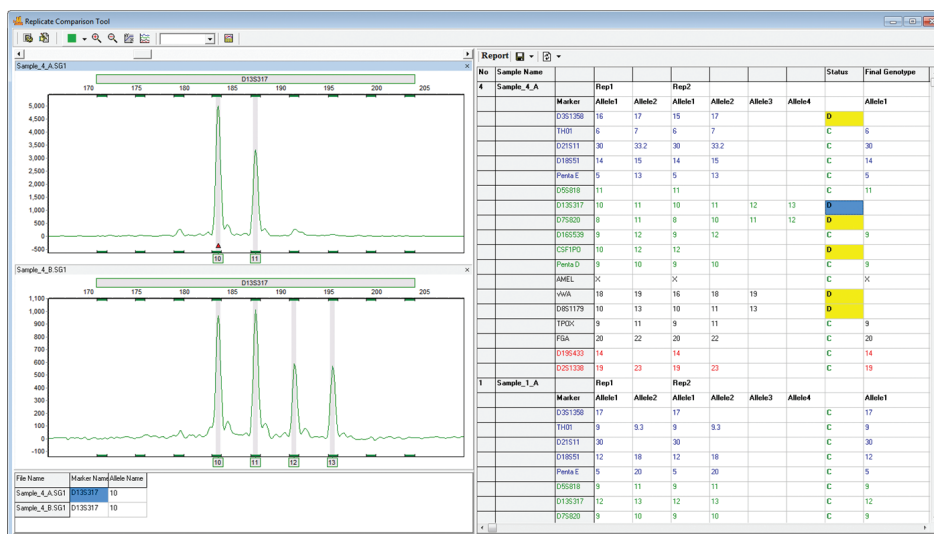
Highlighted Cells in the table on the right differ in one or more of the parameters selected in the Comparison Settings. Click on the highlighted cell to zoom in on the electropherogram and display the results details in the lower table.

Replicate Comparison Tool

The Replicate Comparison Tool automates replicate comparison and determination of consensus genotypes. The replicate comparison tool is a valuable companion to clinical research applications, 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. Otherwise the call is automatically determined by the program.

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.



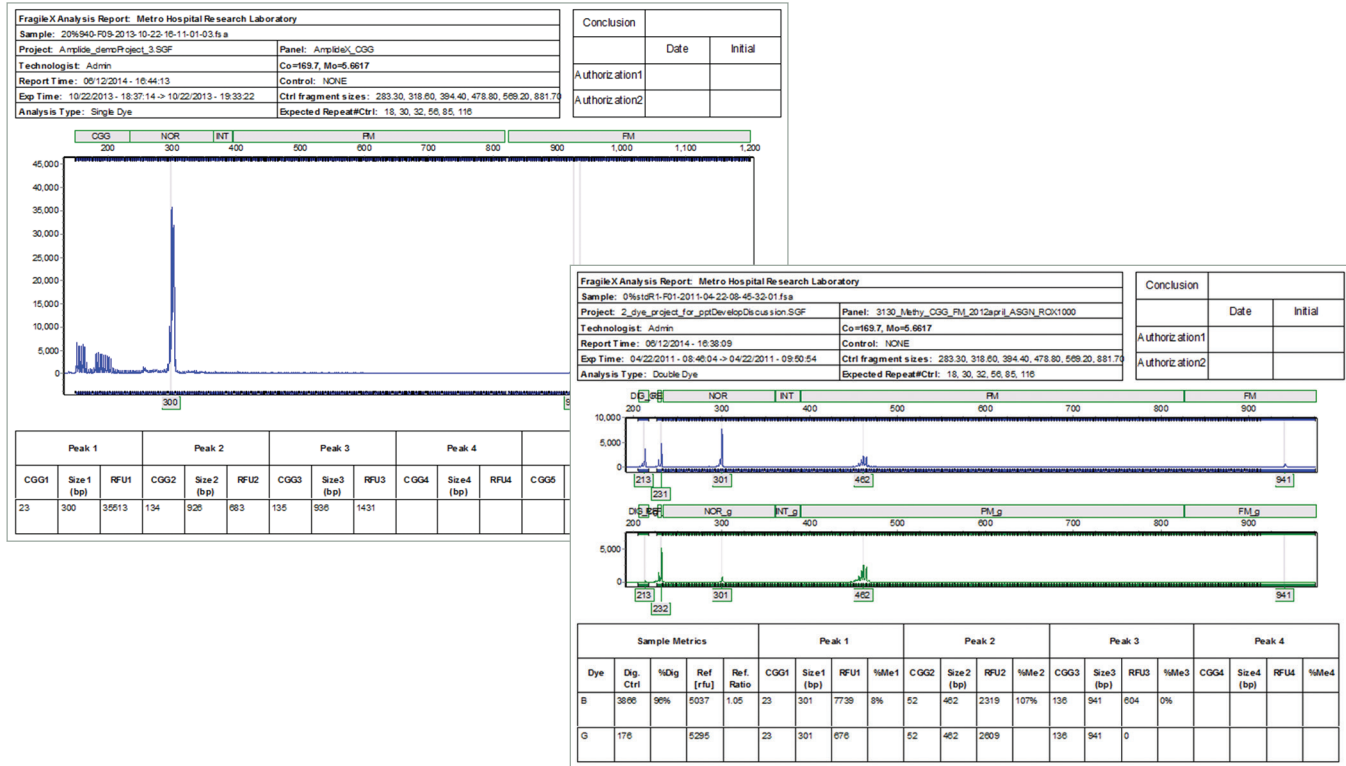
Replicates are displayed visually to the left, and the results of the comparison are displayed in the table to the right. Yellow flagging indicates a discrepancy between the replicates.

Clinical Research Applications

Fragile X Analysis

The Fragile X Application automates the analysis of both single and double-dye Fragile X chemistries, as well as other triplet repeat chemistries.

GeneMarker automatically calculates the triplet repeat number for each sample, and determines if the sample is in the Normal, Intermediate, Pre-Mutation, or Full-Mutation Range. The user may extensively customize the settings, which are conveniently saved by the program to reduce clicks.



The Fragile X Single and Double-Dye reports contain a header with project specific information, the electropherogram, and an informative results table.

The Fragile X Analysis application supports several print and digital exporting options, including excel and text results tables, a CGG Repeat Summary Sheet, and sample-specific summary sheets.

