

GeneMarker® is unique genotyping software with application oriented defaults which dramatically reduce analysis set-up time as well as error prone data transfer. Analysis is typically 3 clicks away from completion...PARAMETER SETTING...DATA SIZING...ANALYSIS REVIEW. The software's robust sizing and pattern recognition technology automatically removes chemistry and separation artifacts.

GeneMarker has been designed to provide genetic researchers with a "biologist friendly" genotyping tool. We incorporated the suggestions and requirements of several research groups into the software. Their main requirements were **ease-of-use, high accuracy, flexibility and low acquisition cost.**

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), SCF files, MegaBACE™ files (*.RSD, *.ESD), Beckman-Coulter files, and slab gel image files (TIFF,BIP,JPEG and TXT) from such systems as the LI-COR, DNA Analyzers and Kodak Image Station when used in combination with our JellMarker® software.

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

User Operation

The basic operation of GeneMarker automatically corrects for most instrument and chemistry errors, such as saturated peaks, noisy data, wavelength bleed-through, instrument spikes, and stutter peaks. GeneMarker's automated Run Wizard is designed to make repetitive analysis quick, easy, and accurate.

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's operation wizard simplifies parameter setting by "walking" the user through 3 simple steps. Templates can either be selected from several embedded templates or the user can create and save additional templates; detection settings are presented in one dialogue box that creates required flexibility in a simple to use format. Panel management and creation requires only a few mouse clicks! Once set the parameters can be saved and called back for future analysis.

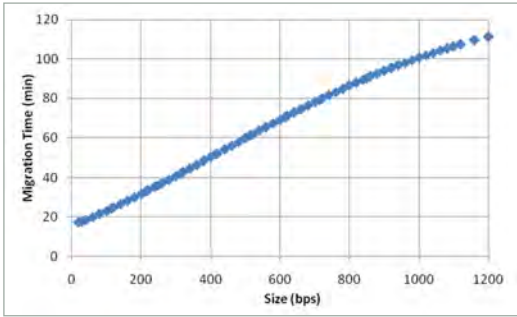
Sizing Technologies

As separation technology is now achieving fragments in excess of 1000 base pair, we have incorporated a new sizing technology in addition to the traditional Local Southern, Cubic Spline and third order least squares for the middle fragments. These older sizing methods have significant difficulties obtaining accurate size calls for data in excess of 800 bp, due to the non linear migration of larger DNA fragments.

Our "Large Size" sizing technology provides accurate, linear sizing using a DNA derivative migration time correction on fragments range from 30 to 1400bp, achieving resolution of 1base pair.

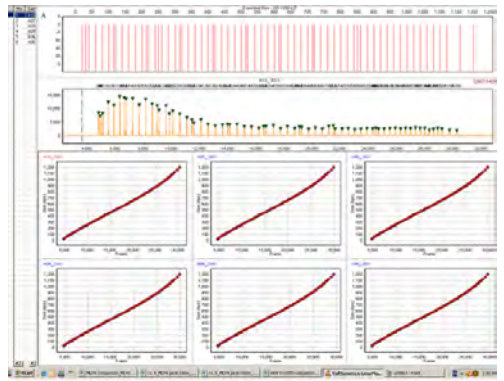
The "Large Size" technology affords many time, cost reducing multiplexing opportunities, such as the addition of 4x probes to clinical assays such as MLPA, Allele Specific Amplification (ARMS™), ASA, OLA, etc.) forensic profiling, and ecology analysis including TILLING®, STR/microsatellite profiles,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

This figure of GeneScan® 1200 in a POP7™ gel illustrates the typical non-linear DNA migration of large fragments. The migration time is linear for 100-800 bp fragments, but smaller and larger fragments deviate from the linear function. GeneMarker's new algorithm provides accurate, linear sizing of the data using a DNA derivative migration time correction to large DNA fragments. The new algorithm extends the linear range by over 50%, enabling accurate sizing of fragments from 30 – 1400 bps.

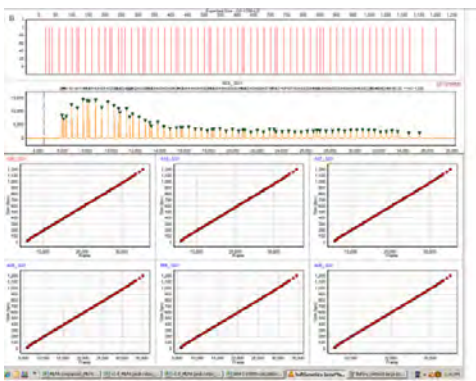


Comparison of the linear range for DNA fragments before with Local Southern (80-800 bps) and after with GeneMarker's Large Size Technology. The migration times are almost a perfect linear function to the DNA fragment size after applying the Large Size call in GeneMarker.

