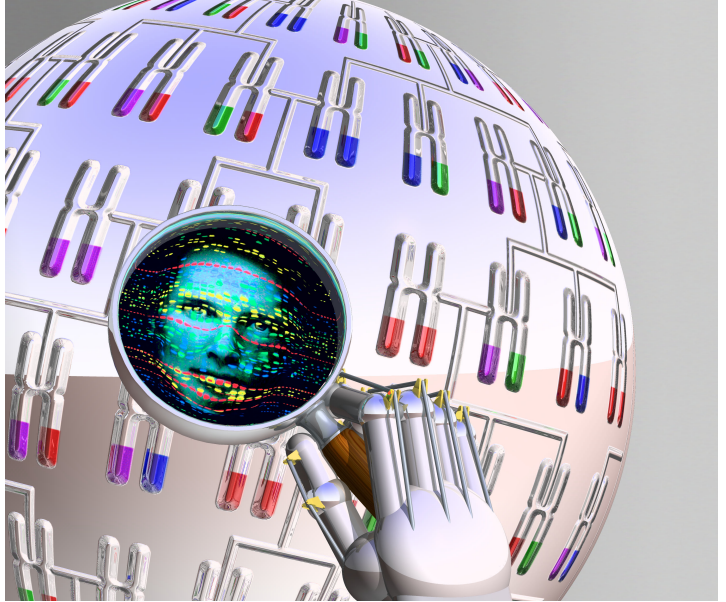




GeneMapper™ *ID* Software Version 3.1

Human Identification Analysis

User Guide

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GeneMapper *ID* Software version 3.1 has undergone a verification process defined by Applied Biosystems. However, human identification laboratories analyzing forensic, paternity, databasing, and single-source samples that choose to use GeneMapper *ID* Software for data analysis should perform their own appropriate validation studies.

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Preface

How to Use This Guide

Purpose of This Guide The *GeneMapper™ ID Software Version 3.1 User Guide* provides reference information about the GeneMapper *ID* software for general use and customization. For specific step-by-step instructions on how to use the software, refer to the *GeneMapper™ ID Software v3.1 Human Identification Analysis Tutorial* (PN 4335523).

Audience This guide is intended for GeneMapper *ID* software users.

Assumptions This guide assumes that you have read all applicable material and installed the GeneMapper *ID* Software Version 3.1 according to the installation instructions included in the software kit.

This guide uses conventions and terminology that assume a working knowledge of the Windows™ operating system, the Internet, and Internet-based browsers.

Text Conventions This guide uses the following conventions:

- **Bold** indicates user action. For example:
Type **0**, then press **Enter** for each of the remaining fields.
- *Italic* text indicates new or important words and is also used for emphasis. For example:
Before analyzing, *always* select the appropriate size standard.
- A right arrow bracket (>) separates successive commands you select from a drop-down or shortcut menu. For example:
Select **File** > **Open** > **Project**.

User Attention Words

Two user attention words appear in Applied Biosystems user documentation. Each word implies a particular level of observation or action as described below:

Note: Provides information that may be of interest or help but is not critical to the use of the product.

IMPORTANT! Provides information that is necessary for proper use of the software, instrument operation, accurate chemistry kit use, or safe use of a chemical.

How to Obtain More Information

Related Documentation

This GeneMapper™ *ID* Software Version 3.1 User Guide describes the GeneMapper™ *ID* Software Version 3.1, and provides information about the GeneMapper *ID* software for general use and customization. The software comes with additional documentation to assist with installation and provide recommended workflows for specific genotyping applications.

The following are related documents:

- *GeneMapper™ ID Software v3.1 Human Identification Analysis Tutorial* (PN 4335523)
- *Microsatellite Analysis with ABI PRISM® GeneMapper™ Software v3.0 Tutorial* (PN 4335525)
- *SNP Genotyping with ABI PRISM® GeneMapper™ Software v3.0 Tutorial* (PN 4335524)
- *Installation ReadMe.txt file* (located on the software CD)

IMPORTANT! You must read the installation instructions and the ReadMe file before installing the software!

Send Us Your Comments

Applied Biosystems welcomes your comments and suggestions for improving its user documents. You can e-mail your comments to:

techpubs@appliedbiosystems.com

How to Obtain Support

Applied Biosystems Web Site

For the latest support information for all locations, go to **<http://www.appliedbiosystems.com>**, then click the link for **Support**.

At the Support page, you can:

- Search through frequently asked questions (FAQs)
- Submit a question directly to Technical Support
- Order Applied Biosystems user documents, MSDSs, certificates of analysis, and other related documents
- Download PDF documents
- Obtain information about customer training
- Download software updates and patches

In addition, the Support page provides access to worldwide telephone and fax numbers to contact Applied Biosystems Technical Support and Sales facilities.

About GeneMapper™ *ID* Software

1

This chapter provides an overview of the GeneMapper™ *ID* Software Version 3.1 software features, supported chemistry kits, and this user guide.

This chapter covers:

Overview of GeneMapper <i>ID</i> Software	1-2
Steps Required to Use GeneMapper <i>ID</i> Software.	1-5
Organization of the User Guide.	1-6

Overview of GeneMapper ID Software

GeneMapper ID Software Version 3.1 is an automated genotyping software solution for forensic, paternity, and database data analysis and other genotyping needs. The GeneMapper ID software combines functions of the GeneScan® Analysis Software and the Genotyper® Software, and it includes additional features and enhancements.

The GeneMapper ID Software Version 3.1 is designed specifically for AmpFISTR data analysis, but it can perform other microsatellite and SNP analysis, and this user guide includes information needed for microsatellite and SNP analysis. To obtain more information about the initial setup and workflow specific to a particular application, refer to one of the three tutorials listed in [“Related Documentation”](#) on [page x](#).

The GeneMapper ID Software Version 3.1 has not undergone specific developmental validation for human identification (HID) applications. Human identification laboratories that choose to use GeneMapper ID Software for data analysis should perform their own developmental verification studies.

Supported Instruments

The GeneMapper ID software v3.1 supports data generated from the following instruments:

- ABI PRISM® 310 Genetic Analyzer
- ABI PRISM® 377 DNA Sequencer
- ABI PRISM® 3100-*Avant* Genetic Analyzer
- ABI PRISM® 3100 Genetic Analyzer

Note: Data generated from a 310 Genetic Analyzer or 377 DNA Sequencer using a Macintosh® platform must be converted to a Microsoft® Windows®-based format before it can be used with the GeneMapper ID software. Refer to [Appendix E, “Converting Macintosh® Sample Files.”](#)

**Supported
Chemistry Kits**

The GeneMapper *ID* software v3.1 is specifically designed to work with the following AmpF $\mathbb{L}$ STR kits and reagents:

- AmpF $\mathbb{L}$ STR $^{\text{®}}$ Identifiler $^{\text{®}}$ PCR Amplification Kit
- AmpF $\mathbb{L}$ STR $^{\text{®}}$ SGM Plus $^{\text{®}}$ PCR Amplification Kit
- AmpF $\mathbb{L}$ STR $^{\text{®}}$ Profiler Plus $^{\text{®}}$ ID PCR Amplification Kit
- AmpF $\mathbb{L}$ STR $^{\text{®}}$ Profiler Plus $^{\text{®}}$ PCR Amplification Kit
- AmpF $\mathbb{L}$ STR $^{\text{®}}$ COfiler $^{\text{®}}$ PCR Amplification Kit
- AmpF $\mathbb{L}$ STR $^{\text{®}}$ SEfiler $^{\text{®}}$ PCR Amplification Kit
- AmpF $\mathbb{L}$ STR $^{\text{®}}$ Profiler $^{\text{®}}$ PCR Amplification Kit
- AmpF $\mathbb{L}$ STR $^{\text{®}}$ Blue $^{\text{™}}$ PCR Amplification Kit
- AmpF $\mathbb{L}$ STR $^{\text{®}}$ Green $^{\text{™}}$ I PCR Amplification Kit

Unique Features of GeneMapper ID Software v3.1

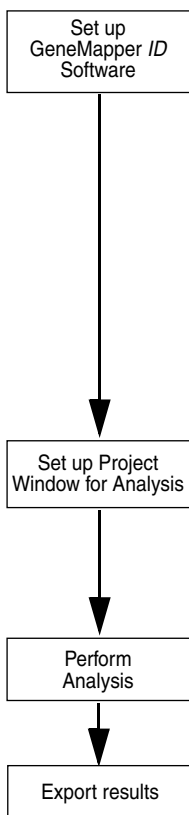
GeneMapper ID software v3.1 includes the following unique features:

Unique Feature	Description
CODIS Export	The software can export results in a CODIS recognized format (cmf v1.0 and 3.0).
Process Component-Based Quality Values (PQV) (See Appendix A for more information.)	The PQV system automatically assigns values to the quality of the data in respect to sizing and allele calling. Poor quality samples are those below the user defined thresholds.
GeneMapper ID Database Note: The database does not store individual sample files.	The GeneMapper ID database stores the following data: • Predefined and custom designed size standard definitions • Panel, marker (loci), and allele bin definitions • Analysis methods • Table profiles (for generating tabular reports) • Saved projects with sizing and genotyping results • Matrix files (310 and 377 instruments only) • Plot settings
Automated concordance checks	• GeneMapper ID software compares genotype concordance between overlapping loci among different AmpFℓSTR kits for the same sample(s), or concordance of genotype calls from duplicate amplifications or duplicate injections of the same sample. • Positive and negative controls give the expected allele calls.

Steps Required to Use GeneMapper ID Software

Using GeneMapper ID Software

The following steps are required to use the GeneMapper ID software. You will be guided through this process in more detail by reading the *GeneMapper™ ID Software v3.1 Human Identification Analysis Tutorial* (P/N 4335523), *Microsatellite Analysis with ABI PRISM® GeneMapper™ Software v3.0 Tutorial* (P/N 4335525), and *SNP Genotyping with ABI PRISM® GeneMapper™ Software v3.0 Tutorial* (P/N 4335524).



- Set up the GeneMapper ID software as described below.
 - Import panels and bins in the Panel Manager.
 - Create an analysis method suitable for your application with appropriate bin sets.
 - Define custom views of analysis tables and plots.
 - View and set HID analysis options.
 - Create the appropriate size standard(s) if your data uses size standards other than those provided with GeneMapper ID software.
 - Convert any Macintosh®-generated GeneScan® sample files to the .fsa format. (See [Appendix E, “Converting Macintosh® Sample Files,”](#) for details.)
- Set up the Project window for analysis of a given set of data:
 - Import the fragment analysis sample files.
 - Select the appropriate analysis method.
 - Choose the appropriate panels.
 - Select the size standard.
- Perform analysis and examine the results:
 - Initiate analysis.
 - Examine analysis results using the Plot window(s).
- Export results:
 - Set up table format for export (using Table Settings Editor).
 - Export (using Export Table command).

Organization of the User Guide

This manual provides technical information to support the GeneMapper ID software user.

Chapters The following chapters explain how to use the major application components:

- [Chapter 2, “Getting Started with GeneMapper ID Software,”](#) describes hardware requirements and procedures for installing, registering, and logging in and out of the GeneMapper™ ID software v3.1.
- [Chapter 3, “Using the Project Window,”](#) describes how to use the main Project window and associated utility windows.
- [Chapter 4, “Using Panel Manager,”](#) describes how to use the Panel Manager to manage all of the chemistry kits, panels, markers (loci), and bin definitions required for automated allele calling.
- [Chapter 5, “Using GeneMapper Manager,”](#) describes how to use the:
 - Projects tab
 - Analysis Methods tab to create custom analysis methods for use in analyzing sample files
 - Table Settings tab to create new profiles, hide and show table columns, and filter the table entries (rows) in Project windows
 - Plot Settings tab to create a custom plot profile for viewing data
 - Matrices tab to create a matrix file
 - Size Standards tab to create a size standard
- [Chapter 6, “Options Dialog Box,”](#) describes how to set up default analysis settings, customize analysis settings, and create new users and passwords.
- [Chapter 7, “Analyzing and Interpreting Data,”](#) describes how to
 - [Set HID analysis options for analysis, interpreting sample PQVs, sizing quality, and precision](#)
 - [Use the Size Match Editor to examine size standard electropherograms, edit the identification of size standard peaks, and view size calling curves](#)

- View Genotypes table, interpreting genotype PQVs and genotype quality
- Perform concordance checks
- Chapter 8, “[Using Plot Windows - Samples and Genotypes](#),” describes how to examine and interpret electropherograms within the samples and genotypes plot windows.
- Chapter 9, “[Exporting Table Data](#),” describes how to use the export and CODIS export features.
- [Chapter 10, “Printing Data,”](#) describes how to print data.

Appendices The following appendices contain reference information:

- Appendix A, “[Process Quality Values](#),” explains how to use the Process Component-Based Quality Values.
- Appendix B, “[Software Genotyping Algorithms](#),” provides a description of the genotyping algorithms used in GeneMapper *ID* software.
- Appendix C, “[Project Window Software Interface](#),” describes the dialog boxes and menu commands associated with the Project Window.
- Appendix D, “[GeneScan Size Standards](#),” provides information on the defined size standards provided with the GeneMapper *ID* software.
- Appendix E, “[Converting Macintosh® Sample Files](#),” describes how to use the Macintosh® AppleScript® program to convert Macintosh-generated fragment analysis sample files to the Windows format.
- [Appendix F, “Transferring Data and Backing Up Data,”](#) describes how to transfer projects, supporting files and data to a different computer running GeneMapper *ID* software.
- [Appendix G, “Sample Sheets and Plate Records.”](#)
- [Appendix H, “Troubleshooting Sheet,”](#)
- [Appendix I, “GeneMapper ID Software Verification,”](#) describes verification testing.
- [Appendix J, “Software Warranty,”](#) describes the Applied Biosystems warranty that comes with the software package.
- [Appendix K, “Workflow for Storing Sample Files, Projects, and Second Reads.”](#)

Getting Started with GeneMapper *ID* Software

2

This chapter describes hardware requirements and procedures for installing, registering, and logging in and out of the GeneMapper™ *ID* software v3.1.

This chapter covers:

Hardware Requirements	2-2
Installing the GeneMapper <i>ID</i> Software	2-3
Registering GeneMapper <i>ID</i> Software	2-6
Logging Onto and Logging Out of the GeneMapper <i>ID</i> Software .	2-7

Hardware Requirements

Hardware Requirements The following table describes the components your computer system requires to run GeneMapper *ID* software.

System Component	Minimum Requirement	Recommended
Computer	• Intel® Pentium® III processor minimum of 540 MHz • 256 MB RAM • 6 GB hard disk (free space)	• Intel Pentium III processor faster than 700 MHz • 512 MB RAM • 10 GB EIDE hard drive (6 GB free space) • 20/48X IDE CD-ROM • 10/100 NIC with RWV (internal)
Monitor	800 x 600 pixels size 17-inch monitor	1024 x 768 pixels size 19-inch or larger monitor
Operating System	Microsoft® Windows NT® v.4.0 (Service Pack 5)	Microsoft Windows® 2000 Professional (Service Pack 3) or Windows NT v.4.0 (Service Pack 5)
Ethernet Capability	Network card for Oracle® installation. TCP/IP must be installed prior to Oracle installation.	Network card for Oracle® installation. TCP/IP must be installed prior to Oracle installation.

In order to perform all functions in GeneMapper *ID* software (with its embedded Oracle database), ensure there are at least 500 MB of free space on the partition where the GeneMapper *ID* database is installed. It is possible to export your projects to a different drive or partition, then delete those projects from the database to make room for new projects. (See [Chapter 3](#), “Using the Project Window.”)

Installing the GeneMapper *ID* Software

Compatibility with Oracle Databases

GeneMapper *ID* software uses an Oracle® database. GeneMapper *ID* software v3.1 is compatible only with the Oracle database installed with ABI PRISM® 3100/3100-*Avant* Data Collection software v2.0.

Special Considerations

The following is a list of special considerations for installing the GeneMapper *ID* software v3.1.

- Compatibility of GeneMapper *ID* Software v3.1 with ABI PRISM® 3100/3100-*Avant* Genetic Analyzer systems:

3100 Data Collection Version	3100- <i>Avant</i> Data Collection Version	Compatible with Instrument Computer?
1.1	–	No, use a separate computer that meets the minimum requirements ^a
–	1.0	
2.0	2.0	Yes

a. GeneMapper *ID* software v3.1 cannot exist with other Oracle servers, except the Oracle database installed by 3100/3100-*Avant* Data Collection version 2.0.

IMPORTANT! To properly install GeneMapper *ID* software v3.1 on the computer that is connected to 3100/3100-*Avant* system using Data Collection v2.0, *the data collection software must be running*. If data collection is not running, the GeneMapper *ID* software v3.1 does not register with the Data Service software.

- GeneMapper *ID* software v3.1 runs on Windows NT® and Windows® 2000 platforms only. (Conversion utilities are included for analyzing data from Macintosh®-based sample files.)
- The version of Oracle® database in the GeneMapper *ID* software v3.1 is an embedded license for use by five (5) named users.

IMPORTANT! To accommodate more than five (5) users, additional GeneMapper *ID* software or Oracle database licenses must be purchased.

- When installing GeneMapper *ID* software v3.1, you must log into the local machine with administrator privileges.
- TCP/IP has to be installed prior to the installation of the Oracle database.

Installing GeneMapper *ID* Software

To install GeneMapper *ID* software version 3.1, use the *InstallationReadMe.txt* file on the CD-ROM, as shown in [Figure 2-1](#).