CGG Repeat Summary Sheet

Job ID: Amplide_demoProject
 Operator:
 Data Processed: 06/11/2014 - 14:35:27
 Sample: 5

Signal > Saturation Limit (FAM)
 Signal < Signal Cutoff

ID	Allele Ranges Detected				Peak 1	Peak 2	Peak 3	Peak 4	Peak 5	Peak 6
Sample File	Nor	Int	PM	FM	CGG1	CGG1	CGG2	CGG2	CGG3	CGG3
20%940-F09-2013-10-22-16-11-01-03.fsa					23	>200	>200			
PC-1_A04_2013-07-26_1_08-23-13.fsa					18	30	56	85	116	>200
PC-2_B04_2013-07-26_1_08-23-13.fsa					18	30	56	85	116	>200
PC-F10-2013-10-22-16-11-01-04.fsa					19	31	33	58	86	117
SC-F11-2013-10-22-16-11-01-04.fsa					31	58	>200	>200		

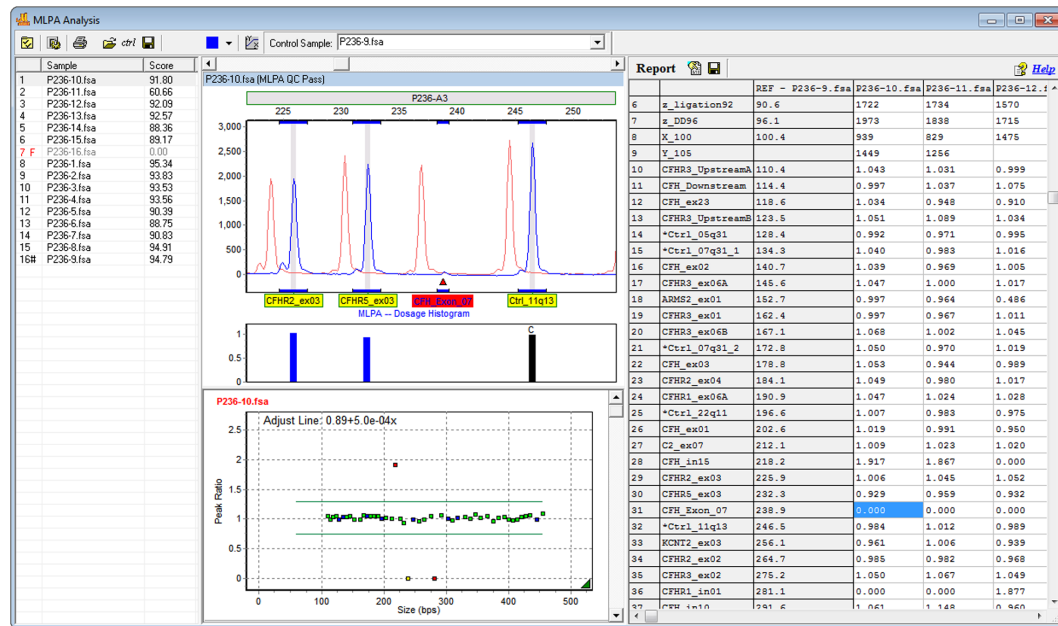
Example CGG Repeat summary sheet, one of the several exports available from the Fragile X Analysis application.

MLPA® Analysis

MLPA analysis using GeneMarker is easy, rapid, and accurate, making GeneMarker an excellent alternative to Genotyper®, GeneMapper®, Coffalyser.Net®, and other software packages. GeneMarker's extensive settings give the user complete control over the project and provide two advanced normalization methods (population and control-probe) to increase the rigor and reliability of analyses.

The MLPA application's visual interface and linked navigation make the program easy and intuitive to use, while eliminating slow and error prone transfer of sizing data. Results are easily interpreted using the electropherogram, ratio plot, and allele table, all of which can be customized, digitally saved, and printed directly.

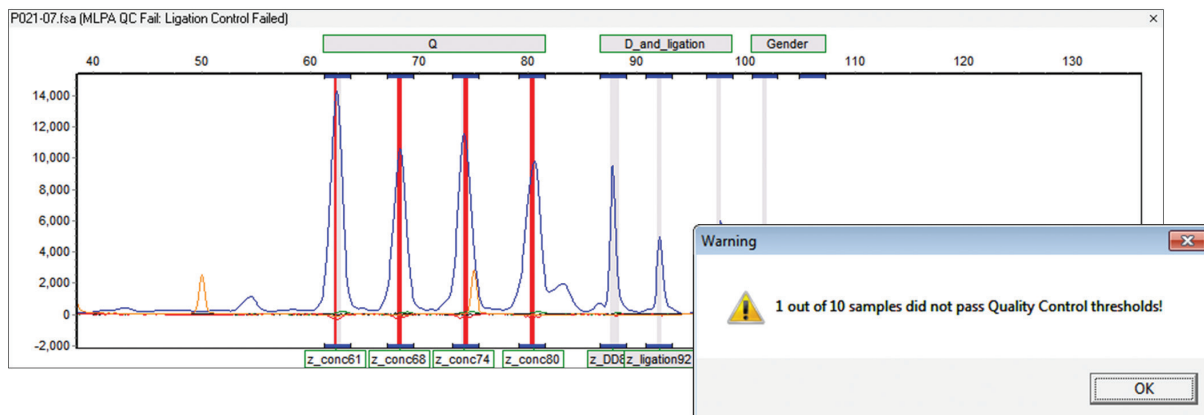
A large suite of panel XML files are pre-loaded in the program. In addition, new panel files can easily be acquired through the SoftGenetics webpage, or by simply emailing SoftGenetics technical support. GeneMarker's advanced panel editor easily facilitates any panel calibrations that may be necessary.



The MLPA Analysis Window, showing the trace comparison, ratio plot, and report table. GeneMarker software's MLPA module automatically indicates samples failing to meet user set quality parameters, indicated by a red F (left). Here the electropherogram is zoomed in to display a homozygous deletion.

MLPA Quality Control

MLPA analysis is enhanced by new quality control settings. The program automatically detects important QC fragments and warns the user if any exceed the pre-determined thresholds recommended by MRC-Holland.



A pop-up message alerts the user of any samples that failed the Quality Control settings. Potentially abnormal peaks are highlighted with red.

Trisomy Analysis

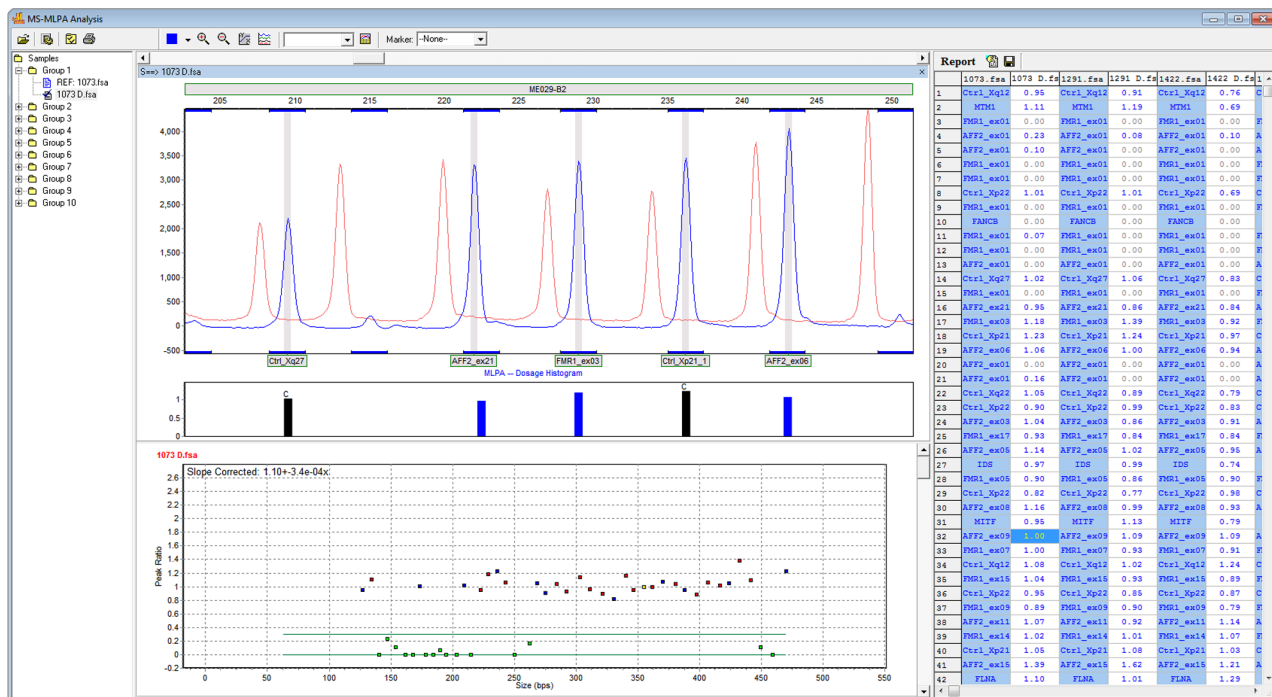
GeneMarker aids technicians and researchers analyzing QF-PCR products to detect aneuploidy with highly accurate size calling and a user-friendly interface. The trisomy application is directly linked to the main analysis screen – there is no need for error prone data transfer. Analysts may select the default analysis and report settings; which follow the **QF-PCR for the Diagnosis of Aneuploidy Best Practice Guidelines by the Association for Clinical Cytogenetics**, or customize the analysis parameters and final report. GeneMarker is ideal for analysis of data generated by major QF-PCR kits such as: Aneufast™, Devyser™, Elucigene™, CyberGeneAB™ and custom chemistries.