Local Southern Algorithm has a limited linear range



New GeneMarker Algorithm provides linear range from 20–1200 bps

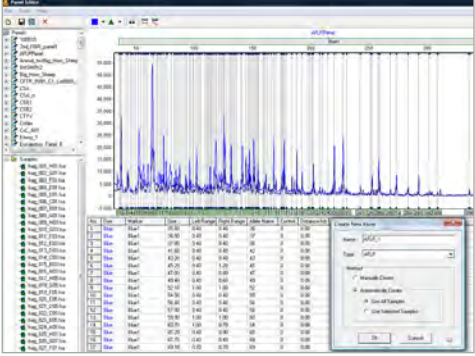


Pattern Recognition Technology

GeneMarker incorporates the latest pattern recognition technology in order to remove sequencing artifacts and other separation/chemistry errors. Automated removal of artifacts eliminates the need for time-consuming analysis editing prevalent with other genotyping analysis programs.

Easy-to-use Panel Editor

Panel templates 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 from GeneMapper. Custom panels are easily created in the panel editor. Users can automatically create custom panels using the signal information from data files or manually: by simply inserting the desired alleles using the mouse.



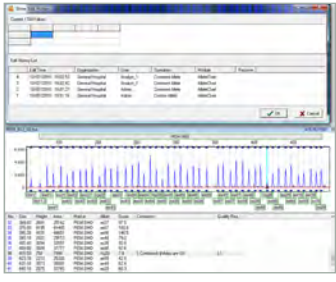
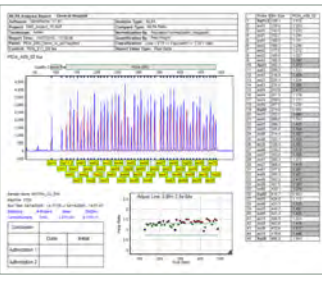
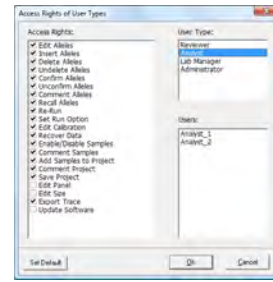
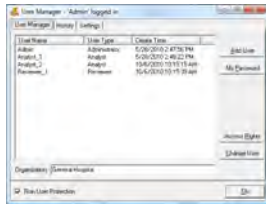
Exclusive Tools Speed and Protect Analysis

GeneMarker software includes many tools designed to assist the quality and speed of analysis. Analysis results are linked to analysis electropherograms, navigation is a simple process of moving your mouse across the electropherograms, or drawing a box around an area of interest to zoom in or out for in-depth review. GeneMarker software includes a user control function that password protects and limits program access, while also providing an audit trail of edits and comments.

User Management and Audit Trail

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

Login Record Access Rights Control Activate Data Edit History



Header has Institution and User ID

User Management records and save edit history and comments

Genotype Merge Tool

Obtaining a complete genetic profile for wildlife and plant research or medical research is often complicated by overlapping marker ranges and/or incompatible chemistry; making it necessary to amplify the same samples multiple times using different sets of markers or locus specific primers. Traditionally, researchers export the genotyping results from several multiplexes into a spread sheet and manually combine allele calls for each individual.

GeneMarker's Genotype Merge Tool addresses this problem. Researchers can conveniently combine two or more GeneMarker projects (each project using a panel containing a unique set of markers or loci) into a single, comprehensive report. This report represents a single view of multiplexes (a super panel) of markers/loci from the individual projects, providing a complete genotype for each sample. This merged report may be saved in a spreadsheet as a genome-wide genotype, or imported into other special applications, such as, Clustering Analysis or Relationship Testing and Kinship Analysis, improving the robustness of these analyses by including information from a greater number of markers.

Sample	Marker1	Marker2	Marker3	Marker4	Marker5	Marker6	Marker7	Marker8	Marker9	Marker10	Marker11	Marker12	Marker13	Marker14	Marker15	Marker16	Marker17	Marker18	Marker19	Marker20	Marker21	Marker22	Marker23	Marker24	Marker25	Marker26	Marker27	Marker28	Marker29	Marker30
Sample1	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110
Sample2	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110
Sample3	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110
Sample4	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110
Sample5	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110
Sample6	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110
Sample7	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110
Sample8	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110
Sample9	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110
Sample10	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110
Sample11	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110
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Sample15	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110
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Sample17	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110
Sample18	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110
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Sample30	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110	110

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 spread sheet 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.