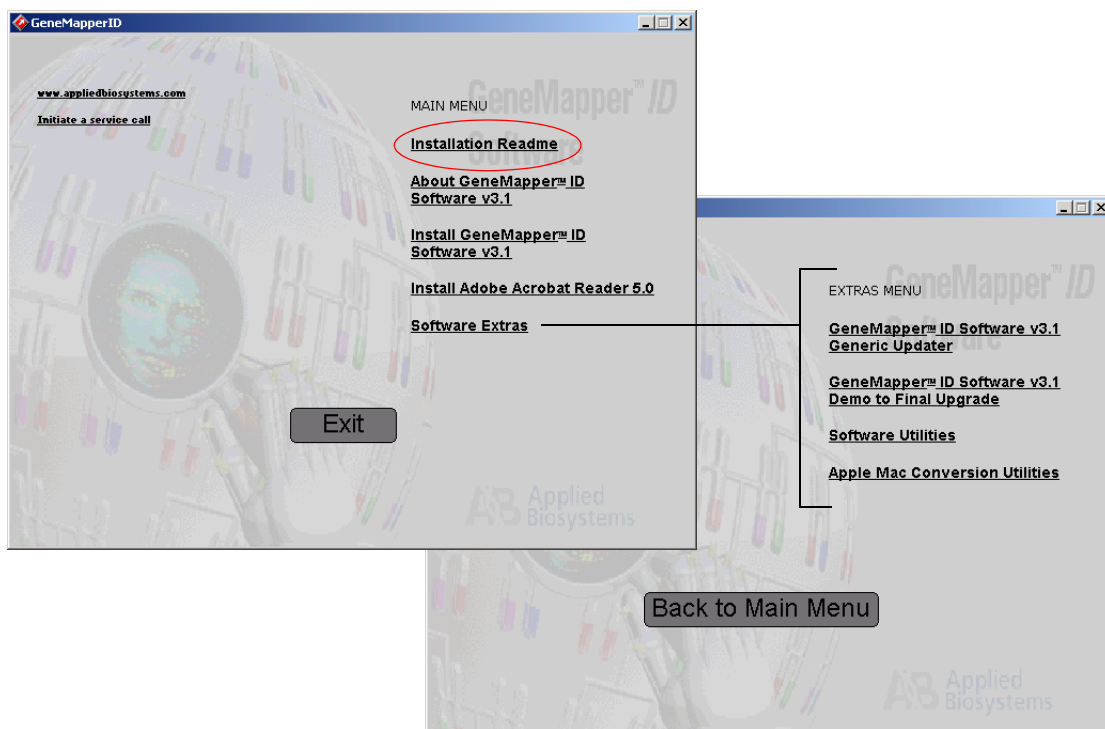


Figure 2-1 Installation options on the GeneMapper *ID* software installation CD-ROM

What gets Installed

The installer on the CD installs:

- GeneMapper *ID* software version
- Oracle database
- Panel folder that contains AmpF Φ STR panels and bins
- Microsatellite and SNP genotyping example data
- Electronic (PDF) version of the user guide
- Size standard folder which contains various size standards
- Default plot and table settings

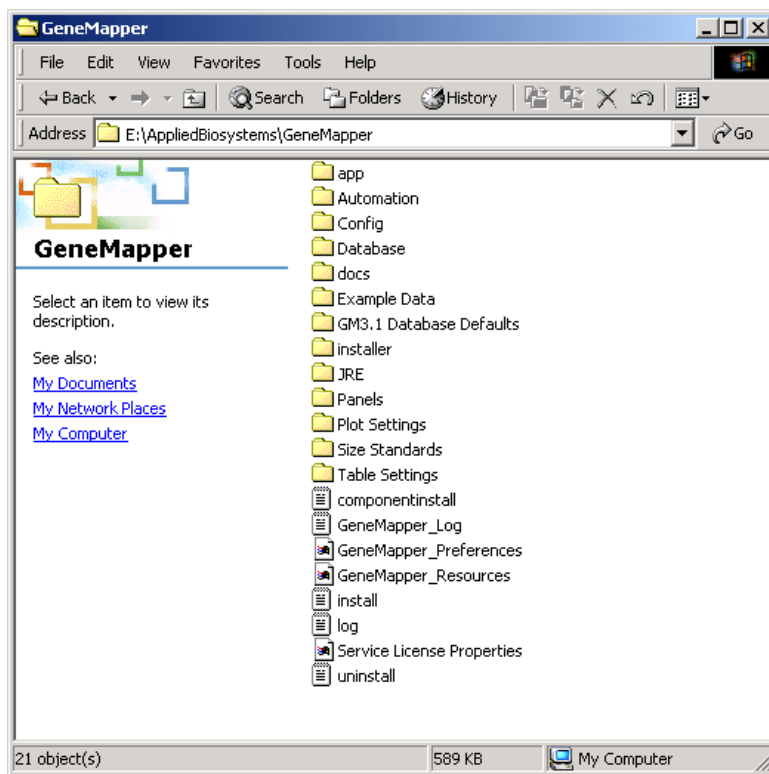


Figure 2-2 Example of the installed GeneMapper folder

Registering GeneMapper ID Software

Completing the Registration Card

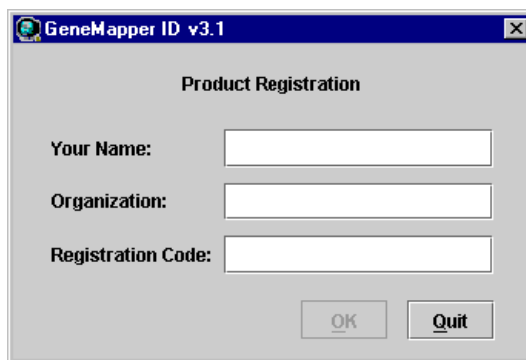
Complete the registration card included in this package and return it to Applied Biosystems. To contact Applied Biosystems technical service and support, see [“How to Obtain Support” on page xi](#) in the Preface.

IMPORTANT! Installation privileges are available only if you have returned your registration card.

Registering GeneMapper ID Software

To register GeneMapper ID software:

1. Select **Start > Programs > Applied Biosystems > GeneMapper > GeneMapperID v3.1** or double-click the GeneMapperID v3.1 desktop shortcut.



2. Register the software.
 - a. In the Product Registration dialog box, enter your name, organization, and registration code (on the registration card).
 - b. Click **OK**.
GeneMapper ID software then verifies the code.

Note: Keep your registration code in a place where you can easily retrieve it. If you need to reinstall the software at any time, you will be prompted for the registration code again.

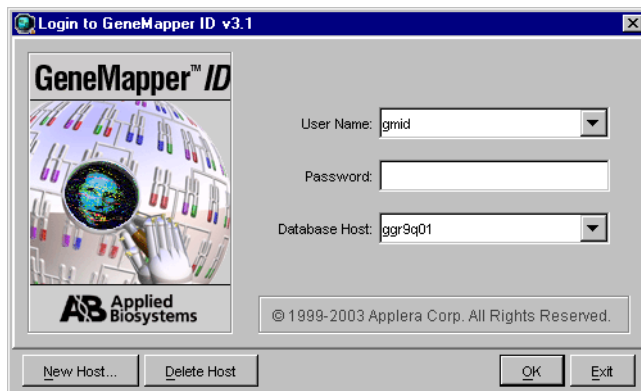
Logging Onto and Logging Out of the GeneMapper ID Software

Introduction This section provides information on logging on to and logging out of the GeneMapper ID software.

GeneMapper ID Software Access Rules Follow these rules to access the GeneMapper ID software.

- The first time you launch the GeneMapper ID software application after installation, you must register the software.
- Only one user can access a database at a time.

Logging On After you enter the registration code (see [“Registering GeneMapper ID Software” on page 2-6](#)), the Login screen appears. You must log on to the GeneMapper ID software program with a user name, so that the system can log information to the database and preserve the options for each user. The first time you log on to the GeneMapper ID software, the following dialog box opens.



To create a password:

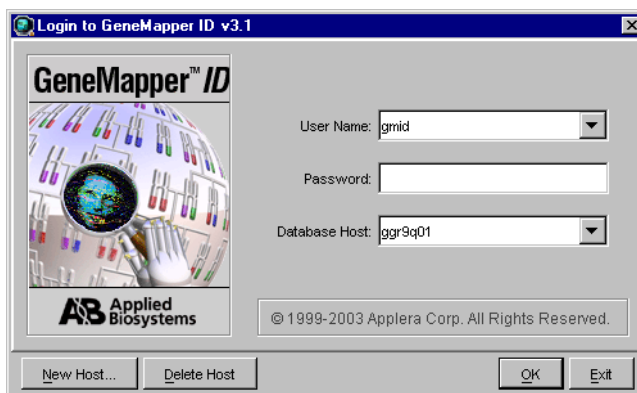
1. The first time you log on leave the User Name as **gmid** and leave the Password field blank.
2. Select the **Database Host** (local computer) from the drop-down menu, and click **OK**.
The software opens a dialog box to change (or create) your password.

3. Create a new password.

Note: The first time you use the GeneMapper *ID* software, leave the User Name as **gmld** and the Old Password field blank.

- a. Enter a new User-defined password.
- b. Confirm the User-defined password and click **OK**.

4. For subsequent launches of the GeneMapper *ID* software the following screen appears.



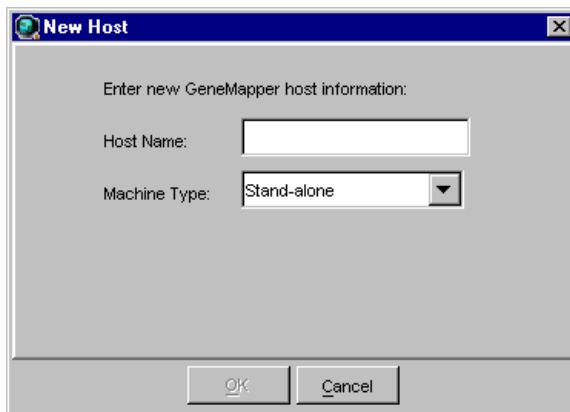
- a. Select the appropriate user name.
- b. Enter your password.
- c. Change the database host if necessary.
- d. Click **OK**.

Note: For information on how to create new users and passwords see “Users Tab” on pages C-24 through C-25.

Database Host Option

The **New Host** option allows you to connect to GeneMapper *ID* software v3.1 databases on other computers from the Login screen.

1. Click the **New Host** button on the Login Screen to open the New Host dialog box.



2. In the **Host Name** field, enter the machine name or IP address of the database host.

The window displays the new database host and its user names list.

Note: If the GeneMapper *ID* software cannot connect to the database host you entered, the following error message opens: “You have entered an invalid host.” Click **OK** to exit and reenter the database host information.

3. Select the appropriate machine type from the drop-down list.
4. Click **OK**.

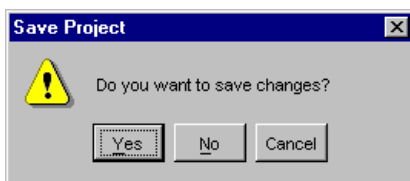
Note: Multiple copies of the GeneMapper *ID* software cannot share the same database. If you want to have the same project, analysis method, and size standard definition as another GeneMapper *ID* database, you need to use the import and export functions in GeneMapper Manager and Panel Manager.

IMPORTANT! Only one user can access a database at a time.

Logging Out Close the GeneMapper *ID* software in one of three ways.

- Select **File > Logout**,
- Select **File > Exit**, or
- Click the **Close** button .

When you log out of the GeneMapper *ID* software, you have the option of saving or discarding the changes you made since the last time you updated the project.



Note: If you close GeneMapper *ID* software with a new or blank Project window (or no changes in your Project), the Save Project (logout) dialog box does not open.

Using the Project Window

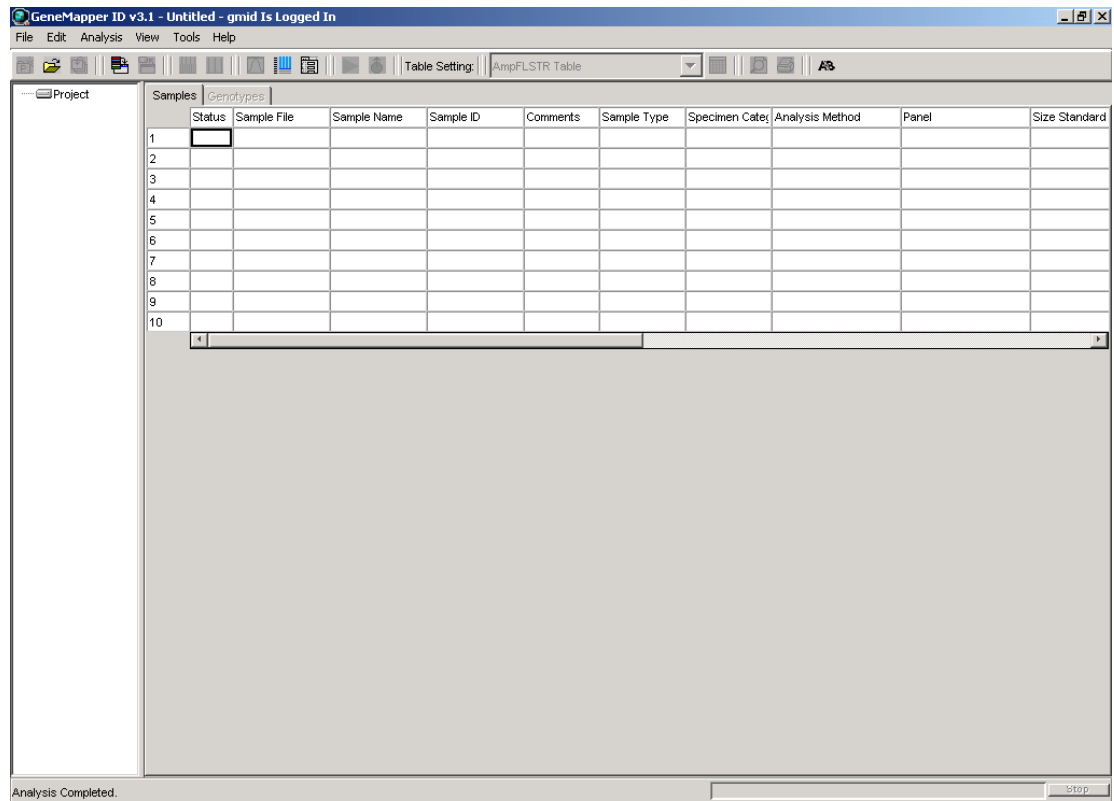
This chapter provides a general overview of the Project window, the central window of the GeneMapper™ *ID* Software Version 3.1 interface.

This chapter covers:

Main GeneMapper <i>ID</i> Project Window	3-2
Overview of the Project Window	3-3
Navigating in the Project Window	3-12

Main GeneMapper ID Project Window

Importance of the Project Window The Project window is the primary window in the GeneMapper ID software user interface. The GeneMapper ID software is organized around the concept of the project to support its purpose of automatically calling alleles or performing genotyping.



For more detailed information on the main project window and its functions and features, see [Appendix C, “Project Window Software Interface.”](#)

The Project window makes it easy for you to:

- Add samples, initiate analysis, and export results
- View sample data and access data plots associated with a particular sample file:
 - The pre-analysis (raw data) version of the data in the Samples view of the Project window
 - The tabular results of analysis in the Genotypes view of the Project window, or
 - Plot(s) of selected Project sample data in the Plot window
- Initiate the process of creating analysis methods and importing and defining panels and bins

Overview of the Project Window

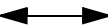
Project Window Interface Conventions

In the Project window you can:

- Minimize the window to an icon on the Microsoft Windows taskbar or to the title bar
- Maximize the window to fill the entire screen
- Resize the window by dragging any window edge or corner

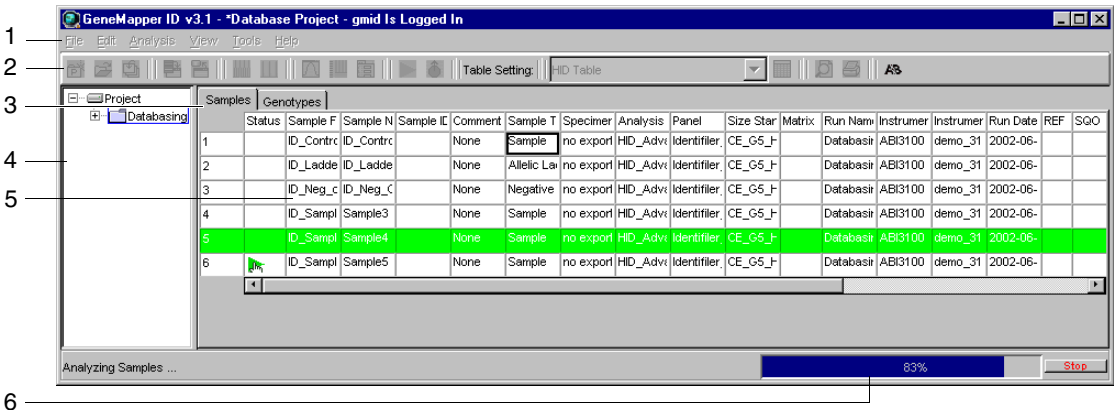
Reformatting the Window

You can reformat the Project window:

- Use the Show Navigator switch on the View menu to show/hide the navigation pane for the Samples and Genotypes views. When the navigation pane is hidden, the table occupies the entire width of the Project window.
- Shift-click a column header to sort the data by that column. The Sort dialog box under the Edit menu allows cumulative sorting over three columns; see [“Sort Dialog Box” on page C-46](#).
- The first column of the table, displaying row numbers, is fixed during horizontal scrolling of the Samples and Genotypes tables.
- Create Hide/Show columns as needed with different table settings created using the Table Settings in the GeneMapper Manager window.
- Resize columns by dragging the separating lines. During resizing, the cursor changes to a resize cursor (a double-headed arrow that looks like this ).

Parts of the Project Window

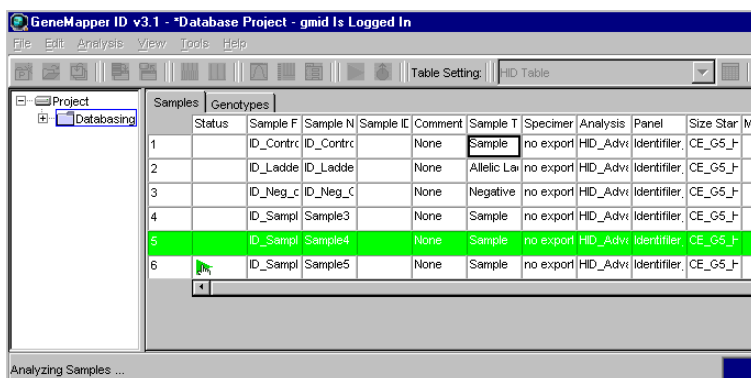
Once you have loaded Sample files into a Project window, it looks like the example below. For more information see [Appendix C](#), “Project Window Software Interface.”



Item	Name	Description
1	Drop-down menus	These menus are described in “ Project Window Menus ” on page C-10.
2	Toolbar	Toolbar icons are described in “ Project Window File Menu Dialog Boxes ” on page C-27.
3	Samples and Genotypes tabs	The use of these tabs is described in “ Samples View ” on page C-2 and “ Genotypes View ” on page C-7.
4	Navigation pane	The use of this pane is described in “ Navigating in the Project Window ” on page 3-12.
5	Samples view columns	These columns are described in “ Samples View Columns ” on page C-4.
6	Progress indicator	The progress of analysis and import/export of projects is shown in this indicator.