The Trisomy Print report showing the header with project information, electropherograms, summary table, and validation box. Samples in the triallelic range are highlighted in gray.



MS-MLPA®

GeneMarker DNA analysis software has been successfully paired with the Multiplex Ligation-dependent Probe Amplification technique for detecting genetic deletions and duplications associated with various diseases including cancer. The technique has been improved to detect methylation sites within promoter regions and for genomic imprinting applications. Promoter Methylation kits from MRC-Holland include ME001B Tumor Suppressor, ME002 Tumor Suppressor, and ME011 for Mismatch Repair genes. Genomic Imprinting kits from MRC-Holland include ME028 PWS-AS and ME030 BWS-RSS. GeneMarker's Methylation Specific – MLPA® module quickly and accurately detects methylation sites for researchers studying promoter methylation and genomic imprinting diseases. GeneMarker's ease of use and professional reporting options are an excellent choice for MS-MLPA® applications.



The MS-MLPA analysis window includes a trace comparison above, and a ratio plot below. In the trace comparison, the reference sample is displayed in red, while the digested sample is displayed in blue. In the ratio plot, the "normal range" is indicated by parallel green lines. Points above these lines are in the methylation range.

Microsatellite Instability (MSI)

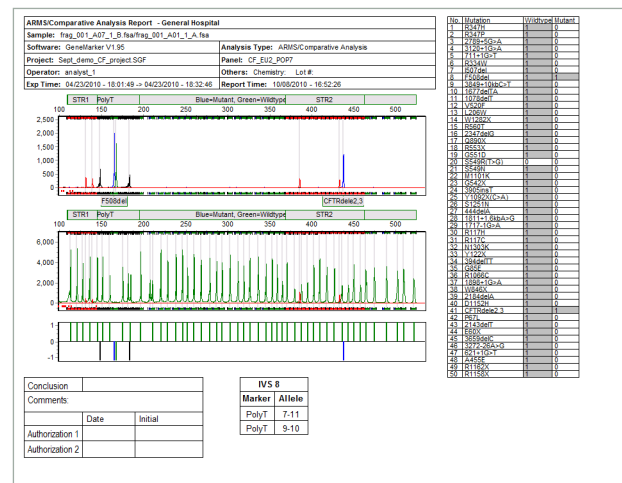
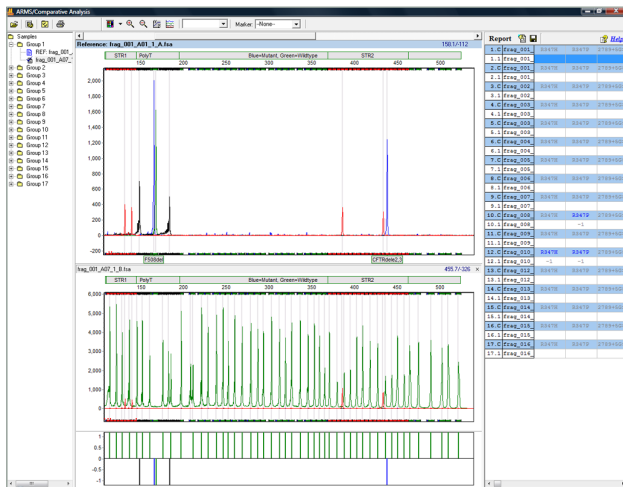
Microsatellites are stretches of DNA where a 1-5 base pair sequence is repeated several times. The most common microsatellite in humans is a dinucleotide repeat of CA which occurs tens of thousands of times across the genome. Microsatellite instability (MSI) is a condition where repeat units are gained or lost within a locus resulting in length polymorphism. Certain repeat regions are known to be highly polymorphic and heritable. Microsatellite instability within and around certain genes can have devastating effects due to the possibility of frameshift mutations.

In GeneMarker's MSI Analysis module, tumor samples are compared to normal samples based on peak-to-peak comparison. Differences between the two traces are displayed in a Trace Comparison Histogram below each electropherogram. Within the electropherogram, the tumor sample trace is overlaid on a light red reference trace. In this way, the researcher can easily visualize where the areas of instability exist.



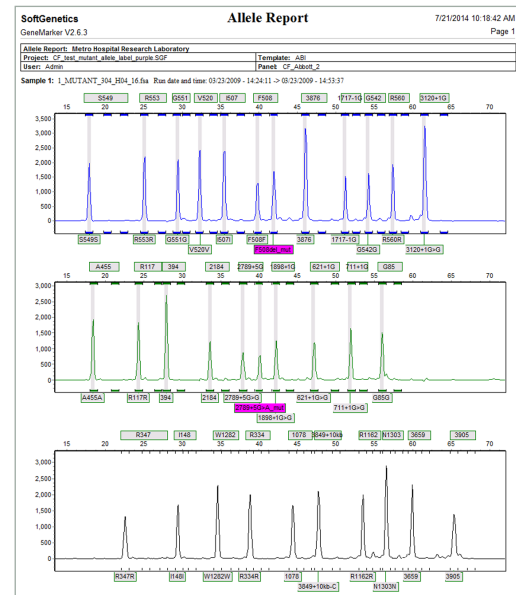
Cystic Fibrosis - Elucigene®

The ARMS/Comparative Analysis application provides comparison of file pairs for a given individual within the same project and a final clinical research report. It is ideal for analysis of samples from Elucigene EU-2 for cystic fibrosis (or any project where the different dye channels for duplicate files need to be compared). The ARMS/Comparative Analysis application provides an accurate, concise comparison of the two files for one individual.