Project Comparison Tool

The Project Comparison tool provides two useful functions. First, it can be used to compare two independent analysts' analyses. Second, it can be used as a validation tool to determine differences in allele calls based on analysis parameters or instrument runs. Replicate 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.

SOFTGENETICS®

Software PowerTools for Genetics Analysis

GeneMarker®

The Biologist Friendly Software

Unique Tools to Increase Genotyping Speed and Accuracy

Compatible with all Major Sequencing Systems

Embedded Applications Eliminate Data Transfer

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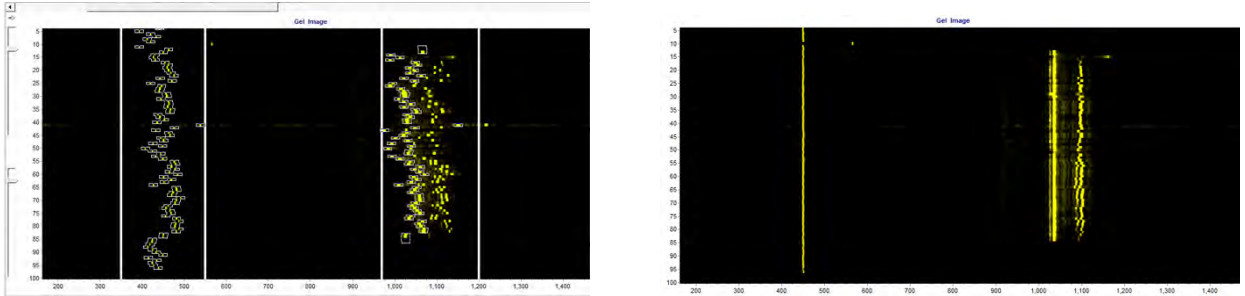
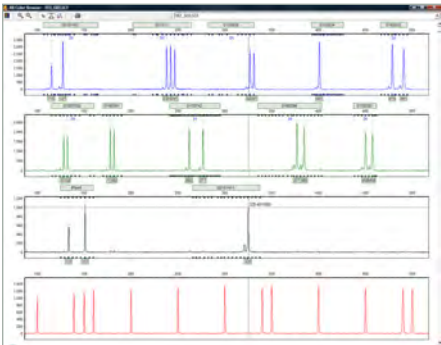
Software PowerTools for Genetics Analysis

All Color Browser

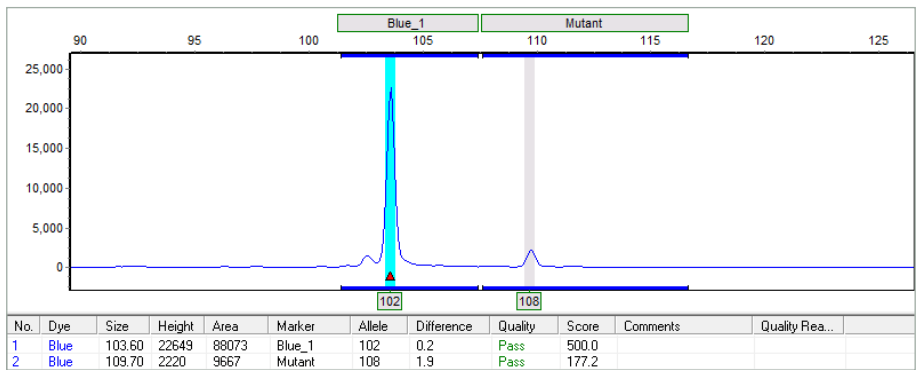
The All Color Browser provides a complete 4 or 5 color review of an analysis. Edits or comments made in this view are automatically recorded in the electropherogram, allele report and peak table of the main analysis window and are saved when the project is saved.

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.