Project Window Views

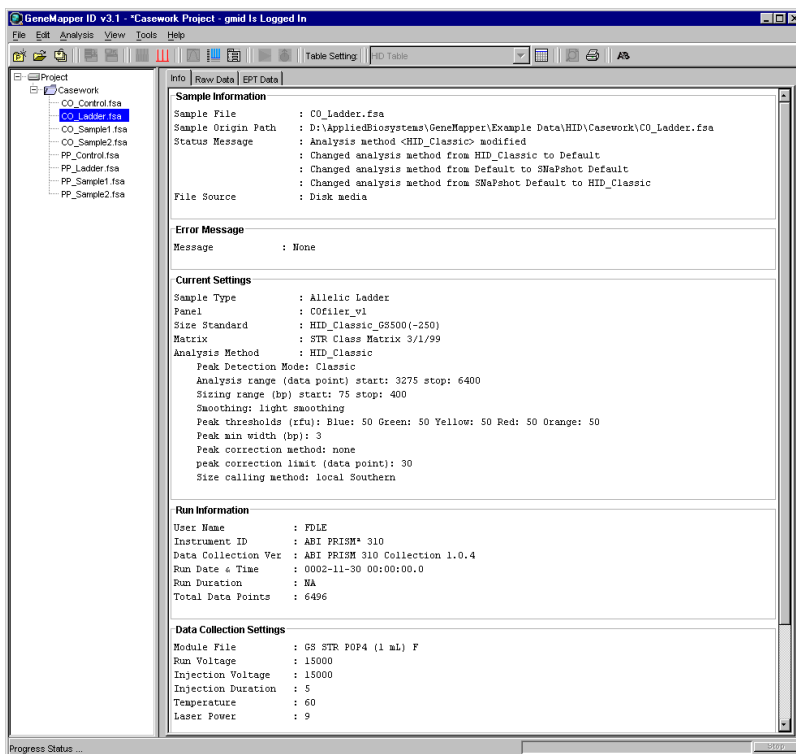
Samples View When you launch the GeneMapper *ID* software, the Samples View is the first view you see. Use this view to open existing projects, import sample files, select analysis settings, analyze data, and view sample PQVs and sizing quality. The Samples tab is always enabled; find it by clicking once on the Project root node in the left navigation pane or by selecting Samples under the View menu.



In the Samples view, you have access to Sample Information, Raw Data, and EPT data. Each of these tabs is discussed in the following pages.

Info Tab

When a Sample file is selected from the navigation pane in the Samples view, a special information window like that shown below is presented and “Info” is the active tab.



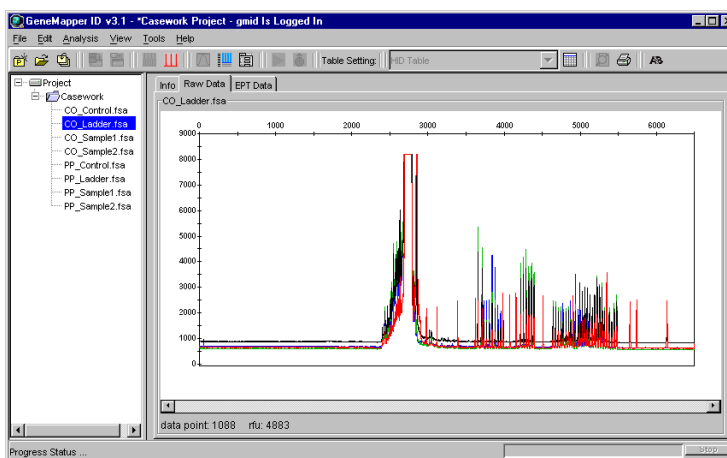
The Info tab displays sample file information such as:

- sample information
- run information
- data collection settings
- gel information
- capillary information
- error messages
- current settings

Sample information comes directly from the Instrument Data Collection and/or Sample sheet.

Raw Data Tab

Clicking the Raw Data tab in the Samples view when the Sample file is selected displays the Raw Data view for the selected Sample file.



Plot-scaling features in the Raw Data view:

- Zoom in on one area by placing the cursor in either the X or Y axis label areas, and click and drag. The software zooms in on the area of selection.
- Return to a full scale plot by double-clicking in the label area again. The plot window is restored to full scale.
- Scale the Y axis to maximum Y or a user defined value by selecting Y-Axis Scale from the View menu.

Use the Raw Data view to evaluate the following:

- baseline problems or noise could result in poor size calling
- start and stop points for analysis

EPT Tab

Clicking the EPT (electrophoresis, power, and temperature) tab in the Samples view, when the Sample file is selected, displays a window that shows the voltage, power, current, and run temperature associated with that sample.



Line	Description
Blue	EP Voltage
Black	EP Power
Green	EP Current
Red	Run Temperature

There are several plot-scaling features in the EPT view:

- Zoom in on one area by placing the cursor in either the X or Y axis label areas, and click and drag. The software zooms in on the area of selection.
- Return to a full scale plot by double-clicking in the label area again. The plot window is restored to full scale.
- Scale the Y-axis to maximum Y or a user defined value by selecting the Y-Axis Scale from the View menu or by specifying upper and lower limits in the Scale to dialog box.

Sample Columns Use columns to set up analyses after you have added samples to a project.

Column Menus

The columns in the Samples view of the Project window have drop-down menus to apply settings to selected cells in the column:

- Sample Type
 - Positive control
 - Negative control
 - Allelic ladder
 - Sample
- Analysis Method
- Panel
- Size Standard
- Matrix
- Specimen Category

Note: To genotype your samples, select Allelic Ladder in the Sample Type column in the allelic ladder row.

Note: The Panel column displays a pop-up window that contains a hierarchical list of kits and panels. A panel is required for any allele-calling analysis.

Use each of these columns to apply needed analysis parameters to selected samples. You can make different list selections to the individual column rows representing samples.

Rules for Columns

The following rules apply to column selections:

- Clicking the header for a column selects the entire column.
- Shift+clicking selects a continuous selection range.
- Ctrl+clicking individual cells makes discontinuous selections.

Applying a List Selection to an Entire Column

To apply a Menu selection:

1. Apply the selection to the top cell in a column:
 - a. Select the top cell in a column to display a drop-down list of items.
 - b. Select and click an item to apply the selection to the cell.
2. Select all other cells in the column, either as a continuous or discontinuous selection, to which the list selection is to be applied.
3. Click **Edit > Fill Down** (Ctrl+D) to apply the choice made for the first cell to all selected cells.

Filling Down a Column

To fill down a column:

1. Double-click the top cell in a column to open the selections.
2. Expand the appropriate folder.
3. Double-click the appropriate panel.
4. Click **Edit > Fill Down**.

Genotypes View

The Genotypes tab becomes enabled only when the data is successfully sized and allele calls are made. In addition, only samples that are sized correctly are displayed in the Genotypes view table. When you perform a sizing only application, this tab is not active after sizing since there are no genotypes to display.

Sample Name	Run Name	Panel	Marker	Dye	Allele 1	Allele 2	AE Contr	AE Contr	ADO	AE	OS	BIN	PHR	LPH
1	CO_Control	Casework	COtlier_v1	D3S1358	B	14	15							
2	CO_Control	Casework	COtlier_v1	D16S539	B	11	12							
3	CO_Control	Casework	COtlier_v1	AMEL	G	X								
4	CO_Control	Casework	COtlier_v1	TH01	G	8	9.3							
5	CO_Control	Casework	COtlier_v1	TPOX	G	8								
6	CO_Control	Casework	COtlier_v1	CSF1PO	G	10	12							
7	CO_Control	Casework	COtlier_v1	D7S820	V	10	11							
8	CO_Ladder	Casework	COtlier_v1	D3S1358	B	12	13							
9	CO_Ladder	Casework	COtlier_v1	D16S539	B	5	8							
10	CO_Ladder	Casework	COtlier_v1	AMEL	G	X	Y							
11	CO_Ladder	Casework	COtlier_v1	TH01	G	5	6							

The genotypes table in this view contains items such as allele calls, genotype PQVs, and overall genotyping quality. This table is customizable and can be easily exported.

- Genotypes View - See [page C-7](#).

The tab for this view is enabled after successful analysis. Samples that do not have a Panel attribute do not display in the Genotypes table.

Navigating in the Project Window

This section provides information on how to navigate as well as how to access different views for samples files.

- Navigation in Samples and Genotypes View
- Info Tab - (See [page 3-6](#))
- Raw Data Tab - (See [page 3-7](#))
- EPT Tab - (See [page 3-8](#))

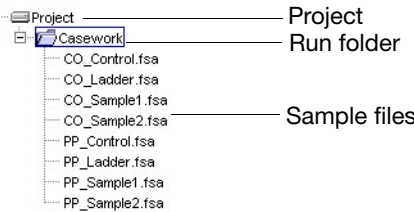
Navigation in Samples and Genotypes Views

The left panel of the Project window has a tree view navigation device. In the Samples view, the tree viewer has three levels. Each sublevel of the tree-view is indented to the right.

In the Genotypes view, the tree viewer has three levels and each sublevel of the tree view is again indented to the right.

Samples View	Genotypes View
● Project	● Panels (root level)
● Run folder	● Panels
● Sample	● Markers (loci)

As soon as you click the tree view controller (indicated in the figure below), the tree view expands with the subordinate level indented. Clicking the controller a second time collapses the level back to its original form.

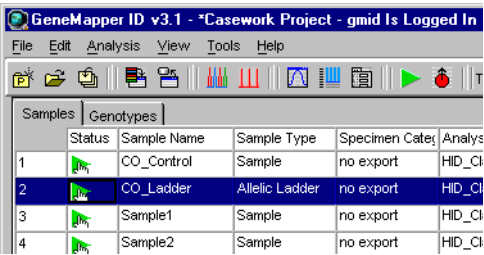
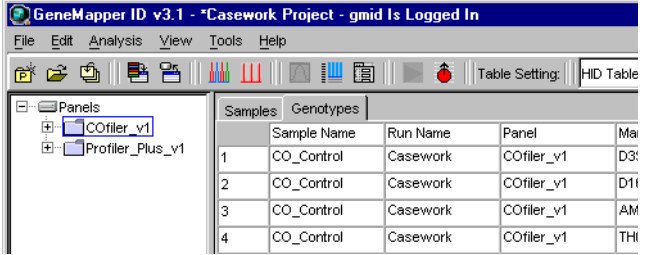
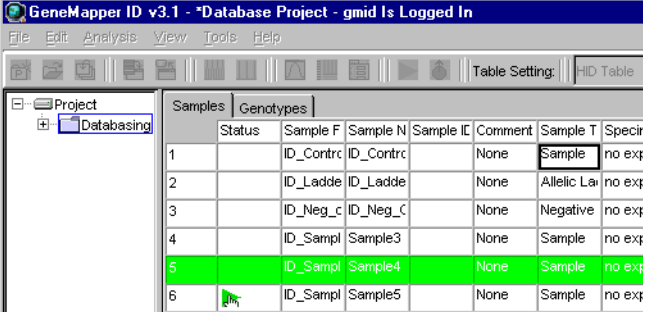


Once you have added files to the Project window:

If you...	Then...
select the project item in the Navigation pane	all samples in the project are displayed in the table.
select a run folder in the Navigation pane	samples in the selected Run folder are displayed in the table.
select a sample file in the Navigation pane	you have access to sample information, raw data and EPT data. Click the tab corresponding to the type of data you want displayed in the right pane.

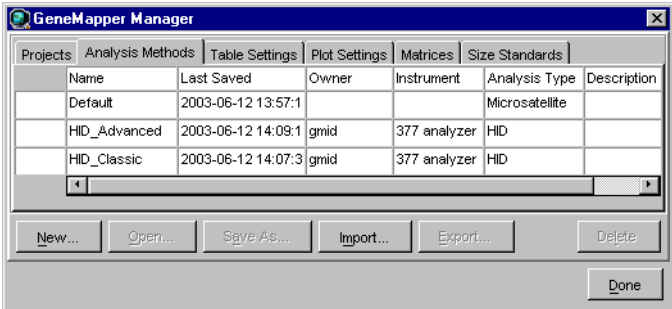
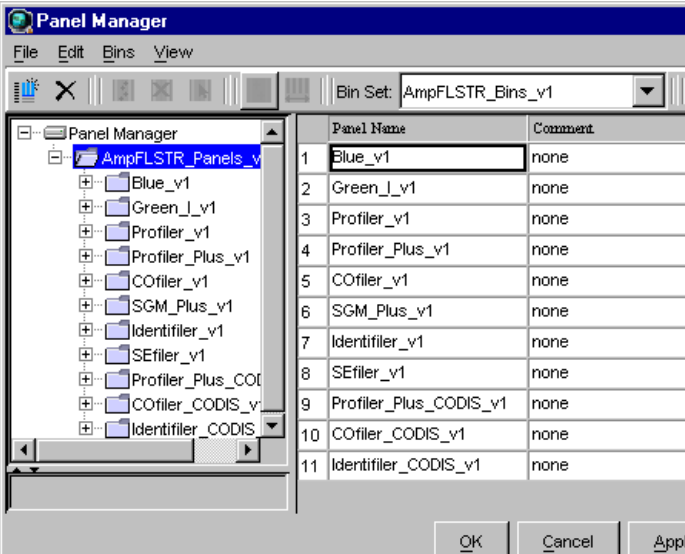
Viewing the Project Window Contents

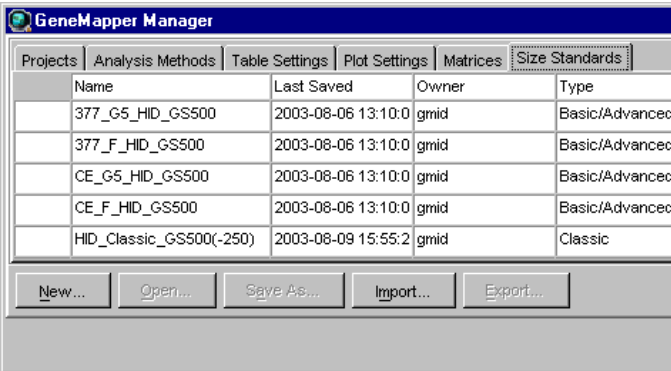
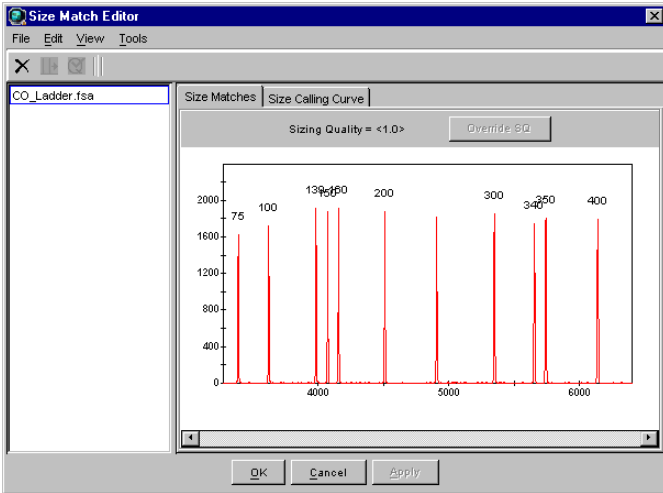
The following table lists how to display information in a Project window.

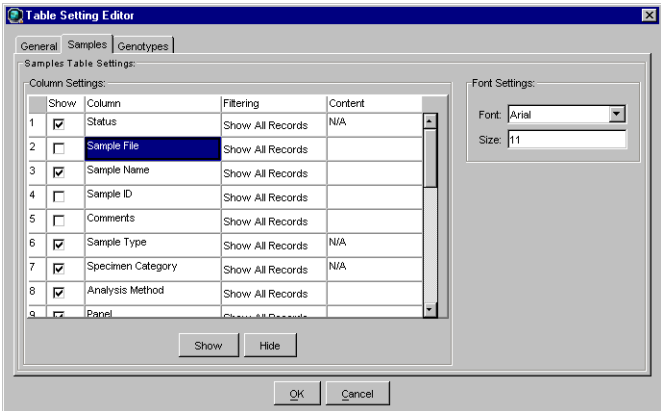
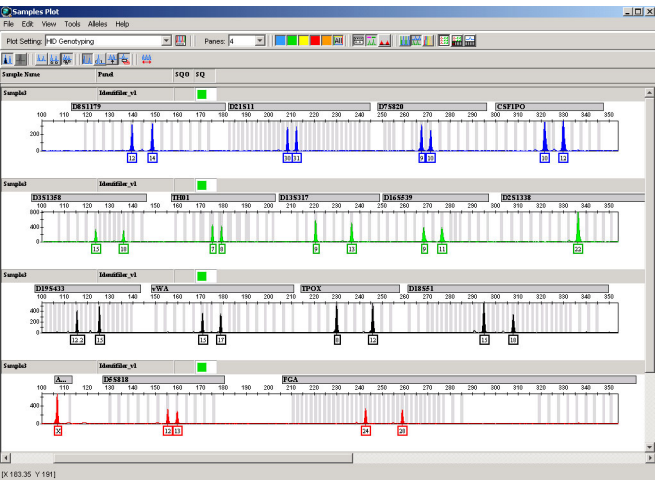
To hide the...	Open the...	Clear the...	And GeneMapper <i>ID</i> software displays the...
Navigation pane	View menu from the Project window Alternative = Ctrl+Shift+N	Show Navigator check mark	Project window without the navigation pane. 
To see the...	Click the...	And GeneMapper <i>ID</i> software displays the...	
Genotypes view Note: Samples must be analyzed to see genotypes.	Genotypes tab Alternatives = Select Genotypes command from the View menu, or Ctrl+Shift+2	Project in the Genotypes view. 	
To see the...	Click the...	And GeneMapper <i>ID</i> software displays the...	
Samples view again	Samples tab Alternatives = Select View > Samples , or Ctrl+Shift+1	Project in the Samples view. 	

Access to Subordinate Application Windows

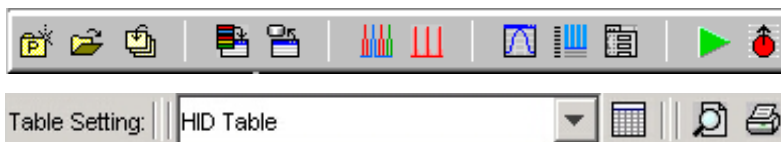
The following table lists how to access subordinate GeneMapper *ID* software windows.

To see the...	Select...	And GeneMapper <i>ID</i> software displays the...
Analysis Methods	Tools > GeneMapper Manager Then select the Analysis Methods tab.	Analysis Methods page. 
Panel Manager	Tools > Panel Manager Alternative = Click the icon: 	Panel Manager window. 

To see the...	Select...	And GeneMapper ID software displays the...
Size Standards	Tools > GeneMapper Manager Then select the Size Standards tab.	Currently Defined Size Standards. 
Size Match Editor	Analysis > Size Match Editor , Alternative = Click the icon: 	Size Match Editor. 

To see the...	Select...	And GeneMapper ID software displays the...
Table Setting Editor	Tools > Table Setting Editor, Alternative = Press Ctrl+T. If samples have been imported, you can click the icon:  Note: To create a new table, select Tools > GeneMapper Manager. Then select the Table Settings tab and click New.	Table Setting Editor. 
Plot window	a sample or results row, and then either select Analysis > Display Plots, press Ctrl+L, Alternative = Click the icon: 	Plot Window. 