Cystic Fibrosis - Abbott®

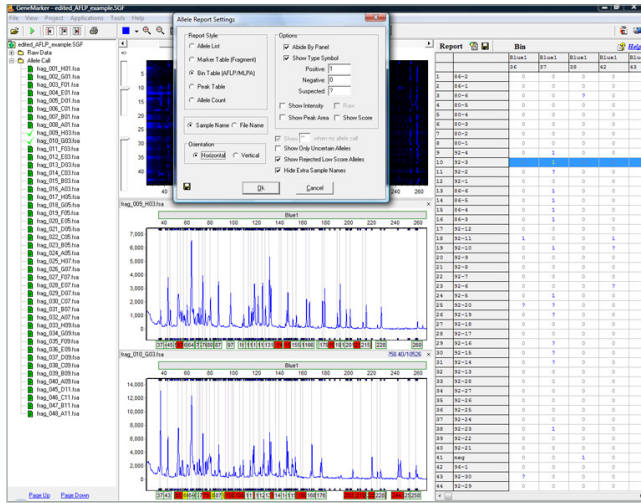
Is ideal for CF analysis using kits such as the Abbott Cystic Fibrosis Genotyping Assay. The main analysis screen provides rapid visual evaluation of mutant alleles the electropherogram and Allele Report. Mutant allele labels are displayed in contrasting color in electropherogram for instant visualization of mutant alleles.



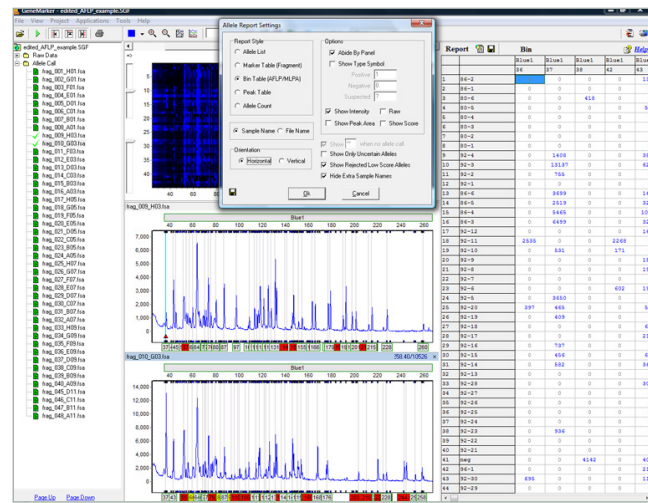
Basic Research Applications

AFLP® Analysis

GeneMarker is an efficient, user-friendly software tool designed for the analysis of data generated by AFLP® technology. The software is compatible with electrophoresis systems worldwide, as well as slab gel output. GeneMarker software features high efficiency allele calling, adjustable parameters and various reporting options including a trace comparison report. GeneMarker's unique sizing and pattern recognition technologies significantly improves analysis accuracy while providing greater analysis speed and less user intervention. GeneMarker's New Large Fragment Sizing Technology expands and reduces the cost of the AFLP technique by affording never before multiplexing opportunities.



Allele Report displaying peak intensities.

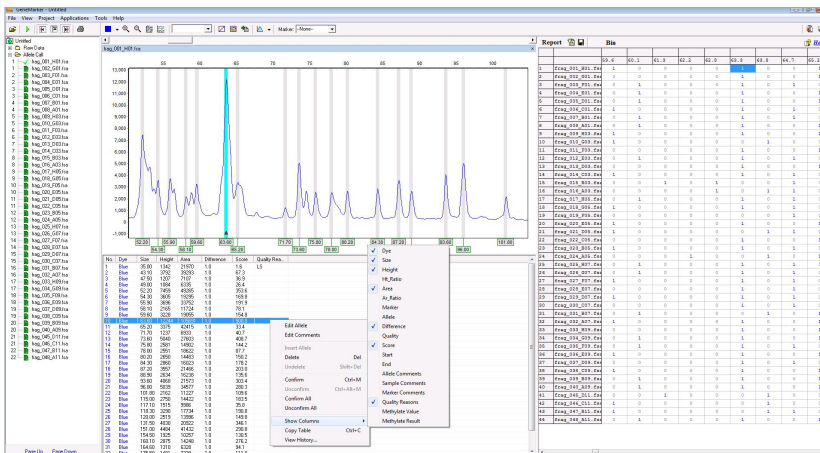


Report displaying presence and absence of alleles.

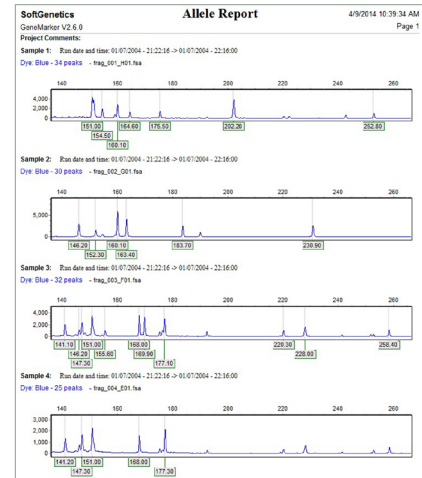
T-RFLP

(Terminal-Restriction Fragment Length Polymorphism) Analysis

Terminal-Restriction Fragment Length Polymorphism (T-RFLP) is a PCR based genetic fingerprinting technique for the study of microbial community structure. One of the primers of a primer pair is labeled with fluorescent dye and used to amplify a selected region of a gene of interest by PCR. The resulting PCR fragment is digested with one (or more) restriction endonuclease(s) and the Terminal Restriction Fragments (T-RFs) are separated with an automated DNA analyzer. Microbial diversity in a community can be estimated by analyzing the number and peak heights of T-RF patterns. GeneMarker software has the ability to produce highly sensitive and reproducible results for T-RFLP analysis by adjusting the AFLP Analysis default settings.



Peak table displayed below a sample electropherogram showing the option menu for editing items in the peak table (clicking the right mouse button with the cursor on a highlighted cell will activate the editing menu).

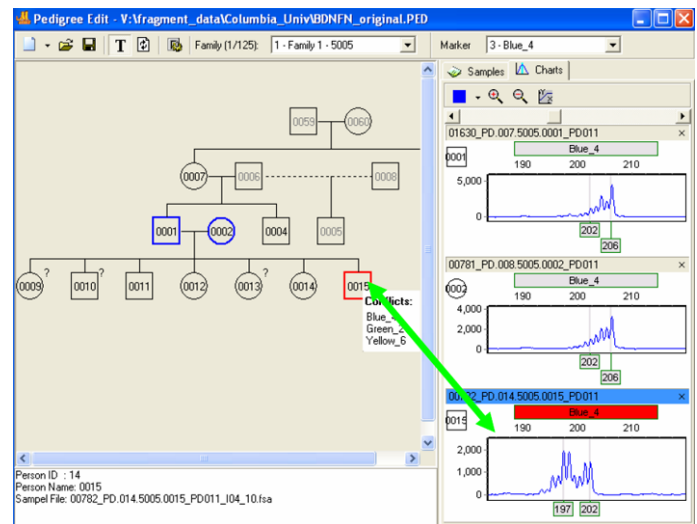


Print Report: Comparison of electropherograms from four samples.