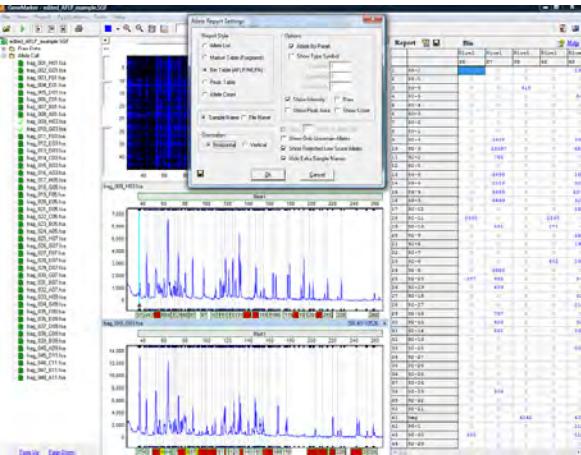


Applications and Reporting

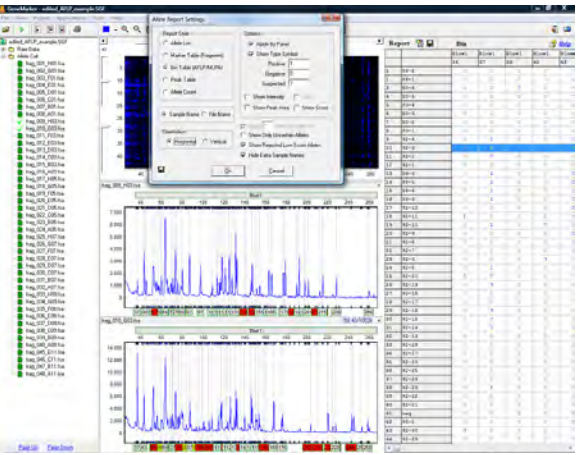
GeneMarker includes several embedded applications that provide analysis of key applications in a single software package eliminating the need to transfer data from a genotyping program to spread sheets or other programs. Most of these applications include a specialized customizable report formats.

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. The 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



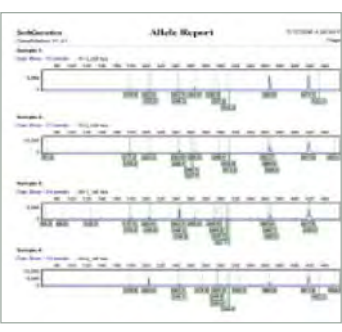
Report displaying presence and absence of alleles

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 (only one sample from a duplicate pair is shown).

Microsatellite Analysis with Linked Pedigree

GeneMarker decreases analysis set-up time through automated correction of common genotyping problems including saturated peaks, noisy data, wavelength bleeding, 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:

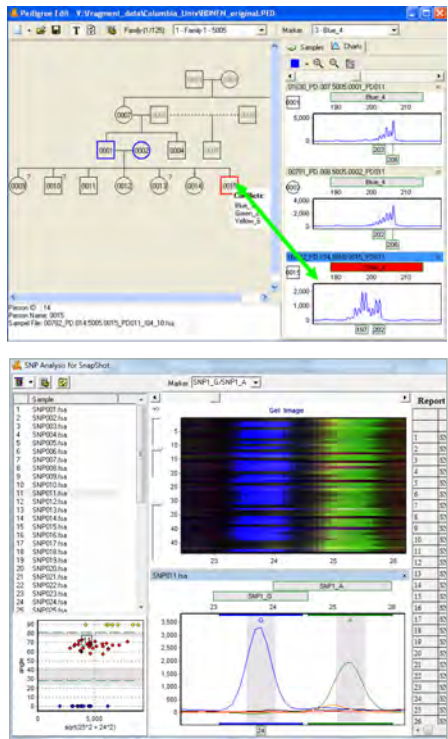
1. Saturation Correction: Analysis of saturated data points by creating a synthetic peak based upon the peak shape before and after saturation.
2. Baseline Subtraction: The software removes the baseline so that the Y axis is above the noise level.
3. Pull-up Correction: This function removes peaks caused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

Linked Pedigree Tool

The user can either open an existing pedigree file of type ped or pre or create a new pedigree file in GeneMarker's pedigree tool. The pedigree chart is designed to aid identification of inheritance patterns and abnormalities. All individuals in the pedigree with sample files are directly linked to the corresponding electropherograms with a mouse click, and individuals with illogical or abnormal allele calls are highlighted in red.

The link between the pedigree and the electropherograms displaying allele calls for each marker makes analysis quick and efficient.