Project Window Toolbar



Project window toolbar icons and descriptions:

Icon	Description
	Creates a New Project Tooltip: New Project
	Opens the Open Project dialog box Tooltip: Open Project
	Saves the Project Tooltip: Save Project
	Opens the Add Samples To Project dialog box Tooltip: Add Samples To Project (Ctrl+C)
	Opens the Export dialog box; exports the contents of the Samples or Genotypes tables in tab or comma delimited format (Ctrl+E) Tooltip: Export Table
	Opens the Plots Table window Tooltip: Display Plots
	Opens the Size Match Editor window Tooltip: Size Match Editor
	Opens Analysis Method Editor Tooltip: Analysis Method Editor
	Opens the Panel Manager window Tooltip: Panel Manager
	Opens GeneMapper Manager Tooltip: GeneMapper Manager
	Starts the analysis; displays progress bar and Stop button on the Status bar during analysis Tooltip: Analyze (Ctrl+R)

Project window toolbar icons and descriptions: *(continued)*

Icon	Description
	Brings samples with lower quality values to the top of the table; sorts by overall Genotype quality Tooltip: Bring Low Quality Samples to the Top (Ctrl+B)
	Opens the Table Setting Editor for the currently selected table Tooltip: Table Setting Editor (Ctrl+T)
	Opens the Find dialog box Tooltip: Find (Ctrl+F)
	Opens the Print dialog box Tooltip: Print (Ctrl+P)

Using Panel Manager

This chapter describes the Panel Manager window in the GeneMapper™ ID Software Version 3.1.

Note: For AmpFLSTR® data analysis, you use the Panel Manager only to import predefined panels and bins supplied by Applied Biosystems. Most of the information provided in this chapter is not relevant to AmpFLSTR data analysis. However, this information is provided to explain various features and actions that you can perform using the Panel Manager for microsatellite and some SNP analysis. For application-specific tutorials refer to the *Microsatellite Analysis with ABI PRISM® GeneMapper™ Software v3.0 Tutorial* (PN 4335525) and the *SNP Genotyping with ABI PRISM® GeneMapper™ Software v3.0 Tutorial* (PN 4335524).

This chapter covers:

Panel Manager Overview	4-2
Panel Manager Window Commands	4-7
Formats of Panel and Bin Text Files	4-22
Panel Table View	4-34
Marker Table View	4-36
Bins View - Microsatellites Only	4-39

Panel Manager Overview

Introduction Use the Panel Manager utility to manage all of the chemistry kits, panels, markers (loci), and bin definitions required for automated allele calling.

Topic	See Page
Related Definitions	4-2
Purpose of the Panel Manager Window	4-3
When to Click the Apply Button	4-3
Displaying the Panel Manager Window	4-4
Hierarchy of Panel Data	4-4
Panel Manager Toolbar	4-5
Panel Manager Navigation Pane	4-6

There are two ways to add information to the Panel Manager:

- Creating kits, panels, markers (loci), and bins manually.
- Importing preformatted text files that contain panels, markers (loci), and bin definitions. (See [“Formats of Panel and Bin Text Files”](#) on page 4-22 for two examples.)

Related Definitions The words kit, panel, marker (locus), bin, and bin set are defined in the following table.

Term	Definition
Panel	Collection of loci (A primer set specific to an AmpF ℓ STR kit. For example, Identifiler has 16 loci.)
Kit	Collection of panels (e.g., a panel is an AmpF ℓ STR kit)
Marker	A locus or primer pair. For example, TH01 or vWA.
Bin	An expected location for a particular allele within a locus.
Bin Set	Collection of expected locations for loci contained within a panel or kit.

Purpose of the Panel Manager Window

The Panel Manager window enables you to create, edit, and import kits, panels, markers (loci), bins, and bin sets. It features the following:

- A tool for creating kit folders
- A table for specifying panels
- A table for defining loci within a panel
- A graphical editor for creating and editing bins

All information imported, entered, or created is stored in the database. This information includes kits, panels, markers (loci), bins, and bin sets.

- Clicking **Apply** after making changes to a panel sends all changes to the database but keeps the window open for further edits.
- Clicking **OK** sends all changes to the database and closes the Panel Manager window.

The changes in panel data are then reflected in the Project and Plot windows after the samples are re-analyzed.

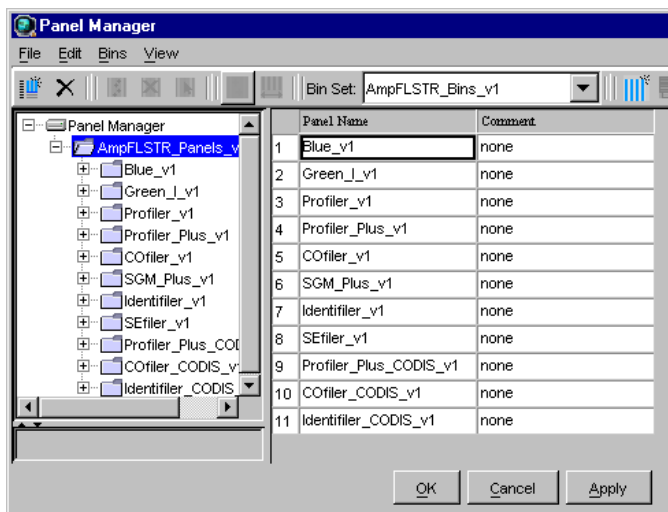
When to Click the Apply Button

- After deleting a kit, panel, locus, or bin set
- After making a number of edits
- After creating a bin set
- After making any change you want to save

Displaying the Panel Manager Window

To display the Panel Manager window, either:

- Click  (Panel Manager) from the Project window toolbar, or
- Select **Tools > Panel Manager** in the Project window.



Hierarchy of Panel Data

Conceptually, the hierarchy of panel data is as follows:

- Kit (chemistry kit or product)
- Panel
- Locus
- Bin

For AmpFLSTR data analysis, different bin sets are not needed for different instrument platforms. The panels and bins supplied by Applied Biosystems for AmpFLSTR data analysis are optimized to accommodate all instruments that support AmpFLSTR data analysis.

The GeneMapper *ID* software requires that kits, panels, and bin sets are unique throughout the database and that locus names are unique within a panel. The Panel Manager enforces these requirements during user entry, editing of names, and data import.

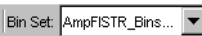
Since the Panel Manager allows users to add comments as needed, no history or audit trail of panel data changes is kept. Such comments provide a way for users to track changes and the reasons for the changes. Each kit, panel, and locus has a comment field.

Panel Manager Toolbar

The toolbar icons enable specific Panel Manager actions similar to their associated menu items as described throughout this chapter. Position your mouse over an icon to view its tooltip description.



Table 4-1 Panel Manager icons and descriptions:

Item	Description	
1		New Kit/Panel/Locus icon automatically updates to reflect the current action depending on the item selected in the navigation pane (File menu).
2		Clear icon performs kit/panel/locus deletion (Edit menu).
3		Icons select bin options: Add Bin, Delete Bin, Edit Bin (Bins menu).
4		Icon allows viewing of Project alleles (Bins menu).
5		Icon switches view to full X-Axis and Y-Axis scales (View menu).
6		Pull-down menu allows selection of a bin set for a selected kit.
7	 Icons select specific kit/panel/locus actions (Bins menu): New Bin Set  Add Reference Data  Auto Bin  Panel Reference Data  Auto Panel 	
8		Dye color toggle icons (View menu).

Panel Manager Navigation Pane

The Panel Manager navigation pane is the primary navigation interface in the Panel Manager window. Since it is found in all Panel Manager views, as shown in the table below, it enables moving back and forth between the views to view, edit, and create kit folders, panels, locus, and bins. The elements of this field are described below.

Element	Description
 Panel Manager	The Panel Manager root node is the container for all panels. It is the top level of the hierarchy.
 Panel Manager  AmpFISTR_Panels_v1	Kits help users organize their panels in product- or project-specific folders. Clicking a kit folder displays the Panel table.
 Panel Manager  AmpFISTR_Panels_v1  Blue_v1  Green_I_v1  Profiler_v1	The Panel folder icon represents a set of loci. Clicking a panel folder displays the locus table. For SNaPshot kits, clicking a panel folder also displays a plot view.
 Panel Manager  AmpFISTR_Panels_v1  Blue_v1  D3S1358  vWA  FGA	The locus icon represents an individual locus. Clicking a microsatellite locus displays the Bin view. Clicking a SNP locus displays the Bin table.

Panel Manager Window Commands

Introduction The tables in this section describe the following Panel Manager views and menus.

Topic	See Page
File Menu	4-7
Edit Menu	4-11
Bins Menu	4-12
View Menu	4-19

File Menu The File menu offers you options to create or export kits, duplicate, import and export panels, and import and export bin sets.



New Kit/Panel/Locus

The New Kit/Panel/Locus command changes its name and function depending on the item selected in the Panel Manager navigation pane as follows:

- **New Kit** – This menu item is enabled when the Panel Manager root node is selected in the navigation pane and allows creation of a new kit folder. When creating a new kit:
 1. Enter in a unique kit name.
 2. Select the appropriate kit type (microsatellite or SNP) from the drop-down menu.
 3. Click **OK** to add kit to navigation pane.

- **New Panel** – This menu item is enabled when a kit folder is selected in the navigation pane. Use New Panel to create a new panel folder. When creating a new panel:
 1. Select the “New Panel” name in the right hand window table and enter in the desired name. Names must be unique within the database.
 2. Repeat to add multiple panels to a kit.
 3. Press **Enter**, then click **Apply** after entering a Panel name to update the navigator pane.
- **New Marker** – This menu item is enabled when a panel folder is selected in the navigation pane. Use New Marker to create a new locus. When creating new loci:
 1. Select the “New Marker” row in the right hand window table and enter in the appropriate information for each column. Refer to [“Marker Table View” on page 4-36](#) for more information.
 2. Repeat to add multiple markers to a panel.
 3. Press **Enter**, then click **Apply** to update the navigator pane.

Duplicate Panel

The Duplicate Panel command creates a copy of a selected panel and places it in the same kit as the original with the name “*Selected Panel-dup*”. For example, the duplicate name for the New Panel is “New Panel-dup.” The panel and all associated bins with all bin sets are duplicated as well. This menu item is enabled when a panel folder is selected in the navigation pane.

Import/Export Panels

Refer to the *GeneMapper™ ID Software v3.1 Human Identification Analysis Tutorial* (PN 4335523) for more information on importing panels and bin sets specific to the AmpF[®]STR PCR Amplification kits.

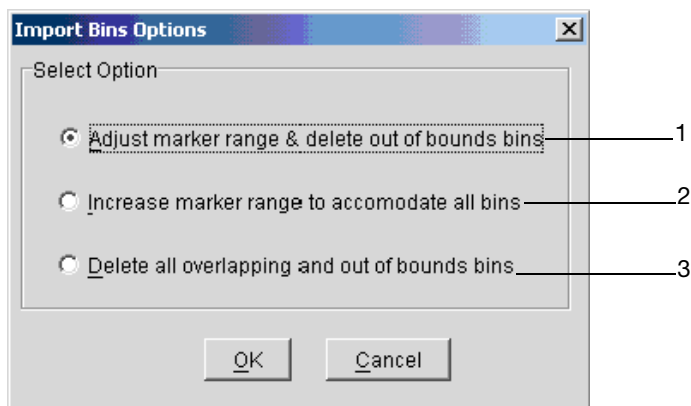
The Import Panels command and the Export Panels command open the corresponding dialog box to allow importing/exporting of kits and panels. This command is enabled when you select the Panel Manager root node in the navigation pane. See [“Formats of Panel and Bin Text Files” on page 4-22](#) for more information.

Export Panels is not available when the Panel Manager root node is selected; it is only active when a kit is selected.

Import/Export Bin Set

The Import Bin Set command and the Export Bin Set command open the corresponding dialog box to allow importing/exporting of bin sets. This command is enabled when you select a kit name in the navigation pane. See [“Formats of Panel and Bin Text Files” on page 4-22](#) for more information.

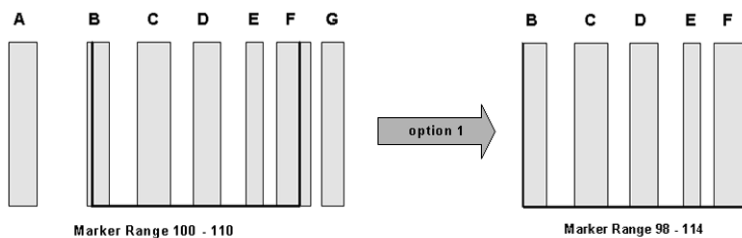
When bins are imported into a kit, GeneMapper *ID* software checks for bins that fall outside of their marker allele size ranges. If bins overlap or fall out of the marker range, the Import Bins Options dialog box will open.



Use the Import Bins Options dialog box to choose how GeneMapper *ID* software handles the bins being imported. The numbered lines in the picture above refer to the individual numbered descriptions in the following figures.

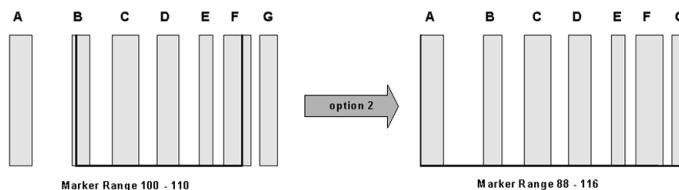
1. The **Adjust marker range & delete out of bounds bins** option shown below adjusts the marker range to accommodate any bins that overlap the marker boundaries and will delete all bins that fall completely outside the marker size range.

1 - Adjust marker range and delete out-of-bounds bins



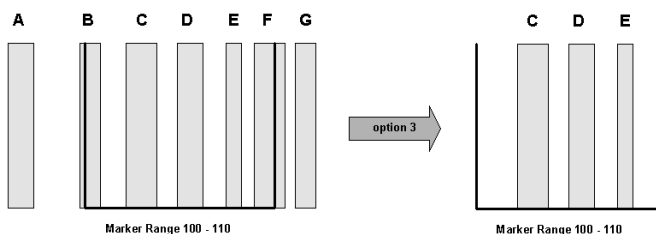
2. The **Increase marker range to accommodate all bins** option shown below increases the marker allele size range to accommodate all overlapping and out of bounds bins.

2 - Increase marker range to accommodate all bins



3. The **Delete all overlapping bins and out-of-bounds bins** option shown below does not affect the marker allele size range; all overlapping and out of bounds bins are deleted.

3 - Delete all overlapping bins and out-of-bounds bins



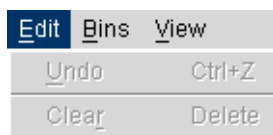
Note: If all the bins in an import are inside their marker ranges, the dialog box does not open.

Export All Kits

The only way to back up panels and bin sets is to export them. You will be prompted for a location and name for the exported item.

The Export All Kits command opens the Export All Kits dialog to allow exporting all kits and their associated panels, markers, and bin sets to a selected location. When you use this option, panels retain their current panel name as listed in the Panel Manager. Bins are named as follows: *[Kit name]_[Bin set name]_bins.txt*.

Edit Menu The Edit menu contains the Undo command and the Clear command.



Undo

The undo command allows for the Clear command to be undone.

Clear

The Clear command deletes items selected in the navigation pane from the GeneMapper *ID* database:

- In the Kits file menu, Clear deletes the kit and all bin sets associated with that kit.
- In the Panels file menu, Clear deletes the panels.
- In the Markers file menu, Clear deletes the markers.

Use the Bins Menu items to delete bins and bin sets.

Bins Menu The Bins menu contains commands to edit markers and bins.

Bins	View
Edit SNP Marker...	Ctrl+H
Delete SNP Marker	Ctrl+Shift+D
Add Bin	Ctrl+B
Edit Bin	Ctrl+H
Delete Bin	Ctrl+D
New Bin Set...	
Delete Bin Set	
Show Project Alleles	
Add Reference Data...	Ctrl+R
Panel Reference Data...	Ctrl+R
Auto Bin	Ctrl+A
Auto Panel	Ctrl+A

Edit SNP Marker

The Edit SNP Marker command opens the Edit SNP Marker dialog box and allows you to edit the marker name and bin information for the selected marker. To enable this feature:

- Select the menu item when a SNP genotyping marker is selected in the Panel Manager Plot tab, or
- Right-click on a SNP genotyping marker or bin in the Panel Manager Plot tab

You can quickly edit SNP genotyping markers and bins by changing the data in the Panel Manager Table tab for a selected SNP genotyping Panel.

Delete SNP Marker

The Delete SNP Marker command deletes the SNP genotyping marker and associated bins selected in the Panel Manager Plot tab. To enable this feature:

- Select the menu item when a SNP genotyping marker or bin is selected in the Panel Manager Plot tab, or
- Right-click on a SNP genotyping marker in the Panel Manager Plot tab

Add Bin

The Add Bin command creates a new bin associated with a panel as follows:

For microsatellite data:

1. Select a marker in the Panel Manager navigation pane.
2. Select the **Add Bin** menu item, or right-click within the allele size range of the marker shown in the Panel Manager right-hand window to select the New Bin option.
3. Drag the length of the desired bin range with the mouse.
4. Edit the bin information in the Add Bin dialog box that opens.

For SNP Genotyping data:

1. Select a SNP genotyping panel in the Panel Manager navigation pane.
2. Select a marker in the Plot tab.
3. Select the **Add Bin** menu item, or right-click on a marker or bin in the Plot tab.
4. Edit the SNP marker using the dialog box that opens.

Delete Bin

The Delete Bin command deletes a marker bin as follows:

For microsatellite data:

1. Select a marker in the Panel Manager navigation pane.
2. Select a bin to be deleted.
3. Select the **Delete Bin** menu item, or right-click the bin and select the Delete Bin option.

For SNP Genotyping data:

1. Select a SNP genotyping panel in the Panel Manager navigation pane.
2. Select a bin to be deleted in the Plot tab.
3. Select the **Delete Bin** menu item, or right-click the bin and select the Delete Bin option.

Edit Bin

The Edit Bin command edits a marker bin as follows:

For microsatellite data:

1. Select a marker in the Panel Manager navigation pane.
2. Select a bin to be edited.
3. Select the **Edit Bin** menu item, or right-click the bin and select the Edit Bin option.
4. Edit the bin information in the dialog box that opens.

For SNP Genotyping data:

1. Select a SNP genotyping panel in the Panel Manager navigation pane.
2. Select a bin to be edited in the Plot tab.
3. Select the **Edit Bin** menu item, or right-click the bin and select the Edit Bin option.
4. Edit the bin information in the Edit SNP Marker dialog box that opens.

To quickly resize a bin, select a bin and adjust the right and left handles to the desired size range.

To move a bin, select a bin and holding down the mouse button, move the bin to the desired location.

Edit Bin Dialog Box

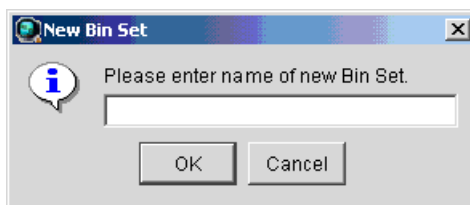
Table 4-2 Edit Bin dialog box elements and descriptions:

Element	Description
Name	Editable; name of the bin can be alphanumeric.
Location	Editable; location of the bin center in base pairs.
Left Offset	Editable; the left boundary of the bin, expressed as the base pair distance to be subtracted from the location (default = 0.4).
Right Offset	Editable; The right boundary of the bin, expressed as the base pair distance to be added to the location (default = 0.4).