Microsatellite Analysis and Linked Pedigree

GeneMarker decreases analysis set-up time through automated correction of common genotyping problems including saturated peaks, noisy data, bleed-through, instrument spikes and stutter peaks. GeneMarker's automated Run Wizard is designed to make analysis quick, easy, and accurate. The Data Analysis window features include:

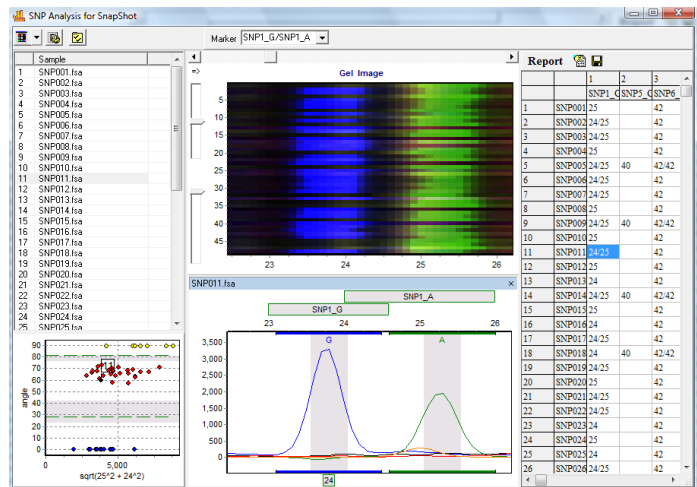
- 1. Saturation Correction:** Analysis of saturated data points by creating a synthetic peak based upon the peak shape before and after saturation.
- 2. Baseline Subtraction:** The software subtracts the baseline so that the Y axis is above the noise level.
- 3. Pull-up Correction:** This function removes peaks caused by bleed-through.
- 4. Spike Correction:** The software automatically removes peaks from voltage spikes caused by micro- air bubbles or debris in the laser path.
- 5. Stutter Peak Correction:** The software automatically filters for stutter peaks caused by PCR slippage.



Pedigree chart showing inheritance conflicts in Family 5005.

Single Base Extension/SnapShot®

GeneMarker software is an excellent tool to determine SNP genotypes is single base primer extension or SBE. An unlabeled primer with its 3' end directly flanking the SNP is extended one nucleotide by Taq polymerase and fluorescently-labeled ddNTPs complementary to the polymorphic base are added. The resulting fragment is one nucleotide longer, but the observed fragment size on a gel will be greater than expected due to the influence of the fluorescent dye on the electrophoretic mobility of these small fragments. SNPs can be identified by the one or two-color peaks associated with the incorporated labeled ddNTP and the length of the primer.

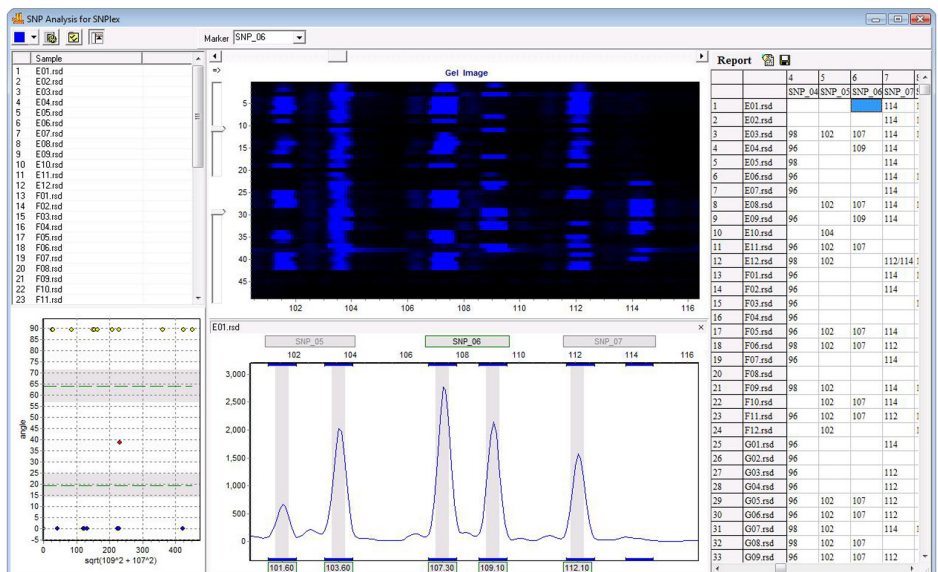


Single Base Extension/SnapShot® image.

SNPlex® & SNPWave® Analysis

One high-throughput method to determine SNP genotypes is SNPWave (Keygene® N.V.). SNPWave uses multiplex oligonucleotide ligation amplification of allele-specific probes coupled with AFLP®-primer selective amplification. SNPWave can detect up to 100 SNPs.

An alternate methodology is the SNPlex Genotyping System (Applied Biosystems) which can interrogate 48 SNPs simultaneously and has been used to investigate SNPs in 92 cancer-related genes in breast cancer and to genotype plants.



GeneMarker's SNPWave Report.

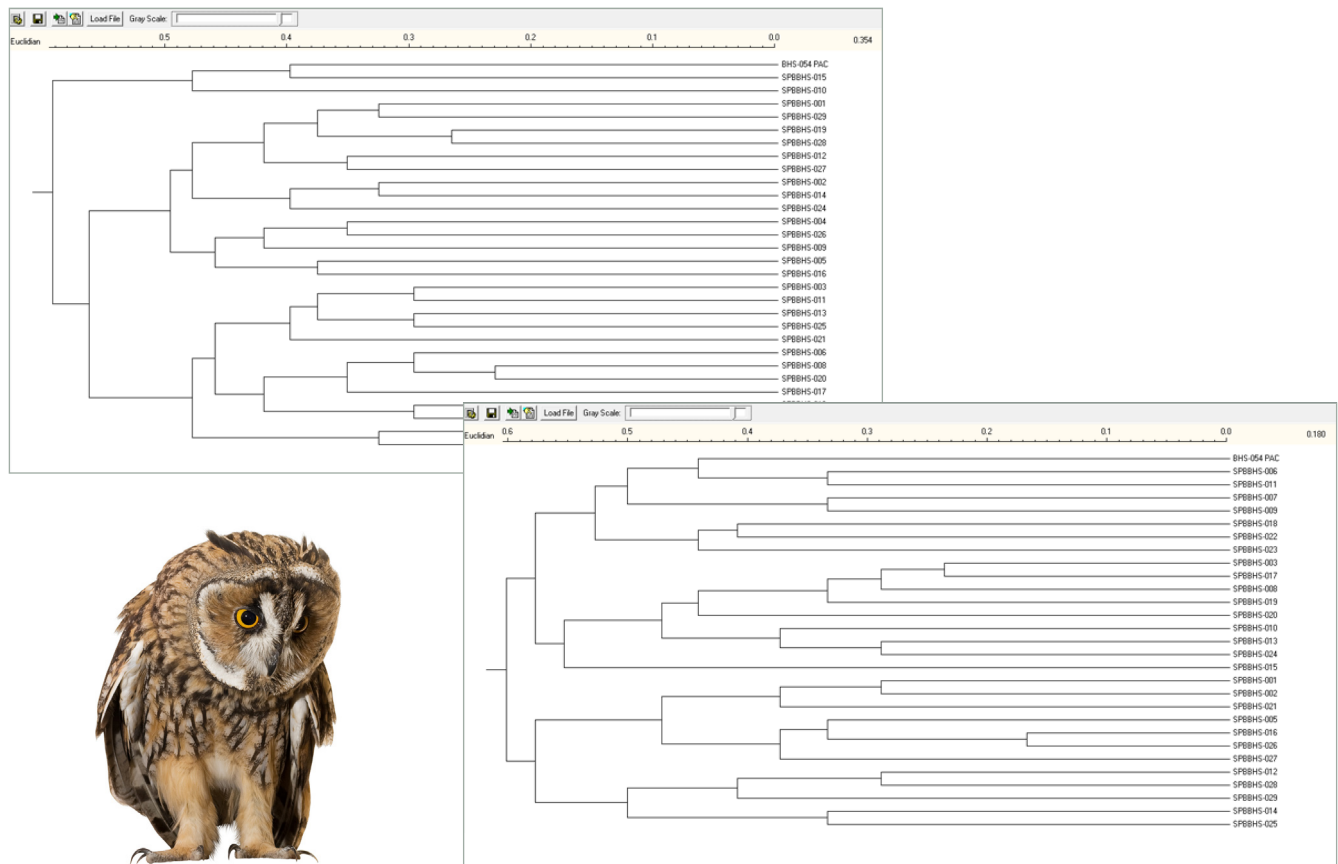
Phylogeny Clustering Analysis and the Merge Project Tool