Pedigree chart showing inheritance conflicts in Family 5005



Single Base Extension/SnapShot® image

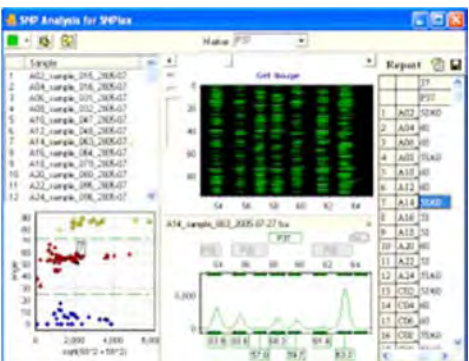
Single Base Extension/SnapShot®

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.

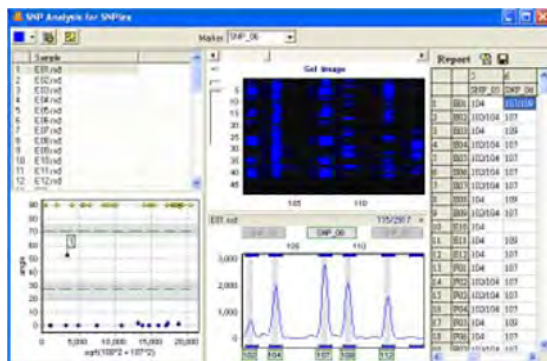
SNPlex® & SNPWave® Analysis

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.



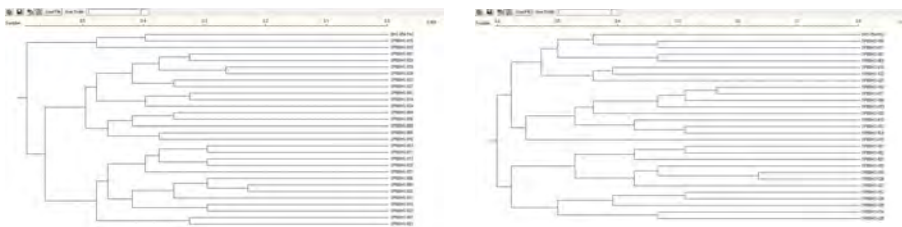
GeneMarker's SNPlex Report



GeneMarker's SNPWave Report

Phylogeny Clustering Analysis

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.

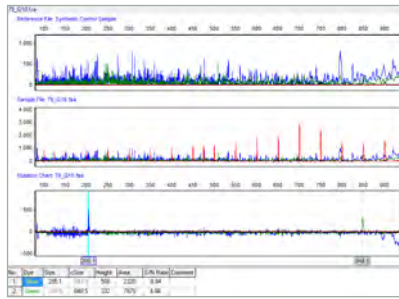


Dendrogram from Cluster Analysis of 30 files using allele calls from one multiplex (left) and dendrogram of the same files based on the results of 3 multiplexes (right). Allele calls were combined using the Genotype Merge Tool prior to Cluster Analysis.

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.

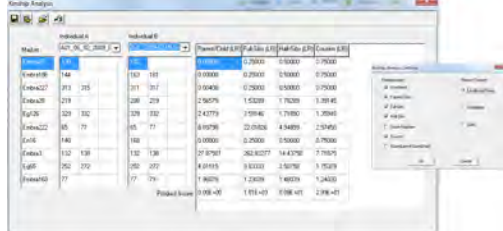


Kinship Analysis of Wild Populations and Data Base Search Tool

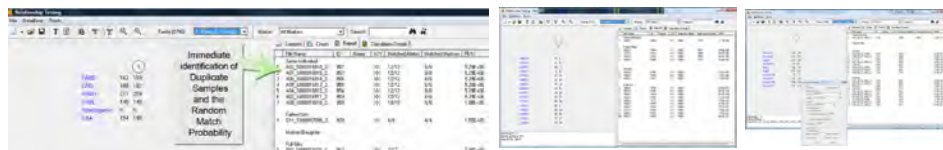
GeneMarker now includes a first of its kind Kinship Analysis and database module which enables ecologists, and other wildlife researchers to easily identify kinships in natural populations, livestock breeding programs and aquaculture.

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 STR 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.



Kinship Analysis Settings Provide Customization of Report Format

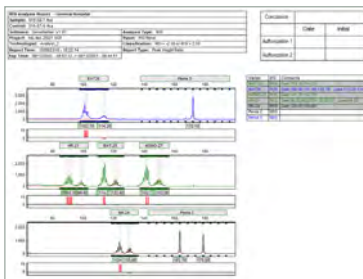


GeneMarker's Database Search Report: Report Settings to Customize Search for Animal or Plant Samples

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.