Table 4-2 Edit Bin dialog box elements and descriptions:

Element	Description
OK	Accepts the pending changes and closes the dialog box. The following criteria apply: • The edited bin must have a name unique in its marker. • The edited bin must have a location and offsets that do not overlap any other bins in the marker. An alert message will be displayed if these criteria are not met.
Cancel	Closes the dialog box without accepting the pending changes
New Offset Default (checkbox)	Checking this box sets a left and right offset default, to be used for subsequent creation of bins.
Mutant Bin	Use this feature to distinguish microvariant bins from known allele bins. Bin color is red.

New Bin Set

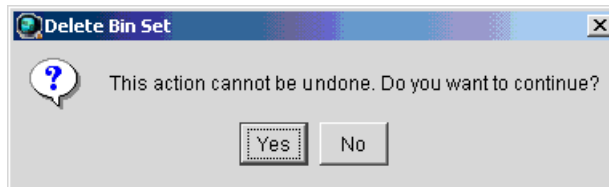


The New Bin Set command opens the New Bin Set dialog box, allows you to create a new bin set for the selected kit. Clicking **OK** checks the name for uniqueness; if the name is not unique, an alert is shown and the dialog box closes. The new bin set name then displays at the top of the Panel Manager window in the Bin Set pull-down menu.

Note: Bin sets are always created at the kit level.

Note: SNaPshot kits can only contain one bin set.

Delete Bin Set



The Delete Bin Set command deletes the bin set currently listed for a kit in the Bin Set drop-down menu at the top of the Panel Manager window. If there are multiple bin sets associated with a kit, use the drop-down menu to select the appropriate bin set to be deleted.

Note: This action cannot be undone.

Show Project Alleles

The Show Project Alleles command displays all of the marker's allele calls that are present in the genotypes table of the open project. Allele calls are displayed as blue asterisks. This option is enabled when a marker is selected in the Panel Manager navigation pane and a profile exists for that marker. The option is for microsatellite data only.

Add Reference Data

The Add Reference Data command opens the Add Reference Data dialog box, which allows you to associate data with a microsatellite panel or SNaPshot kit for creating markers and bins.

For microsatellite panel reference data, samples must have been analyzed using the panel for which the data serves as a reference and the appropriate size standard.

For SNaPshot kit reference data, samples must have been analyzed using the appropriate size standard and analysis method with SNP genotyping as the analysis type.

Panel Reference Data

This feature works for both SNP genotyping and microsatellite data.

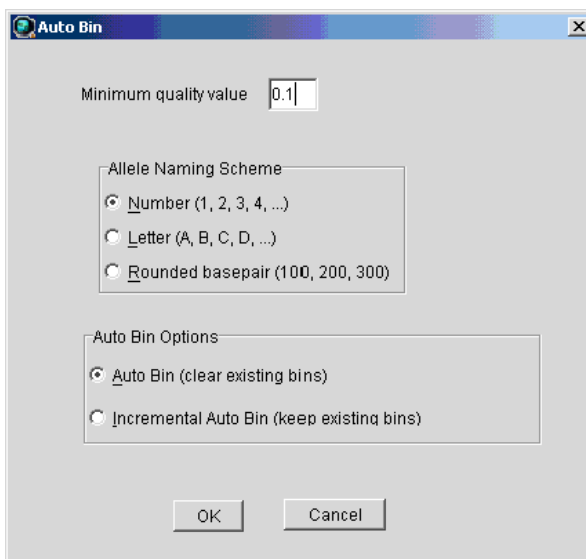
- SNP genotyping data

The Panel SNP Reference Data command opens the SNP Reference Data dialog box, which allows you to organize the reference data imported at the kit level into panels. This feature is only used when creating panels manually, not when using the Auto Panel feature. The panel is enabled when a SNP genotyping panel is selected and reference data has already been added at the kit level.

- Microsatellite data

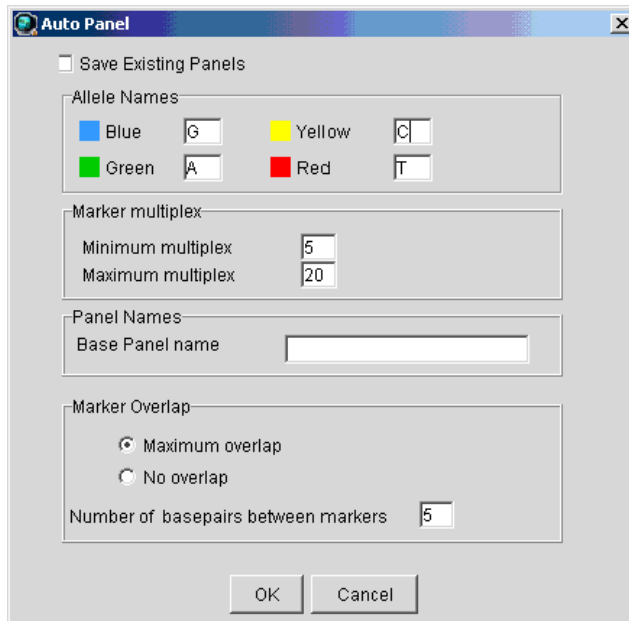
Use the Panel Reference Data command to remove reference samples from the selected microsatellite panel.

Auto Bin



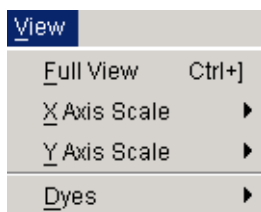
The Auto Bin command opens the Auto Bin dialog box to automatically create bins for a microsatellite panel. This feature is enabled when a microsatellite panel is selected and reference data has been selected. Refer to the Microsatellite Tutorial for more information.

Auto Panel



The Auto Panel command opens the Auto Panel dialog box to automatically create panels, markers, and bins for a SNaPshot Kit. This feature can only be used with reference data generated from the ABI PRISM® SNaPshot Primer Focus Kit. Refer to the SNaPshot Tutorial for more information.

View Menu



Full View

Depending on the X- and Y-axes mode, the Full View command resets the X- or Y-axis to the:

- maximum range
- panel size range
- most recent Scale To range

X Axis Scale for SNP genotyping data



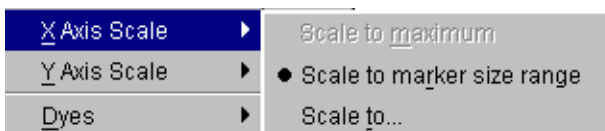
The X Axis Scale menu command controls the horizontal scale of the reference data electropherogram being viewed in the Panel Manager as follows:

- **Scale to maximum** – The electropherogram being viewed is scaled to its maximum base pair length as collected from the instrument.
- **Scale to panel size range** – The electropherogram being viewed is scaled to the size range available for the selected panel.
- **Scale to** - The user can define the horizontal range being viewed.

To zoom in to a specific region on the X-axis of the electropherogram:

- Click and drag with the left mouse button the desired region on the X-axis, or
- Right-click on the X-axis to open the X-Axis Scale menu command options.

X Axis for microsatellite data



The X Axis Scale menu command controls the horizontal scale of the reference data electropherogram being viewed in the Panel Manager as follows:

- **Scale to maximum** -The electropherogram being viewed is scaled to its maximum base pair length as collected from the instrument.
- **Scale to marker size range** - The electropherogram being viewed is scaled to the size range available for the selected marker.
- **Scale to** - You can define the horizontal range being viewed.

To zoom in to a specific region on the X-axis of the electropherogram:

- Click and drag with the left mouse button the desired region on the X-axis, or
- Right-click on the X-axis to open the X-Axis Scale menu command options.

Y Axis Scale



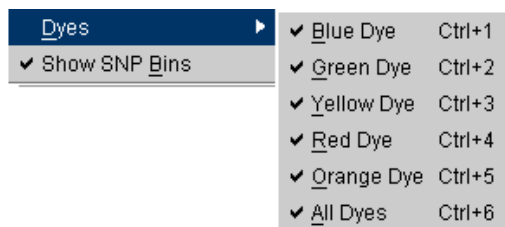
The Y-Axis Scale menu command controls the vertical scale of the reference data electropherogram being viewed as follows:

- **Scale to maximum** - The electropherogram being viewed is scaled to its maximum height as collected from the instrument.
- **Scale to** - You can define the vertical range being viewed.

To zoom in to a specific region on the Y-axis of the electropherogram:

- Click and drag with the left mouse button the desired region on the Y-axis, or
- Right-click the Y-axis to open the Y-Axis Scale menu command options.

Dyes



The Dyes menu item controls the dye colors shown when viewing SNP reference data and microsatellite reference data electropherograms in the Panel Manager Plot view checked when active. You may select multiple dye colors.

Formats of Panel and Bin Text Files

This section provides an example of a file used to import panel definitions and a file used to import bin definitions. These files are created using Microsoft® Excel or other spreadsheet programs. Files must be saved as a tab-delimited text file.

Topic	See page
Panel Definition Example	4-23
Bins Definition Example	4-27
Panel Definitions for SNP Genotyping	4-29
Bins Definitions for SNP Genotyping	4-30
Import and Export Files	4-33

Panel Definition Example

The following examples show panels defined in the format used by GeneMapper *ID* Software v3.1 for a panel definition in a tab-delimited text file (.txt) using notepad or wordpad, or a Microsoft Excel spreadsheet saved as a .txt file.

AmpFLSTR_Panels_v1 - Notepad												
File Edit Format Help												
#GeneMapper ID v3.1 Last edited 071403												
#GMV3.0 for import into GeneMapper ID v3.1												
Version GMV3.0												
Kit type Microsatellite												
Chemistry Kit AmpFLSTR_Panels_v1												
Panel Blue_v1												
D3S1358	blue	98.00	148.00	14,15	4	0.110	none	"12,13,14,15,16,17,18,19"				
VWA	blue	151.00	203.00	17,18	4	0.110	none	"11,12,13,14,15,16,17,18,19,20,21"				
FGA	blue	206.25	360.00	23,24	4	0.110	none	"18,19,20,21,22,23,24,25,26,26.2,27,28,29,30"				
Panel Green_I_v1												
AMEL	green	106.00	114.00	x	9	0.060	none	"X,Y"				
TH01	green	159.00	205.00	8,9,3	4	0.060	none	"5,6,7,8,9,9,3,10"				
TPOX	green	212.00	254.00	8	4	0.060	none	"6,7,8,9,10,11,12,13"				
CSF1PO	green	275.00	323.00	10,12	4	0.090	none	"6,7,8,9,10,11,12,13,14,15"				
Panel Profiler_v1												
D3S1358	blue	98.00	148.00	14,15	4	0.110	none	"12,13,14,15,16,17,18,19"				
VWA	blue	151.00	203.00	17,18	4	0.110	none	"11,12,13,14,15,16,17,18,19,20,21"				
FGA	blue	206.25	360.00	23,24	4	0.110	none	"18,19,20,21,22,23,24,25,26,26.2,27,28,29,30"				
AMEL	green	106.00	114.00	x	9	0.000	none	"X,Y"				
TH01	green	159.00	205.00	8,9,3	4	0.060	none	"5,6,7,8,9,9,3,10"				
TPOX	green	212.00	254.00	8	4	0.060	none	"6,7,8,9,10,11,12,13"				
CSF1PO	green	275.00	323.00	10,12	4	0.090	none	"6,7,8,9,10,11,12,13,14,15"				
D5S818	yellow	128.00	180.00	11	4	0.100	none	"7,8,9,10,11,12,13,14,15,16"				
D13S317	yellow	192.00	242.00	11	4	0.100	none	"8,9,10,11,12,13,14,15"				
D7S820	yellow	251.00	298.50	10,11	4	0.090	none	"6,7,8,9,10,11,12,13,14,15"				
Panel Profiler_plus_v1												
D3S1358	blue	98.00	148.00	14,15	4	0.110	none	"12,13,14,15,16,17,18,19"				
VWA	blue	151.00	203.00	17,18	4	0.110	none	"11,12,13,14,15,16,17,18,19,20,21"				
FGA	blue	206.25	360.00	23,24	4	0.110	none	"18,19,20,21,22,23,24,25,26,26.2,27,28,29,30"				
AMEL	green	106.00	114.00	x	9	0.000	none	"X,Y"				
D8S1179	green	118.00	183.50	13	4	0.120	none	"8,9,10,11,12,13,14,15,16,17,18,19"				
D21S11	green	184.50	247.50	30	4	0.130	none	"24,2,25,26,27,28,28.2,29,29.2,30,30.2,31,31.2,32,32.2"				
D18S51	green	264.49	350.00	15,19	4	0.160	none	"9,10,10.2,11,12,13,13.2,14,14.2,15,16,17,18,19,20,20.2"				
D5S818	yellow	128.00	180.00	11	4	0.100	none	"7,8,9,10,11,12,13,14,15,16"				
D13S317	yellow	192.00	242.00	11	4	0.100	none	"8,9,10,11,12,13,14,15"				
D7S820	yellow	251.00	298.50	10,11	4	0.090	none	"6,7,8,9,10,11,12,13,14,15"				
Panel cofiler_v1												
D3S1358	blue	98.00	148.00	14,15	4	0.110	none	"12,13,14,15,16,17,18,19"				
D16S539	blue	229.00	279.00	11,12	4	0.130	none	"5,8,9,10,11,12,13,14,15"				
AMEL	green	106.00	114.00	x	9	0.000	none	"X,Y"				
TH01	green	159.00	205.00	8,9,3	4	0.060	none	"5,6,7,8,9,9,3,10"				
TPOX	green	212.00	254.00	8	4	0.060	none	"6,7,8,9,10,11,12,13"				
CSF1PO	green	275.00	323.00	10,12	4	0.090	none	"6,7,8,9,10,11,12,13,14,15"				
D7S820	yellow	251.00	298.50	10,11	4	0.090	none	"6,7,8,9,10,11,12,13,14,15"				
Panel SGM_plus_v1												
D3S1358	blue	98.00	148.00	15,16	4	0.110	none	"12,13,14,15,16,17,18,19"				

Figure 4-1 Example of a Panel Definition in Tab-Delimited Text Format

Note: There are no extra rows between markers and panels.

Column	Descriptions
1	Marker name = locus name
2	Dye color
3	Minimum ASR = allele size range (actually, marker size range)
4	Maximum ASR = allele size range (actually, marker size range)
5	Control allele names as comma-separated bin names (1, 2) Note: If none, there must be a dash “-”
6	Type of repeat (2 = dinucleotide, 3 = trinucleotide, 4 = tetranucleotide, 5 = non-repeat)
7	Marker-specific stutter ratio (if none, use zero “0.0”)
8	Comments (if none, write “none”)
9	Alleles present in the allelic ladder for each marker

```

AmpFLSTR_Panels_v1 - Notepad
File Edit Format Help
#GeneMapper ID v3.1 Last edited 071403
#GMV3.0 for import into GeneMapper ID v3.1
Version GMV3.0
Kit type Microsatellite
Chemistry kit AmpFLSTR_Panels_v1
Panel Blue_v1
D3S1358 blue 98.00 148.00 14,15 4 0.110 none "12,13,14,15,16,17,18,19"
VWA blue 151.00 203.00 17,18 4 0.110 none "11,12,13,14,15,16,17,18,19,20,21"
FGA blue 206.25 360.00 23,24 4 0.110 none "18,19,20,21,22,23,24,25,26,26.2,27,28,29,30"

Panel Green_I_v1
AMEL green 106.00 114.00 x 9 0.060 none "X,Y"
TH01 green 159.00 205.00 8,9,3 4 0.060 none "5,6,7,8,9,9,3,10"
TPOX green 212.00 254.00 8 4 0.060 none "6,7,8,9,10,11,12,13"
CSF1PO green 275.00 323.00 10,12 4 0.090 none "6,7,8,9,10,11,12,13,14,15"

Panel Profiler_v1
D3S1358 blue 98.00 148.00 14,15 4 0.110 none "12,13,14,15,16,17,18,19"
VWA blue 151.00 203.00 17,18 4 0.110 none "11,12,13,14,15,16,17,18,19,20,21"
FGA blue 206.25 360.00 23,24 4 0.110 none "18,19,20,21,22,23,24,25,26,26.2,27,28,29,30"
AMEL green 106.00 114.00 x 9 0.000 none "X,Y"
TH01 green 159.00 205.00 8,9,3 4 0.060 none "5,6,7,8,9,9,3,10"
TPOX green 212.00 254.00 8 4 0.060 none "6,7,8,9,10,11,12,13"
CSF1PO green 275.00 323.00 10,12 4 0.090 none "6,7,8,9,10,11,12,13,14,15"
D15S818 yellow 128.00 180.00 11 4 0.100 none "7,8,9,10,11,12,13,14,15"
D13S317 yellow 192.00 242.00 11 4 0.100 none "8,9,10,11,12,13,14,15"
D7S820 yellow 251.00 298.50 10,11 4 0.090 none "6,7,8,9,10,11,12,13,14,15"

Panel Profiler_Plus_v1
D3S1358 blue 98.00 148.00 14,15 4 0.110 none "12,13,14,15,16,17,18,19"
VWA blue 151.00 203.00 17,18 4 0.110 none "11,12,13,14,15,16,17,18,19,20,21"
FGA blue 206.25 360.00 23,24 4 0.110 none "18,19,20,21,22,23,24,25,26,26.2,27,28,29,30"
AMEL green 106.00 114.00 x 9 0.000 none "X,Y"
D8S1179 green 118.00 183.50 13 4 0.120 none "8,9,10,11,12,13,14,15,16,17,18,19"
D21S11 green 184.50 247.50 30 4 0.130 none "24,25,26,27,28,28.2,29,29.2,30,30.2,31,31.2,32,32.2"
D18S51 green 264.49 350.00 15,19 4 0.160 none "9,10,10.2,11,12,13,13.2,14,14.2,15,16,17,18,19,20,21"
D5S818 yellow 128.00 180.00 11 4 0.100 none "7,8,9,10,11,12,13,14,15,16"
D13S317 yellow 192.00 242.00 11 4 0.100 none "8,9,10,11,12,13,14,15"
D7S820 yellow 251.00 298.50 10,11 4 0.090 none "6,7,8,9,10,11,12,13,14,15"

Panel Cofiler_v1
D3S1358 blue 98.00 148.00 14,15 4 0.110 none "12,13,14,15,16,17,18,19"
D16S539 blue 229.00 279.00 11,12 4 0.130 none "5,8,9,10,11,12,13,14,15"
AMEL green 106.00 114.00 x 9 0.000 none "X,Y"
TH01 green 159.00 205.00 8,9,3 4 0.060 none "5,6,7,8,9,9,3,10"
TPOX green 212.00 254.00 8 4 0.060 none "6,7,8,9,10,11,12,13"
CSF1PO green 275.00 323.00 10,12 4 0.090 none "6,7,8,9,10,11,12,13,14,15"
D7S820 yellow 251.00 298.50 10,11 4 0.090 none "6,7,8,9,10,11,12,13,14,15"

Panel SGM_Plus_v1
D3S1358 blue 98.00 148.00 15,16 4 0.110 none "12,13,14,15,16,17,18,19"

```

Figure 4-2 Example of a panel definition from a spreadsheet saved as a .txt document

Note: There are no extra rows between markers and panels.