Biological applications of data clustering calculations include phylogeny analysis and community comparisons in ecology, gene expression pattern, enzymatic pathway mapping, and functional gene family classification in the bioinformatics field. It has been successfully paired with the AFLP, microsatellite and RAPD analysis techniques for a variety of applications. GeneMarker uses the Hierarchical Clustering method, treating each data point as a single cluster and successively merges clusters until all points have been merged into a single remaining cluster. Researcher may select from different linkage types – Single, Complete or the Average – for the clustering algorithm. Results are presented as a dendrogram and a table providing Euclidian distances between each point.

Using GeneMarker's built in **Merge Project Tool**, the results of several multiplexes can be combined to create a complete genetic profile and enhance resolution.

	MAF65	MAF64	BM1222	BM848	OARCP0024	R19	BM1225	MAF209	BM4505	QanAE16	TGLA0387	PCB266	TGLA1222															
1 BHS-054 PAC	128	116	120	112	291	285	234	232	136	130	134	132	252	246	114	110	277	263	98	80	137	131	89	101	134	131		
2 SPBBHS-001	118	116	116	116	287	285	236	236	138	136	132	128	**	**	110	106	271	263	94	80	141	135	91	89	133	133		
3 SPBBHS-002	118	116	116	116	287	285	244	236	144	144	136	132	252	248	106	106	268	268	94	82	141	131	95	101	134	134		
4 SPBBHS-003	118	118	118	118	291	285	244	236	146	146	138	128	246	242	118	106	263	262	86	82	141	133	95	91	134	133		
5 SPBBHS-004	Disabled																											
6 SPBBHS-005	116	118	122	116	287	285	236	236	138	138	128	122	252	246	114	110	271	271	86	80	141	135	97	89	134	134		
7 SPBBHS-006	116	116	122	120	291	285	244	236	146	126	134	128	260	260	118	106	271	262	86	82	137	131	95	101	135	133		
8 SPBBHS-007	118	116	122	110	285	285	232	228	146	146	128	128	260	248	118	118	262	262	82	82	141	137	95	95	134	133		
9 SPBBHS-008	118	116	122	118	285	285	244	236	126	126	134	128	260	248	118	118	263	262	86	82	141	137	95	95	135	133		
10 SPBBHS-009	118	116	118	110	285	285	236	232	148	144	128	128	248	248	114	106	271	262	**	**	141	131	95	101	134	133		
11 SPBBHS-010	118	116	122	118	287	285	244	234	146	136	132	132	252	250	118	114	277	275	98	80	141	131	95	101	134	133		
12 SPBBHS-011	116	116	120	110	291	285	236	228	146	126	128	128	248	246	118	106	268	263	86	82	141	133	95	91	134	133		
13 SPBBHS-012	124	116	122	116	291	285	236	234	148	138	136	128	252	252	106	106	277	277	80	80	141	135	97	89	134	134		
14 SPBBHS-013	116	116	122	118	291	291	244	244	146	146	128	128	248	246	118	118	275	258	82	80	141	137	95	95	134	133		
15 SPBBHS-014	126	124	122	122	291	287	238	232	144	144	132	132	252	246	120	106	263	263	92	80	141	137	95	95	134	133		
16 SPBBHS-015	128	128	118	118	287	287	234	232	136	130	138	120	244	244	114	114	263	263	92	86	141	137	95	95	134	133		
17 SPBBHS-016	126	118	122	112	291	285	236	236	144	138	128	128	252	248	114	114	263	258	82	80	141	137	95	95	134	133		
18 SPBBHS-017	118	118	122	122	291	285	244	236	126	126	128	128	260	242	106	106	275	262	98	86	141	137	95	95	134	133		
19 SPBBHS-018	118	116	120	116	287	285	244	240	146	146	138	128	260	242	118	118	268	258	86	86	141	137	95	95	134	133		
20 SPBBHS-019	118	118	122	118	291	285	236	228	138	126	132	128	260	260	118	106	263	258	82	82	141	137	95	95	134	133		
21 SPBBHS-020	124	118	118	110	285	285	244	244	126	126	128	128	260	248	118	106	262	262	86	82	141	137	95	95	134	133		
22 SPBBHS-021	118	118	118	116	287	285	236	228	146	126	128	122	260	246	106	106	277	258	86	82	141	137	95	95	134	133		
23 SPBBHS-022	118	116	120	118	291	285	244	238	146	146	138	124	260	246	118	118	262	258	86	80	141	137	95	95	134	133		
24 SPBBHS-023	118	118	120	110	291	287	244	234	144	126	128	128	240	240	120	110	262	258	80	80	141	137	95	95	134	133		
25 SPBBHS-024	**	**	122	118	**	**	244	228	146	146	132	128	252	248	120	118	276	268	86	80	141	137	95	95	134	133		
26 SPBBHS-025	126	124	122	122	291	287	244	236	146	126	128	128	250	248	118	106	258	258	98	98	141	137	95	95	134	133		
27 SPBBHS-026	126	118	122	122	291	285	236	236	144	138	128	128	248	246	118	110	271	258	98	80	141	137	95	95	134	133		
28 SPBBHS-027	126	118	116	110	291	285	236	234	146	138	132	128	252	250	114	106	277	263	98	80	141	137	95	95	134	133		
29 SPBBHS-028	124	116	116	116	291	291	236	236	144	138	132	128	260	260	106	106	263	263	94	94	141	137	95	95	134	133		
30 SPBBHS-029	126	124	116	116	291	291	244	234	138	138	132	128	260	250	110	106	263	262	80	80	141	137	95	95	134	133		

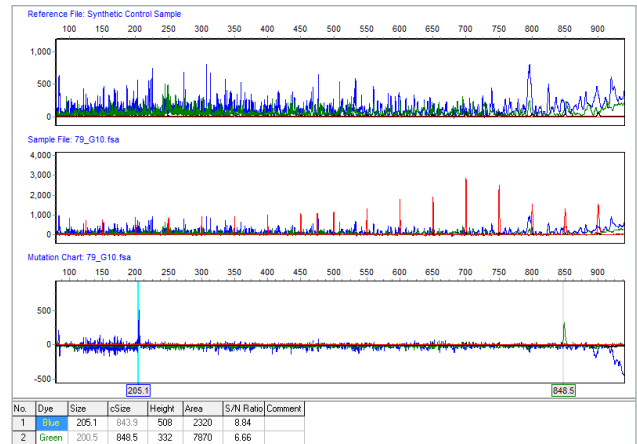
Three different multiplexes were used to amplify 30 DNA samples. Each multiplex contained primers for 4 or 5 independently assorting loci. The merge project tool in GeneMarker provided a single genotype for each individual with 14 markers. Allele drop out is indicated by **. The report style pictured here is a Marker Table suitable for further analysis in Relationship Testing or Kinship Analysis. Saving as a Peak Table would provide a spreadsheet with marker and allele name, fragment size (MW), peak height, height ratio, peak area, area ratio. The Bin Table format would be selected for further analysis using clustering algorithms for phylogeny.