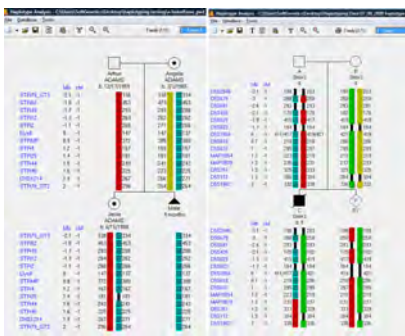
In GeneMarker's MSI Analysis module, tumor samples are compared to normal samples based on peak-to-peak comparison. Differences between the two traces are displayed in a Trace Comparison Histogram below each electropherogram. Within the electropherogram, the tumor sample trace is overlaid on a light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



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.

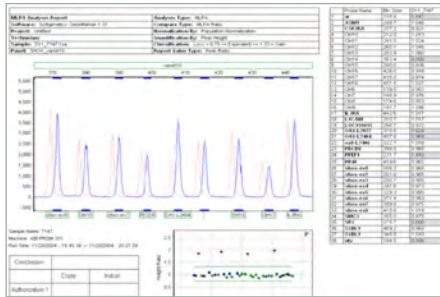
X-linked Example: Personal information Phase assignment Carrier status Standard symbols for male, female, Miscarriage (SAB)



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

MLPA® Analysis

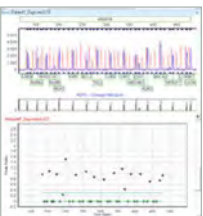
Multiplex Ligation-dependent Probe Amplification is a simple method for simultaneous quantification of up to 45 nucleic acid sequences in a single reaction. Amplification products are separated by sequence gel electrophoresis, and MLPA® probes are able to distinguish between sequences that differ in only one base pair. Although the technique is efficient, inexpensive, and simple, there is a lack of an integrated software analysis package that performs data collection, normalization and patient reporting. GeneMarker's MLPA® function integrates all the analysis steps into a single convenient package. Due to the variations of PCR efficiencies from small to large DNA fragments or from sample to sample, two selectable normalization methods are provided. The first normalization method is the traditional method based upon the control probes. The second method, Population Normalization, unique to GeneMarker, normalizes peak intensities based upon the statistically most probable median providing a more robust analysis when a large number of mutation probes are used. The software features pre-made panels based on the MRC Holland probe sets. These pre-made panels can be imported into the panel editor and will need minor adjustments to fit your experimental data.



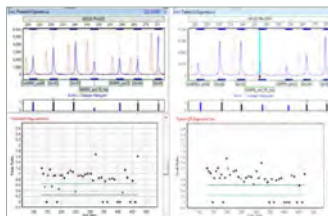
Customizable Patient Report available as a .PDF or printed file.

MS-MLPA®

GeneMarker DNA analysis software has been successfully paired with the Multiplex Ligation-dependent Probe Amplification technique for detecting genetic deletions and duplications in various diseases including cancer. Recently, 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 new 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.



Methylation of the promoter region of tumor suppressor gene ESR1 is indicated by the red plot point that appears above the methylation threshold line.



The image on the left is a normal patient. Five genes of interest (methylation specific) from the PWS/AS region of chromosome 15 appear in the "Normal" region between the methylation detection threshold lines. The image on the right is a sample from a PWS patient. The five genes of interest are in a 1:1 ratio with the reference trace indicating methylation at these five sites (one copy of 15q11.2 and one copy of 15q12 have been deleted).

Trisomy Analysis

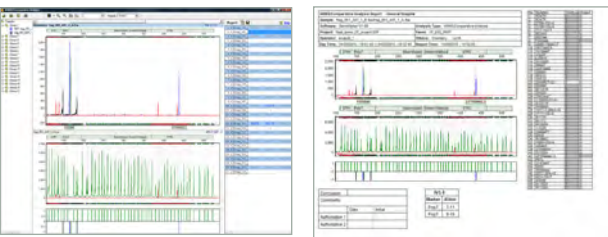
GeneMarker aids technicians and researchers analyzing QF-PCR products to detect aneuploidy with highly accurate size calling and a user-friendly interface. The trisomy application is directly linked to the main analysis – there is no need for error prone data transfer. Analysts may select the default 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™, Finnzymes™ and custom chemistries.

Patient Report: Highlighted samples are consistent with user specified triallelic range and samples marked with a '?' are consistent with user specified intermediate (inconclusive) ranges.



Cystic Fibrosis Analysis

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 GenProbe® 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.



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

The Highlight Abnormal Allele option

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.