Column	Descriptions
1	Marker name = locus name
2	Dye color
3	Minimum ASR = allele size range (actually, marker size range)
4	Maximum ASR = allele size range (actually, marker size range)
5	Control allele names as comma-separated bin names (1, 2). If none, there must be a dash “-”
6	Type of repeat (2 = dinucleotide, 3 = trinucleotide, 4 = tetranucleotide, 5 = non-repeat)
7	Marker-specific stutter ratio (if none, use zero “0.0”)
8	Comments (if none, write “none”)

Bins Definition Example

The examples below show bins defined in the format used by GeneMapper ID Software v3.1 in a tab-delimited text file (.txt) using notepad or wordpad or an Excel spreadsheet saved as a .txt file.

Note: The allele size range (ASR) in the bin definitions file should match the ASR in the Panels definition file. Also, kit, panel, and marker names must be the same between the two files.

5
6
7
8
9

```
#GeneMapper ID v3.1 Last edited 071403
#GMv3.0 for import into GeneMapper ID v3.1
Version          GMv3.0
Chemistry Kit     AmpFLSTR_Panels_v1
BinSet Name       AmpFLSTR_Bins_v1
Panel Name        Blue_v1
Marker Name       D3S1358
```

11	110.00	0.5	0.5
12	114.00	0.5	0.5
13	118.00	0.5	0.5
14	122.00	0.5	0.5
15	126.00	0.5	0.5
15.2	128.00	0.5	0.5
16	130.00	0.5	0.5
16.2	132.00	0.5	0.5
17	134.00	0.5	0.5
17.2	136.00	0.5	0.5
18	138.00	0.5	0.5
18.2	140.00	0.5	0.5
19	142.00	0.5	0.5
20	146.00	0.5	0.5

Marker	Name	VWA	
10	153.00	0.5	0.5
11	157.00	0.5	0.5
12	161.00	0.5	0.5
13	165.00	0.5	0.5
14	169.00	0.5	0.5
15	173.00	0.5	0.5
15.2	175.00	0.5	0.5
16	177.00	0.5	0.5
17	181.00	0.5	0.5
18	185.00	0.5	0.5
18.2	187.00	0.5	0.5
19	189.00	0.5	0.5
20	193.00	0.5	0.5
21	197.00	0.5	0.5
22	201.00	0.5	0.5

Marker	Name	FGA	
17	215.00	0.5	0.5
17.2	217.00	0.5	0.5
18	219.00	0.5	0.5
18.2	221.00	0.5	0.5
19	223.00	0.5	0.5
19.2	225.00	0.5	0.5
20	227.00	0.5	0.5
20.2	229.00	0.5	0.5
21	231.00	0.5	0.5
21.2	233.00	0.5	0.5
22	235.00	0.5	0.5

1 2 3 4

Note: See column and row descriptions on the following page.

Column	Description
1	Bin name: The name of the allele at an expected location within a locus.
2	Bin center: The location of the bin center in base pairs, <i>e.g.</i> , 110±0.5 bp.
3	Bin left offset: The left boundary of the bin expressed as the base pair distance to be subtracted from the location (<i>e.g.</i> , 0.5).
4	Bin right offset: The right boundary of the bin expressed as the base pair distance to be subtracted from the location (<i>e.g.</i> , 0.5).

Row	Description
5	GeneMapper <i>ID</i> version number
6	Kit name: Name of chemistry, <i>i.e.</i> , AmpF ℓ STR_Panels_v1
7	Bin set name: The collection of all AmpF ℓ STR bin sets
8	Panel name: An AmpF ℓ STR kit
9	Marker name: The locus name

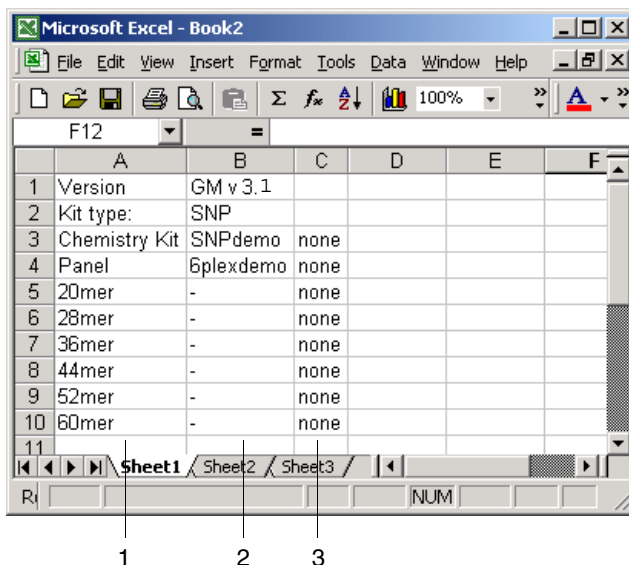
Panel Definitions for SNP Genotyping

The examples below show SNP genotyping panels defined in the format used by GeneMapper *ID* software v3.1 in a tab-delimited text file (.txt) using Notepad or WordPad or an Excel spreadsheet saved as a .txt file.

```

Version GM v 3.1
Kit type:      SNP
Chemistry kit  SNPdemo none
Panel          6plexdemo none
20mer          -      none
28mer          -      none
36mer          -      none
44mer          -      none
52mer          -      none
60mer          -      none
  
```

Column	Description
1	Marker name
2	Control allele names as comma-separated bin names (A, G) (if none, there must be a dash "-")
3	Comments (if none, write "none")

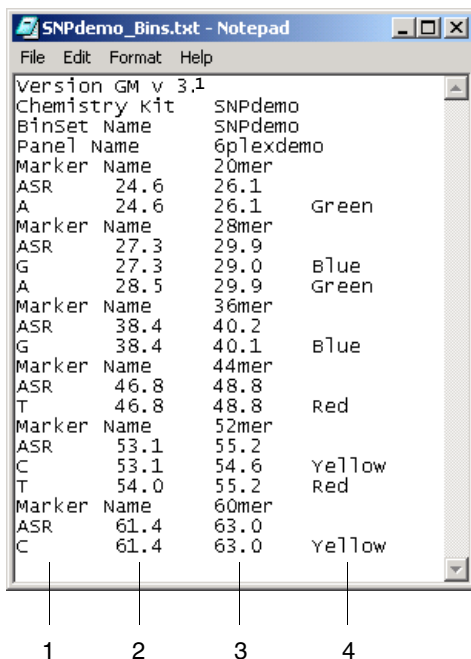


Column	Description
1	Marker name = locus name
2	Control allele names as comma-separated bin names (A, G) (if none, there must be a dash "-")
3	Comments (if none, write "none")

Bins Definitions for SNP Genotyping

The examples below show SNP genotyping bin definitions in the format used by GeneMapper *ID* software v3.1 in a tab-delimited text file (.txt) using notepad or wordpad or an Excel spreadsheet saved as a .txt file.

Note: ASR = Total allele size range. The allele size data automatically exports and then the range is printed.



Column	Description
1	Bin name: The name of the allele at an expected location within a locus.
2	Bin size range minimum
3	Bin size range maximum
4	Bin color

	A	B	C	D	E
1	Version	GM v 3.1			
2	Chemistry Kit	SNPdemo			
3	BinSet Name	SNPdemo			
4	Panel Name	6plexdemo			
5	Marker Name	20mer			
6	ASR	24.6	26.1		
7	A	24.6	26.1	Green	
8	Marker Name	28mer			
9	ASR	27.3	29.9		
10	G	27.3	29.1	Blue	
11	A	28.5	29.9	Green	
12	Marker Name	36mer			
13	ASR	38.4	40.2		
14	G	38.4	40.2	Blue	
15	Marker Name	44mer			
16	ASR	46.8	48.8		
17	T	46.8	48.8	Red	
18	Marker Name	52mer			
19	ASR	53.1	55.2		
20	C	53.1	54.6	Yellow	
21	T	54	55.2	Red	
22	Marker Name	60mer			
23	ASR	61.4	63		
24	C	61.4	63	Yellow	

Column	Description
1	Bin name
2	Bin size range minimum
3	Bin size range maximum
4	Bin color

Import and Export Files

The Panel Manager imports and exports two text file data formats, panel data and bin set data. These files are tab-delimited and the following rules apply:

- Files to be imported must have the correct number of columns in the correct order.
- On import, panel and bin set names are checked for uniqueness in the GeneMapper *ID* database and markers are checked for uniqueness within the panel that will contain them. An alert is presented if a name is not unique and such a file cannot be imported.
- If any errors are found during import, the Panel Manager does not accept data from the file.
- Lines beginning with “#” are comments and are ignored on import.
- Both Panel and Bin set file formats have a header line with a GeneMapper *ID* software version string:

Version GM *ID* v 3.1

If the first line of a file to be imported does not begin with “Version GM,” then GeneMapper *ID* software handles the import as a GeneMapper *ID* software 1.x panel import. If the file is in GeneMapper *ID* software 1.x format, then it is imported without errors. If the file is in GeneMapper *ID* software 2.x or 3.x format, the file will not be imported and the following error alert will be presented: “The format of this file is wrong. Correct the file and try again.”

If the data type label does not match the type specified in the import command, the file is not imported and an alert is presented.

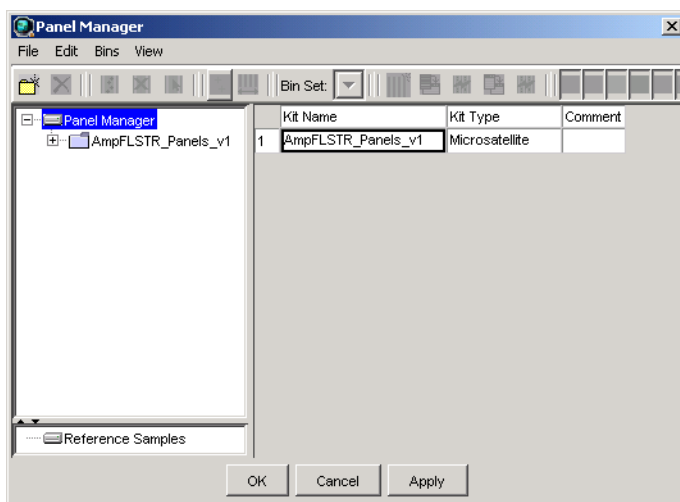
A PanelImportLog.txt file is generated during import of a panel or bin set and will list any errors encountered. This file is found in the *GMdistribution\app* folder on your hard drive.

Panel Table View

Introduction Selecting the Panel Manager icon or text in the navigation pane (left pane shown below) shows a table of the current Panel Manager folders or kits in the right portion of the window. Selecting a particular kit produces a list of panels in the right portion of the window. This list is the Panel Table view. In the Panel Table view, you can:

- Edit panel names and enter comments in the panel table
- Create new panels using the New Panel command
- Export panels and bin sets used for project analysis to backup

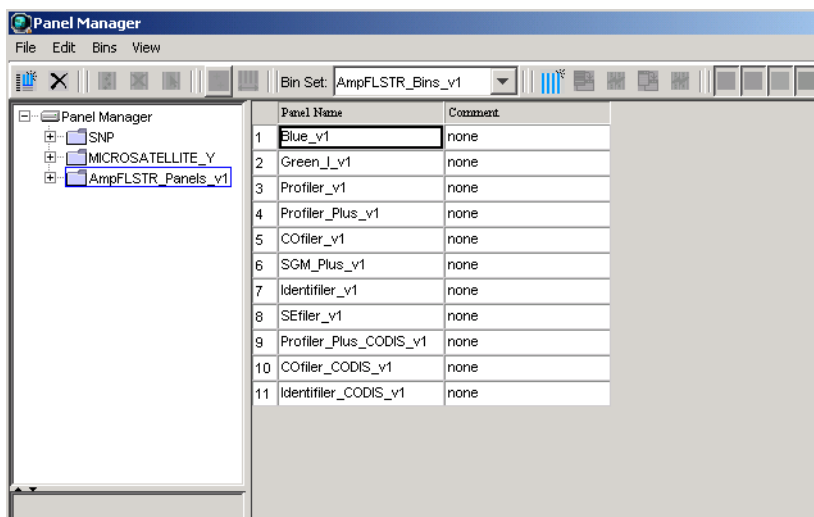
Note: Editing or deleting kits, panels, and bin sets prevents you from displaying the original data. Export any panels and bin sets that you might use at a later date, or for reanalysis of old projects.



Note: A warning alert is shown whenever kits or panels are deleted to remind you that this reference data may have been used previously in sample analysis.

Panel Table Columns

The table below lists the columns in the Panel table:



The screenshot shows the 'Panel Manager' application window. On the left is a tree view with folders for 'SNP', 'MICROSATELLITE_Y', and 'AmpFLSTR_Panels_v1'. The main area displays a table with two columns: 'Panel Name' and 'Comment'. The table contains 11 rows of data, with the first row highlighted. The 'Bin Set' dropdown at the top is set to 'AmpFLSTR_Bins_v1'.

	Panel Name	Comment
1	Blue_v1	none
2	Green_I_v1	none
3	Profiler_v1	none
4	Profiler_Plus_v1	none
5	COfiler_v1	none
6	SGM_Plus_v1	none
7	Identifier_v1	none
8	SEfiler_v1	none
9	Profiler_Plus_CODIS_v1	none
10	COfiler_CODIS_v1	none
11	Identifier_CODIS_v1	none

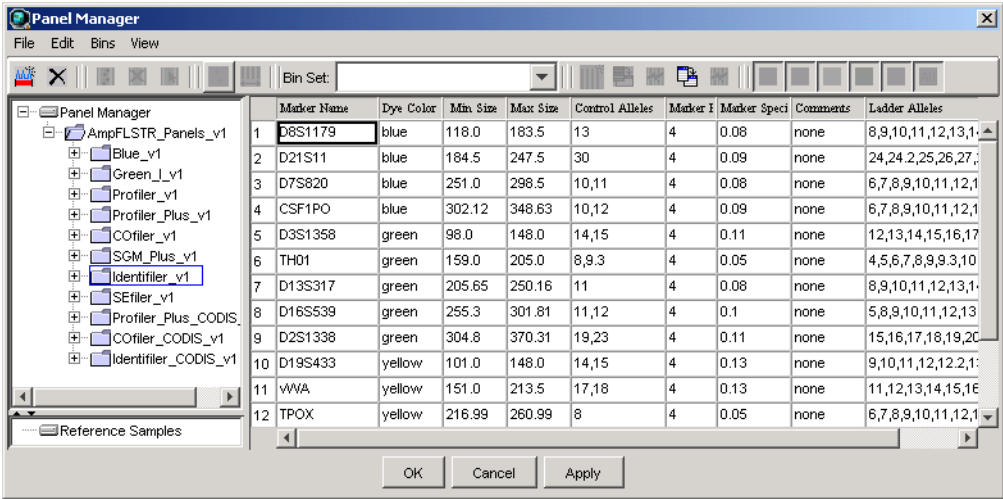
Note: All columns are resizable and editable.

Column Name	Description
Panel Name	Contains the name of the panel. Cell accepts alphanumeric characters (restricted for any invalid symbol characters – includes all Microsoft Windows invalid characters). Note: When the cell is closed, the software must check that the name is unique. An alert message will display if the name is not unique.
Comments	Contains comments regarding the panel. Cell accepts alphanumeric characters.

Panel Name Rule Panel names must be unique within a kit and across kits.

Marker Table View

Introduction Selecting a panel in the navigation pane displays the Marker table for that panel. The Marker table, shown below, enables you to view, create, and edit marker names and associated data.



This figure shows the columns in the AmpFLSTR marker table. They are further described in the following table.

Table 4-3 Marker table column descriptions:

Column	Description
Marker Name	Contains the name of the marker. Cell accepts alphanumeric characters from the keyboard. The field is accepted if the marker name is unique in the panel. Editable. Note: Marker names are case insensitive (<i>i.e.</i> , D1S83 is the same as d1s83).
Dye Color	Contains the color names blue, green, yellow, red, orange. The field is accepted if it is one of these text strings, which are not case sensitive. Editable.

Table 4-3 Marker table column descriptions: *(continued)*

Column	Description
Min Size	Contains the starting base pair number for the allele size range. Cell accepts numeric characters from the keyboard. Editable. The range defined by the Minimum Size parameter should include the stutter peaks for the smallest allele.
Max Size	Contains the ending base pair number for the allele size range. The cell accepts numeric characters from the keyboard. Editable.
Control Alleles	Contains a comma separated list of control allele names in the marker. Editable. For AmpF$\mathcal{L}$STR data analysis, the control alleles correspond to Control DNA 9947A or Control DNA 007.
Marker Repeat	Indicates a di-, tri-, or tetra-nucleotide repeat. Contains a number for the number of repeats: 2, 3, 4. The field also accepts 9 for nonrepeat containing markers. The field is valid if it contains 2, 3, 4, or 9; any other number is invalid. Editable.
Marker Specific Stutter Ratio	Column provided for AmpF$\mathcal{L}$STR markers. Contains the ratio of stutter peak to main peak. Sets the maximum value of stutter for a specific marker, as opposed to using a global value for all markers. Editable.
Comments	Contains comments regarding the marker. Cell accepts alphanumeric characters from the keyboard. Editable.
Ladder Alleles	Column provided for the allelic ladders that come with the AmpF$\mathcal{L}$STR kits. Comma-separated list of allele names in the allelic ladder. Applicable only to Human Identification (HID) genotyping or applications that use allelic ladders. Editable.

Editing Marker Tables

To edit information in the Marker Table view, highlight the cell you want to edit and enter the new information.

IMPORTANT! If you are going to make changes to one or more of the AmpF ϵ STR panels, duplicate the panel(s) first, then make changes to the copy. If you forget to make a copy, you can export and reimport the AmpF ϵ STR panels and bins to restore the recommended default values; this procedure requires you to add the panels and bins back into your analysis methods. Therefore, it is best to make a copy of the panel before you modify it.