Dendrogram from Cluster Analysis of 30 files using allele calls from one multiplex (left) and dendrogram of the same files based on the results of 3 multiplexes (right). Allele calls were combined using the Genotype Merge Tool prior to Cluster Analysis.

TILLING® & EcoTilling of Capillary and Slab gel data

The techniques of Targeted Induced Local Lesions In Genomes, or TILLING, and EcoTILLING have been widely used since 2000 to detect Single Nucleotide Polymorphisms (SNPs) in organisms ranging from Arabidopsis to zebrafish. The test samples may be experimentally mutagenized (ethylmethanesulfonate, radiation, etc) or from natural populations or derived from tumors or diseased tissues. When using GeneMarker's Tilling Application module, the size standard peaks are identified and all sample peaks are aligned. The peaks are smoothed, baseline is subtracted, and lane intensities are normalized. Low quality data is automatically rejected. A synthetic reference trace (Synthetic Control Sample) is constructed using median peak intensities from all of the high quality traces. This reference is subtracted from each sample trace generating a Mutation Chart that automatically identifies the sample's variations as shown in figure with the amplicon length of 1049bps.



The top panel shows the Synthetic Control Sample obtained from the median intensity after peak alignment. The middle panel shows sample 79. The bottom panel shows the Mutation Chart, generated by subtracting the reference from the sample, identifying individual variations. A blue peak at 205.2bps and a green peak at 848.8bps have been automatically identified. The original amplicon size is 1049bps.

Kinship Analysis of Wild Populations with Pedigree and Data Base Search Tool

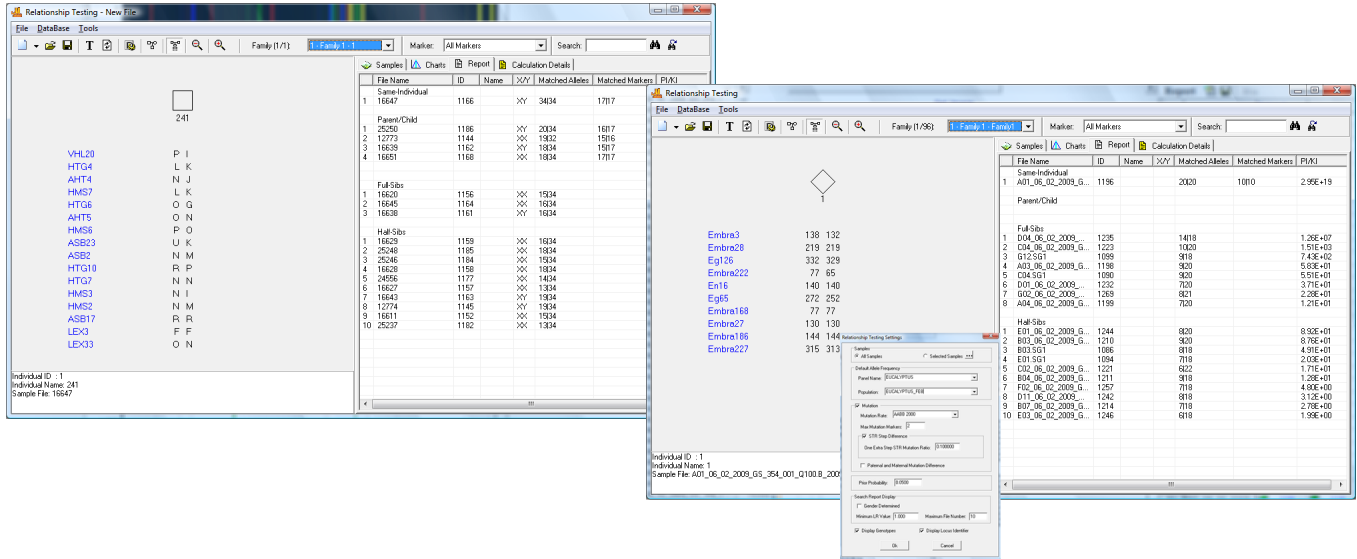
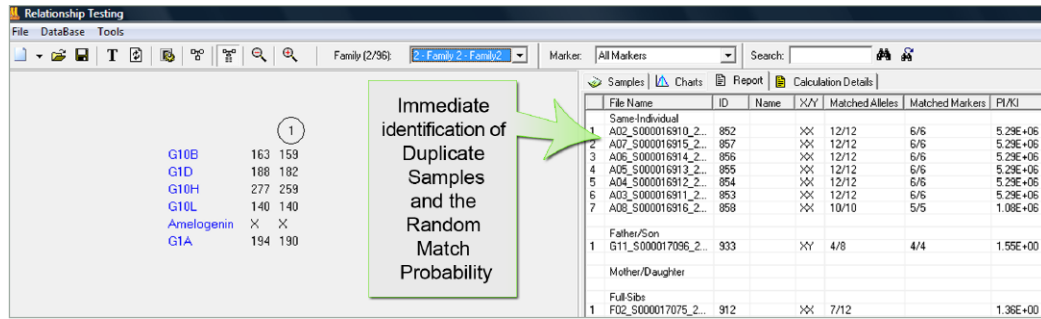
GeneMarker includes a first of its kind Kinship Analysis and database module which enables ecologists, and other wildlife researchers to easily identify kinships in natural populations, livestock breeding programs and aquaculture.

The "Kinship Analysis" tool provides a report table with probabilities and likelihood ratios across three generations for sample pairs. The rigorous statistical analysis to determine levels of kinship uses identity by descent (IBD) follows the methods of Brenner and uses stochastic matrices of Li and Sachs. GeneMarker database search tool identifies samples with the same genetic profile and calculates the random match probability (the probability that a randomly selected individual from a population will have an identical STR profile at the DNA markers tested). The "Find Family" tool searches the database and identifies files with the highest likelihood ratio for each relationship level to the experimental sample. Genetic Analysis Parameters allow setting tolerances for mistyping or mutation. The save to database function in GeneMarker can accept allele frequency tables for species specific markers and can accept previously archived genotype .txt or .cmf files, providing easy database updates.