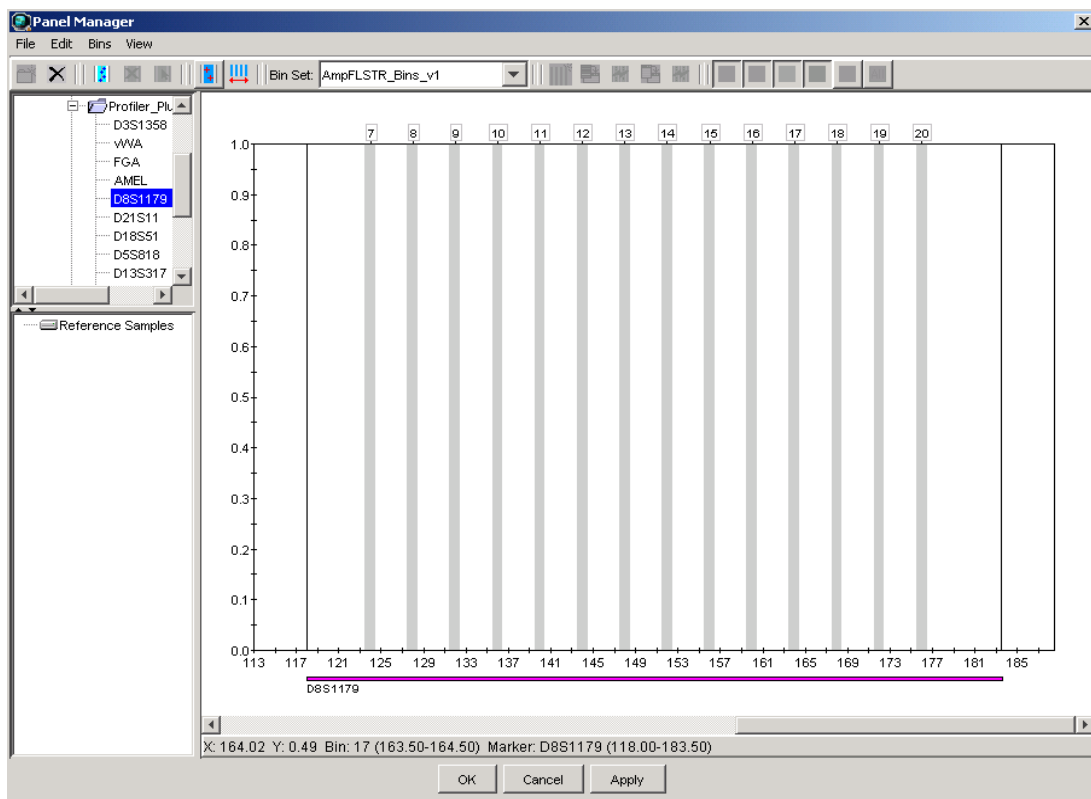
Note: Applied Biosystems has optimized the panels and bins provided for AmpF ϵ STR data analysis. Therefore, Applied Biosystems does not recommend changing the marker information. If this information is changed, Applied Biosystems requests that you analyze with the default values if applications support is needed.

Bins View - Microsatellites Only

Note The information contained in this section (Bins View) is not relevant to AmpFLSTR data analysis.

Introduction The Bins view enables you to view, create, and edit bins. The data displayed in the Bins view, imported into the Panel Manager using the Import Bin Set command, is used to create the bins associated with the marker.

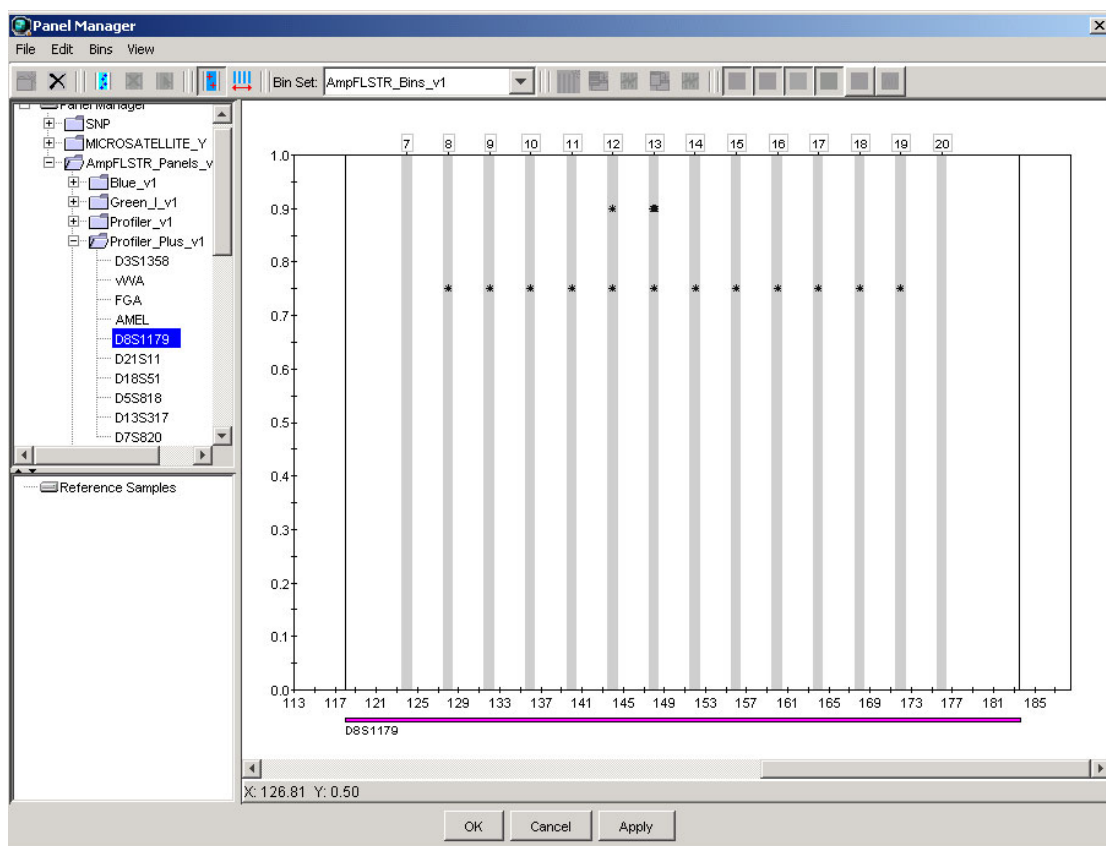
Bins allows you to average the minor variations in size that occur from run to run and capillary to capillary.



Description of the Bins View

Reference Data, Bins, and Allele Calls

Reference data includes descriptions of bins (name, location, and boundaries) and the sample allele calls used to construct the bins. In the Bins view, the vertical bars are bins that are originally associated with the markers imported as reference data. The “+” symbols shown in the figure above (marked in red on the monitor) are from the reference alleles used to build the bin set indicated in the drop-down menu at the top of the window. The red +s are shown when those alleles are used in the autobinning process or in the manual process of bin creation.



If you analyze data using a bin set like that shown above, you will see the + s as shown above. Then, when you click the Show Project Alleles icon,  the + s will be overlaid by a set of “*” symbols (marked as blue on the monitor), which represent the data associated with a particular bin for the current project in the Project window and called according to the allele associated with the bin.

Bins may be used for calling alleles with or without allele calls in the reference data, but project alleles (* s) are required for automatic bin generation (the Auto Bin function). Reference alleles (+ s) are identified as a result of the Auto Bin command.

Selecting a reference data sample in the navigation pane displays that particular sample’s electropherogram with bins overlaid to provide easy viewing while editing markers.

X- and Y-axes, Genotype Quality Symbols, and Cursor Location Information

The Bins view X-axis is the fragment size in base pairs; the Y-axis is quality, or signal height if a reference sample is selected. Vertical shaded bars are the bins that will be used to call alleles for the selected marker. The symbols (blue stars), described above as Allele Call indicators, represent Genotype Quality values on the Y-axis for the alleles of samples that have been analyzed and are currently displayed in the open project. The equivalent positions in the reference data are marked with red + s.

The Genotype Quality values, representing analysis results, are not editable. Bin location, boundaries, and names are editable. Cursor location in base pairs for any selected bin are shown under the X-axis to the left. The Bin Sets drop-down menu, located just above the Bin view pane, selects the bin set that is applied to the marker.

Marker Allele Size Range (ASR)

Reference data for bins includes the marker ASR. This is the size range of bins in a marker to allow multiplexing of markers in a sample.

ASR boundaries can be edited in the marker table or in the Bins view by adjusting the ASR handles on the right and left hand sides of the screen. The bins in a marker must not extend beyond the marker ASR. When bins are created and viewed in the Bins view, they are restricted so that their range does not extend beyond the marker ASR. The Show Alleles command may show new allele calls for a marker

outside that marker's ASR, but bins cannot be created or extended to include alleles outside the ASR. To create bins for alleles that fall outside the ASR, edit that marker's ASR, click **Apply**, and add a new bin.

Bins must not overlap in a microsatellite marker. When bins are created and edited, their locations and offsets are restricted to avoid overlap.

Import and export commands for panels and bin sets are enabled in the table views of the Panel Manager. Imported bin set reference data includes the bin boundaries but not sample reference data (the "+ s").

Using GeneMapper Manager

5

This chapter provides information on how to use the GeneMapper Manager window of the GeneMapper™ ID Software Version 3.1.

This chapter covers:

Introduction	5-2
Section 5.1 Projects Tab	5-3
Section 5.2 Analysis Methods Tab	5-9
Section 5.3 Table Settings Tab	5-27
Section 5.4 Plot Settings Tab	5-41
Section 5.5 Creating and Evaluating a Matrix	5-53
Section 5.6 Size Standards Tab.	5-65

Introduction

The GeneMapper Manager is a centralized interface within the GeneMapper *ID* software v3.1 used for managing and organizing projects and other various information. Six tabs assist you in customizing your data analysis.

- Projects tab
- Analysis Methods tab
- Table Settings tab
- Plot Settings tab
- Matrices tab
- Size Standards tab

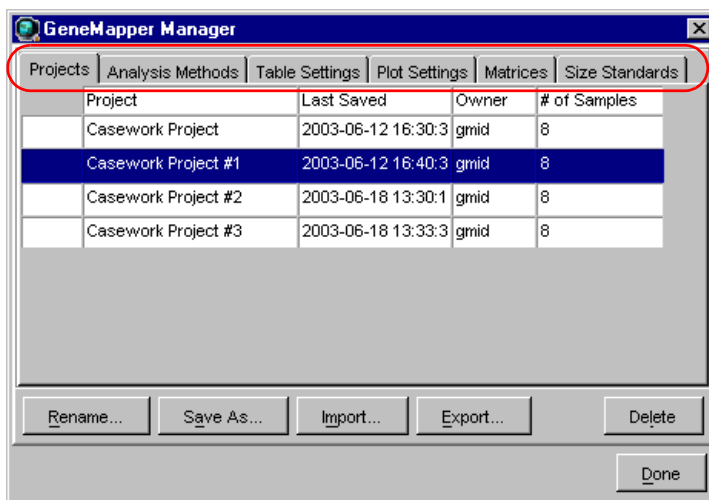


Figure 5-1 Tabs of the GeneMapper Manager

Section 5.1 Projects Tab

Overview The GeneMapper Manager Projects page, shown below, is a utility used to rename, save as, import, export, or delete projects. To display the Projects tab, select **Tools > GeneMapper Manager > Projects**.

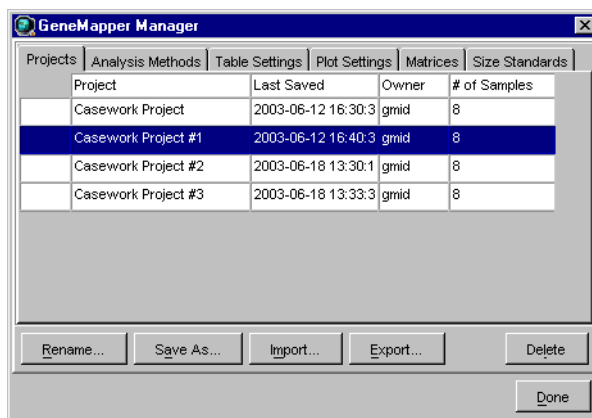


Figure 5-2 Projects Tab of the GeneMapper Manager

Select the project you would like to change and click the appropriate button. If you click the Delete button, the GeneMapper Deletion Alert box displays. Click **No** if you do not want to delete the project. Click **Yes** only if you want to delete the project permanently.

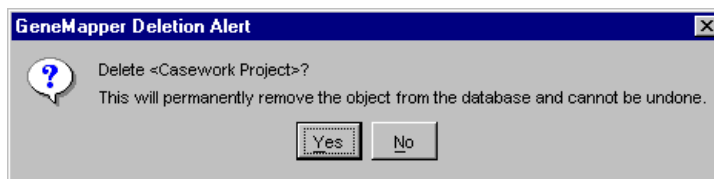


Figure 5-3 GeneMapper Deletions Alert Box

Projects Tab Elements

Table 5-1 Description of elements on the Projects tab

Item	Description
Project column	Project name
Last Saved column	Date/Time stamp showing when the project was last saved
Owner column	User name of the person who created the project
Number of Samples column	Number of samples contained in the project
Rename button	Opens a Rename dialog box for renaming the project Enabled when a project is selected
Save As button	Displays the Save Project As dialog box Enabled when a project is selected
Import button	Displays a dialog box for Importing projects
Export button Note: This exports projects only, not the sample files.	Displays a dialog box for Exporting selected projects Enabled when projects are selected Note: Projects can be exported as a group by holding down the shift key and selecting multiple projects. This feature works with one or more selected projects.
Delete button	Deletes the selected projects Enabled when a project is selected Note: This feature works with one or more selected projects.
Done button	Closes the GeneMapper Manager

Export Project Window

Use the Export Project window to export a project out of the GeneMapper *ID* database into a specified location. See an explanation of each area on this window in [“Using the Export Project Window” on page 5-6](#).

Note: The only method of backing up your projects is to **export** them.

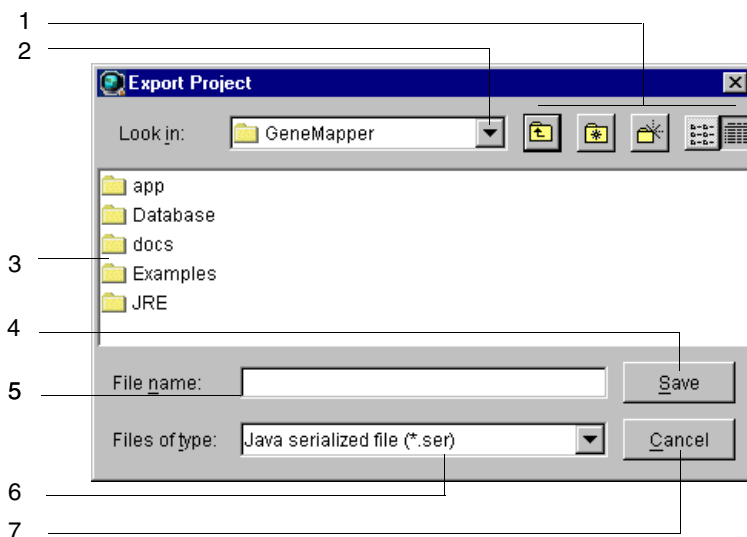


Figure 5-4 Export Project Window

Note: The sample files, analysis method, size standard, panel, and bin set used to analyze a project are **not** exported with the projects.

Using the Export Project Window

The elements called out in the figure in “Export Project Window” on [page 5-5](#) are described in the table below.

Table 5-2 Export Project Callouts

Item	Name	Description
1	Toolbar	Click these icons to:
		 Move display up one level in main pane.
		 Move display to Home level in main pane. This level is usually Profiles\<user>.
		 Create a new folder at the present directory level.
		 Present a list of the contents of the selected folder.
		 Present details of the selected folder.
2	Drop-down directory menu	Select drive letter and/or folder.
3	Folder/file display pane	The contents of the folder selected in the toolbar are displayed here.
4	Save button	Saves the exported project.
5	File name field	Enter a file name to use for saving your data.
6	Files of type field	This field is a display filter for files (folders are always shown). Use to select the file extensions for display: • All Files (*.*) shows all files • Java Serialized file (*.ser) shows only files with the extension .ser, which is how GeneMapper ID projects are exported. Note: Only one preset file extension (.ser) can be used.
7	Cancel button	Closes the Export Project window without exporting a project.

Import Project Window

The Import Project window enables you to import project data previously saved. See an explanation of each area in [“Using the Import Project Window” on page 5-8](#).

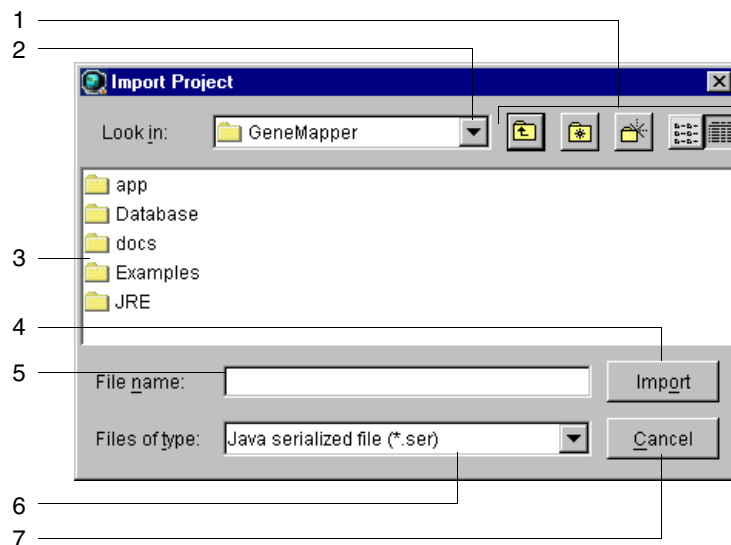


Figure 5-5 Import Project Window

Using the Import Project Window

The elements called out in the figure in “Import Project Window” on page 5-7 are described in the table below.

Table 5-3 Import Project Callouts

Item	Name	Description
1	Toolbar	Click these icons to:
		 Move display up one level in main pane.
		 Move display to Home level in main pane. This is usually “Profiles\<user>”.
		 Create a new folder at the present directory level.
		 Present a list of the contents of the selected folder.
		 Present details of the selected folder
2	Drop-down directory menu	Select drive letter and/or folder.
3	Folder/file display pane	The contents of the folder selected in the toolbar are displayed here.
4	Import button	Imports the selected project.
5	File name field	Enter the name of selected project being imported.
6	Files of type field	This is a display filter for files (folders are always shown). Use to select the file extensions for display: - All Files (*.*) shows all files - Java Serialized file (*.ser) shows only files with the extension .ser, which is how GeneMapper <i>ID</i> database projects are exported. Note: Only one preset file extension (.ser) can be used.
7	Cancel button	Closes the Import Project window without importing a project.

Section 5.2 Analysis Methods Tab

Overview The Analysis Methods Tab is used to create custom analysis methods for analyzing sample files. An analysis method is a set of algorithm parameters that are applied to data during analysis. For additional information about algorithms see [Appendix B, “Software Genotyping Algorithms.”](#)

The Analysis Methods tab contains a list of analysis methods in the database. To display the Analysis Methods tab, select **Tools > GeneMapper Manager > Analysis Methods**.