Marker	Individual A			Individual B			Parent/Child (LR)	Full-Sibs (LR)	Half-Sibs (LR)	Cousins (LR)
Emb27	130			102			0.00000	0.25000	0.50000	0.75000
Emb186	144			163	181		0.00000	0.25000	0.50000	0.75000
Emb227	313	315		311	317		0.00408	0.25000	0.50000	0.75000
Emb28	219			208	219		2.56579	1.53289	1.78289	1.39145
Eg126	329	332		329	332		2.43779	3.59146	1.71890	1.35945
Emb222	65	77		65	77		8.89798	22.01826	4.94899	2.97450
En16	140			168			0.00000	0.25000	0.50000	0.75000
Emb3	132	138		132	138		27.87501	262.83277	14.43750	7.71875
Eg65	252	272		252	272		4.01515	9.83333	2.50758	1.75379
Emb168	77			77	79		1.96078	1.23039	1.48039	1.24020
Product Score:							0.00E+00	1.51E+03	5.08E+01	2.99E+01

Kinship Analysis Settings Provide Customization of Report Format.

The Database Search Tool

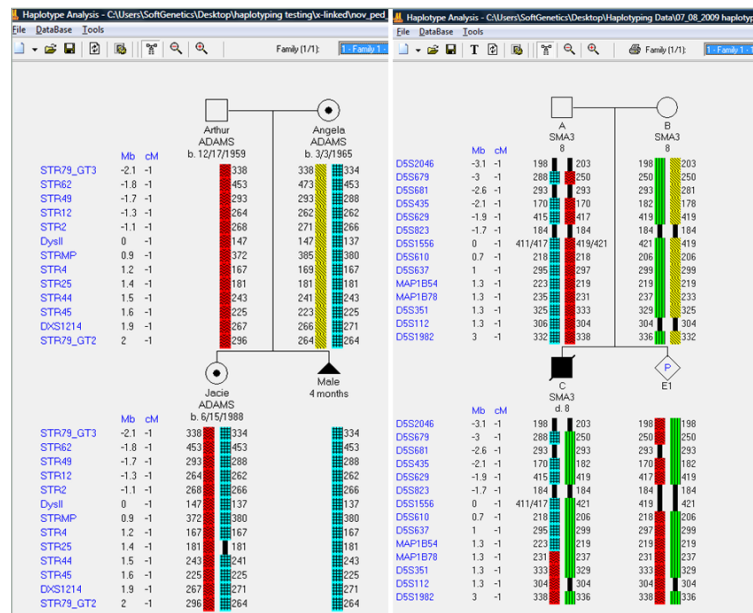


GeneMarker's Database Search Report: Report Settings to Customize Search for Animal or Plant Samples.

Haplotype Analysis

Familial DNA fragment data is used for haplotype analysis in areas such as genetic disorder research and preimplantation studies. In GeneMarker, the Haplotype Analysis application combines allele call information from multiple kits to obtain a complete genetic profile for each individual. The 'Family Group Tool' assists researchers by automating the process of drawing pedigrees. The software uses the allele calls of children and parents to assign a first-order-approximation phase of the alleles from the familial data. Whenever the alleles are informative for phase assignment a pattern/color bar is assigned to indicate most probable phase.

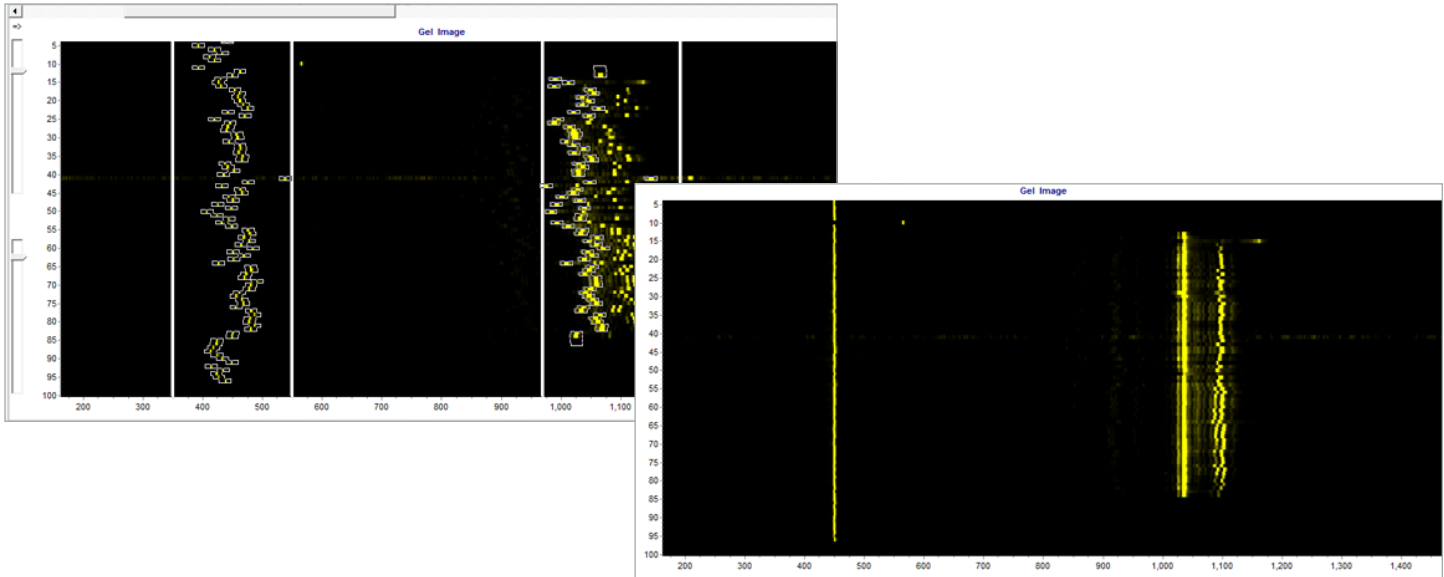
X-linked Example:
 Personal information
 Phase assignment
 Carrier status
 Standard symbols for male, female, Miscarriage (SAB)



Autosomal linkage Example:
 Phase assignment
 Cross-over in child 1
 Standard symbols for male, female, deceased, pregnancy

Macromolecule Analysis Tool

The Macromolecule Tool aids with analysis of macromolecules without an internal lane size standard. Depending on the macromolecule size and configuration, migration rates through capillary electrophoresis vary greatly. Internal size standards of the same macromolecule may not be readily available. This tool enables researchers to physically identify reference peaks known to have the same size and uses the information to calibrate from one capillary to another. Characteristics of the aligned data (such as relative size, peak height, peak area) can then be exported in an excel sheet or printed as an allele report.



*Use GeneMarker's synthetic gel image to draw a box around two size ranges (left), click apply and align the capillaries (right).
Use the run wizard to provide aligned electropherograms, relative size of peaks, peak height and peak area.*

Please open disc to review your applications of interest and to install a **30-day** trial of GeneMarker software.

Disc Contains:

Application Notes
30-day free GeneMarker software Trial
User Manual

Minimum Hardware Requirements

PC

32 or 64 bit Windows® OS: XP, Vista, 7 or 8
Processor: Pentium III, 1 GHz
RAM: 128MB
20GB Minimum available hard disk space.

Intel Powered Macintosh Computer

OS: 10.4.6, with Parallels desktop for MAC or
Apple Boot Camp
RAM: 2GB
20GB Minimum available on hard disk

If trial disc is not present please email info@softgnetics.com
for a free 30-day trial

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