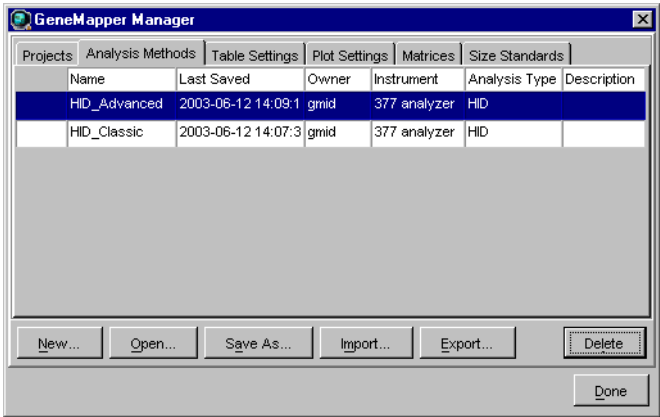


Figure 5-6 Analysis Methods Tab of the GeneMapper Manager

Analysis Methods Tab Elements

Table 5-4 Description of elements on the Analysis Methods tab

Item	Description
Name column	Analysis method name
Last Saved column	Date/Time stamp showing when the analysis method was last saved
Owner column	User name of the person who created the analysis method
Instrument column	The type of instrument used

Table 5-4 Description of elements on the Analysis Methods tab

Item	Description
Analysis Type column	Identifies the analysis mode of the analysis method (<i>i.e.</i> , HID, Microsatellite, SNP Genotyping)
New button	Opens the New Analysis Method dialog box. Always enabled. This dialog box asks you to select the analysis type before opening the Analysis Method Editor. The analysis type you select: • Sets the analysis algorithm • Displays the appropriate fields for that type of analysis in the Analysis Method Editor dialog box.
Open button	Opens the editor for a selected analysis method. Enabled when a analysis method is selected.
Save As button	Displays the Save As dialog box. Enabled when an analysis method is selected.
Import button (.xml file type)	Displays a dialog box for Importing analysis methods. Always enabled.
Export button (.xml file type)	Displays a dialog box for Exporting selected analysis methods. Enabled when one or more analysis methods are selected.
Delete button	Deletes the selected analysis method(s).
Done button	Closes the GeneMapper Manager.

**Default Analysis
Methods**

GeneMapper *ID* software v3.1 contains the following standard Applied Biosystems profiles:

Default	Analysis Type	Bin Set	Detection Algorithm
Microsatellite	Microsatellite	AmpF $\mathcal{L}$ STR_Bins_v1	Classic Advanced
SNP Genotyping	SNP Genotyping	None	Basic

Note: These profiles can be altered but should not be deleted. They are intended to be used as models for customizing new analysis methods.

Note: The *GeneMapper™ ID Software v3.1 Human Identification Analysis Tutorial* (PN 4335523) shows you how to create methods that are used as a guide for AmpF $\mathcal{L}$ STR data analysis.

Analysis Method Editor

Introduction

The Analysis Method Editor allows you to create or edit analysis methods to be used for analyzing your sample files. The analysis method determines the type of application being analyzed and allows for customization of the peak detection algorithm and quality value settings.

Description of the Analysis Method Editor Process

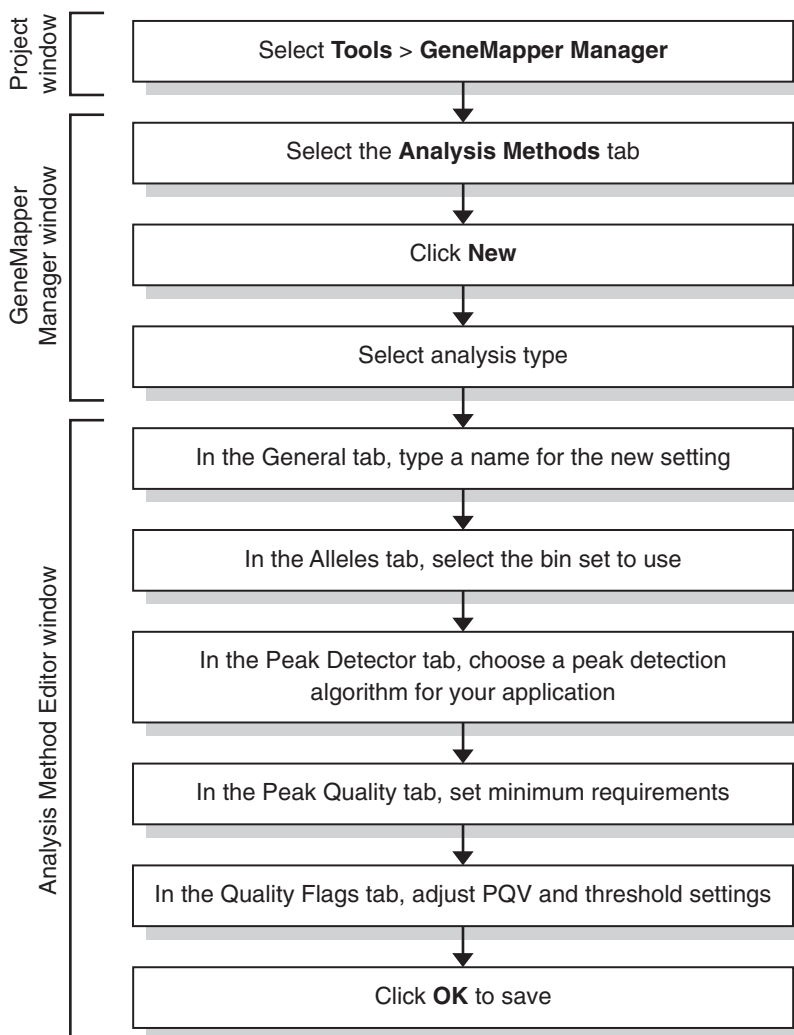


Figure 5-7 Analysis Method Editor Process

Accessing the Analysis Method Editor

Access the Analysis Method Editor in the following ways:

- Select **Tools > GeneMapper Manager > Analysis Methods Tab > New or Open**
- In the Project Window, click the **Samples** tab. Click **New Analysis Method** to create a new method, or double click an existing method to edit it.
- Select a sample in the project window and click the Analysis Method Editor icon on the toolbar. Edit the currently selected samples' analysis method.

Note: The Analysis Method Editor icon is not active unless a sample with an analysis method is selected.

Analysis Method Editor - HID

General | **Allele** | **Peak Detector** | Peak Quality | Quality Flags

Peak Detection Algorithm: **Advanced**

Ranges

Analysis	Sizing
Partial Range	Partial Sizes
Start Pt: 2700	Start Size: 75
Stop Pt: 7600	Stop Size: 450

Smoothing and Baseline

Smoothing: ☐ None ☒ Light ☐ Heavy

Baseline Window: 51 pts

Size Calling Method

☐ 2nd Order Least Squares
☐ 3rd Order Least Squares
☐ Cubic Spline Interpolation
☒ Local Southern Method
☐ Global Southern Method

Peak Detection

Peak Amplitude Thresholds:

B: 50	R: 50
G: 50	O: 50
Y: 50	

Min. Peak Half Width: 2 pts
 Polynomial Degree: 3
 Peak Window Size: 15 pts

Slope Threshold

Peak Start: 0.0
Peak End: 0.0

Factory Defaults

OK Cancel

Figure 5-8 Analysis Method Editor

New Analysis Method Dialog Box

When you create a new analysis method, the following dialog box opens prior to the analysis method editor. This dialog box allows you to select the analysis type for your samples, and controls the items available for customization within the Analysis Method Editor tab.

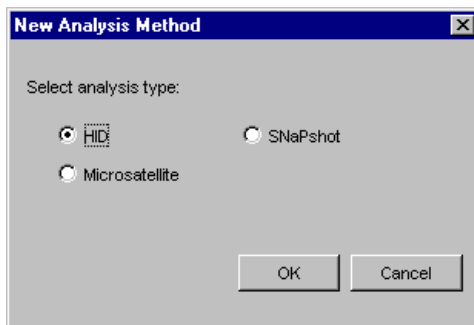
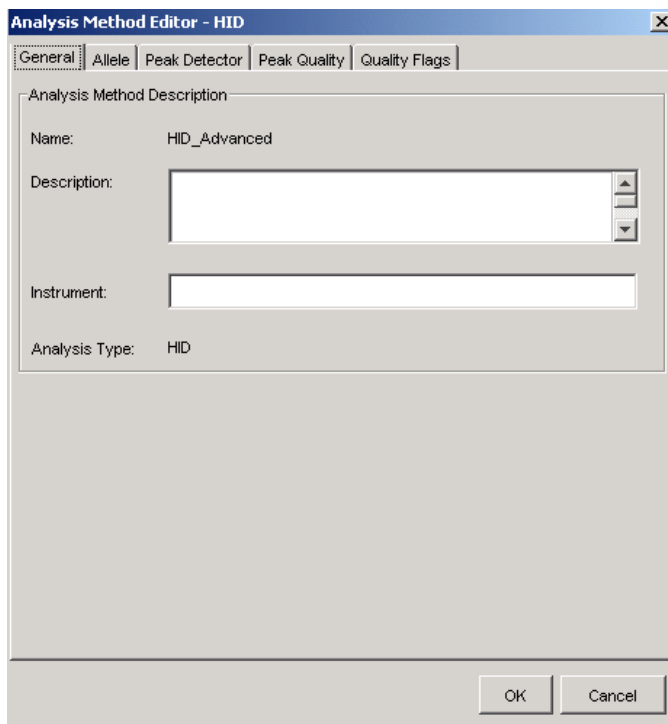


Figure 5-9 New Analysis Method Dialog Box

Note: Select HID for AmpF $\mathbb{L}$ STR data analysis.

General Tab In the General tab, enter a name and description for the analysis method. Create an analysis method description for your specific platform (*i.e.*, capillary or gel-based electrophoresis). Capillary and gel-based electrophoresis may require different start and stop points in the analysis range or peak amplitude thresholds.



The screenshot shows a dialog box titled "Analysis Method Editor - HID". It has five tabs: "General", "Allele", "Peak Detector", "Peak Quality", and "Quality Flags". The "General" tab is selected. Inside the "General" tab, there is a section titled "Analysis Method Description". This section contains four fields: "Name:" with the value "HID_Advanced", "Description:" with an empty text box and vertical scroll arrows, "Instrument:" with an empty text box, and "Analysis Type:" with the value "HID". At the bottom right of the dialog box are "OK" and "Cancel" buttons.

Figure 5-10 General Tab of the Analysis Method Editor

Note: The Analysis Type reflects the choice selected in the New Analysis Method dialog box.

Allele Tab In the Allele tab, you can set a number of parameters controlling allele calling, including:

- Bin Set
- Marker Repeat Type (for microsatellites)
- Amelogenin Cutoff value
- Range Filter

Note: Applied Biosystems recommends that you leave all other parameters in the default setting after selecting the AmpF[®]STR bin set for data analysis.

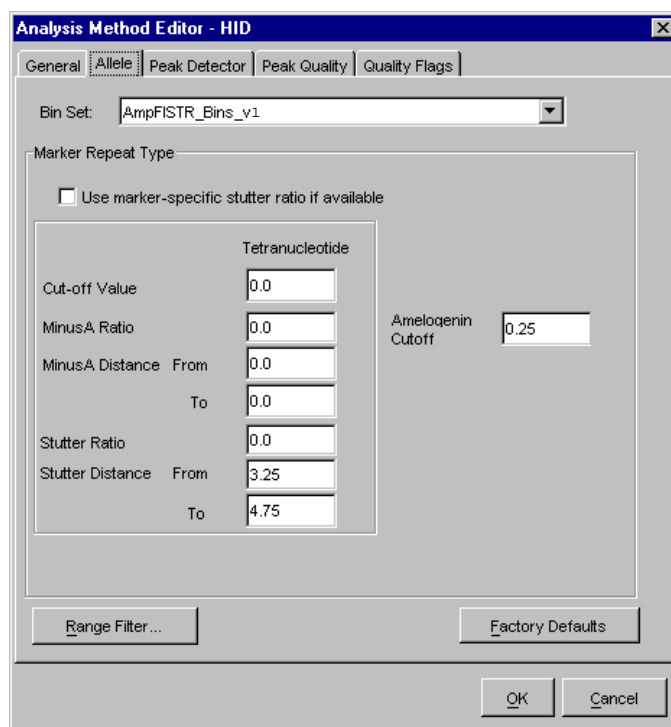


Figure 5-11 Allele Tab of the Analysis Method Editor

Bin Set

Bins are a way of matching your data to the allele definitions contained in the panel. Bins are the locations in which you expect an allele to size. Alleles are defined in bins centered around the average size in base pairs, for example 110.00 ± 0.5 bp. Sample peaks that fall within a bin are genotyped accordingly.

You must choose a bin set for your analysis in order to guide the algorithms in assigning allele calls. This drop-down list enables you to choose the bin set to be used for analysis. The bin set chosen for analysis must match the kit/panel chosen for analysis.

Note: For AmpF $\mathbb{L}$ STR analysis, you import the bin set into Panel Manager before you select it from the menu. For **non**-AmpF $\mathbb{L}$ STR analysis, you create the bin set in Panel Manager before it appears as an option in the drop-down menu.

Marker Repeat Type (for AmpF $\mathbb{L}$ STR and other microsatellites *only*)

As stated on the Allele tab view, no entries are required for dinucleotide repeats because the application calculates them automatically.

Check the “Use marker-specific stutter ratio if available” check box to insert stutter ratio information into the Panel Manager marker’s table. If you know the stutter ratio for an individual marker, you can set this number under the “Marker Specific Stutter%” column in the Panel Manager. Checking this box on the Allele tab makes the algorithm use the stutter ratios you previously defined in Panel Manager and not those defined in the Stutter Ratio box and the Allele tab. Different values can be used for each marker.

IMPORTANT! Use the marker-specific stutter ratio for AmpF $\mathbb{L}$ STR kits. These values are provided in panels and bins supplied by Applied Biosystems.

The “Cutoff value” ignores all peaks less than the cut-off ratio of the largest peak in the allele size range.

The “MinusA ratio” and “Stutter ratio” refer to the ratios you are expecting. For example, if you expect a stutter percentage of 20% for your markers, enter in 0.2.

The following table provides information about the ranges of possible values.

Table 5-5 Value ranges for tetranucleotide:

Parameter	Min	Max	Tetranucleotide Default
Cut-off value	0.0	1.0	0.0
MinusA ratio	0.0	1.0	0.0
MinusA distance	0.0	–	0.0
Stutter ratio	0.0	1.0	0.0
Stutter distance	0.0	–	from 3.25 to 4.75 bp
Amelogenin cut-off	0.0	1.0	0.25

Range Filter

The Range Filter allows you to remove labels from peaks within a specific size range for each color.

Peak Detector Tab

Use the Peak Detector tab to select the algorithm to use for detecting peaks:

- Advanced
- Classic

The Peak Detector tab with Advanced and Classic peak detection algorithm settings are described in the following pages. See [Appendix B, “Software Genotyping Algorithms,”](#) for additional information on Advanced and Classic peak detection algorithms.

Advanced Peak Detection Algorithm

This algorithm is similar to the method of analyzing size standards and performing peak detection that is used in the ABI PRISM® GeneScan® software on the Windows NT operating system. Information on the individual parameters can be found in [Appendix B, “Software Genotyping Algorithms.”](#) Information about ranges and peak amplitude thresholds is as follows:

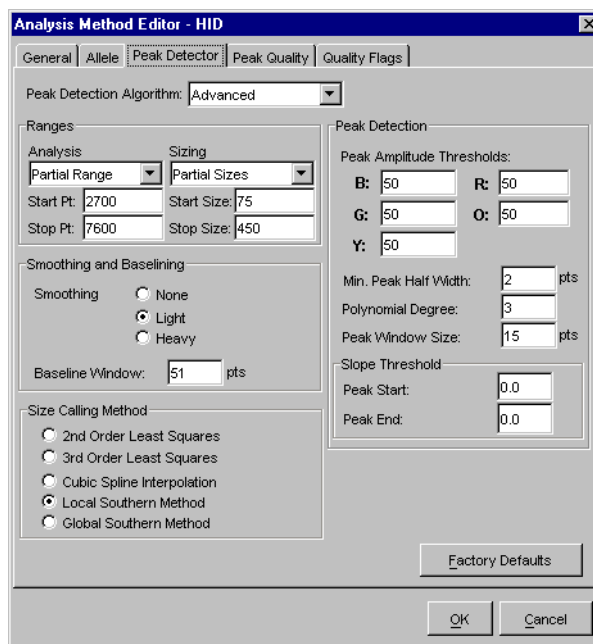


Figure 5-12 Peak Detector Tab of the Analysis Method Editor – Advanced Peak Detection Algorithm

- **Analysis Range** (defined in scans, not by size)

Full Range - the software analyzes the raw data in the entire scan region as collected by the genetic analysis instrument, including the primer peak.

Partial Range - the software analyzes data between the user defined scan points. When this option is selected, Applied Biosystems recommends that you select a Start Point after the primer peak and before the first size standard peak you want to define. The Stop Point should be after the last size standard fragment you define. This range may vary from instrument to instrument and platform to platform.

- **Sizing Range** (in base pairs)

Full Range – the software reports all peaks within the range of the matched size standard in the tabular data.

Partial Sizes – the software reports peaks within this user defined size range in the tabular data.

Note: To ensure successful sizing, the analysis range should contain all size standard fragments includes in the sizing range.

For AmpF $\mathcal{L}$ STR data analysis, follow the below chart for recommended sizing ranges.

Table 5-6 Recommended Sizing Ranges for AmpF $\mathcal{L}$ STR Data Analysis

Kit	Sizing Range
• AmpF$\mathcal{L}$STR[®] Profiler Plus[®] kit • AmpF$\mathcal{L}$STR[®] COfiler[®] kit • AmpF$\mathcal{L}$STR[®] Profiler[®] kit • AmpF$\mathcal{L}$STR[®] Blue[™] kit • AmpF$\mathcal{L}$STR[®] Green[™] kit • AmpF$\mathcal{L}$STR[®] SGM Plus[®] kit	75—400
• AmpF$\mathcal{L}$STR[®] Identifiler[®] kit • AmpF$\mathcal{L}$STR[®] SEfiler[®] kit	75—450

- **Peak Amplitude Thresholds**

The values you enter for each color represent the minimum peak height (in relative fluorescent units) that the software will detect and size. The software ignores any peak heights below this value. For all dyes, the default is 50.

For more information and a detailed description of all other parameters, refer to [Appendix B, “Software Genotyping Algorithms.”](#)

Note: Applied Biosystems recommends that you use the default settings for all other analysis parameters for AmpF Φ STR data analysis.

Classic Peak Detection Algorithm

This algorithm is similar to the GeneScan software on the Macintosh operating system method of analyzing size standards and performing peak detection.

The screenshot shows the 'Analysis Method Editor - HID' dialog box with the 'Peak Detector' tab selected. The 'Peak Detection Algorithm' is set to 'Classic'. The 'Ranges' section includes 'Analysis' (Full Range) and 'Sizing' (All Sizes) dropdowns, with 'Start Pt' and 'Stop Pt' fields set to 0 and 10000 respectively, and 'Start Size' and 'Stop Size' fields set to 0 and 1000 respectively. The 'Data Processing' section has 'Baseline' and 'MultiComponent' checked, and 'Smoothing' set to 'Light'. The 'Size Calling Method' section has 'Local Southern Method' selected. The 'Peak Detection' section includes 'Peak Amplitude Thresholds' (B: 50, R: 50, G: 50, O: 50, Y: 50), 'Min. Peak Half Width' set to 3 pts, and 'Split Peak Correction' set to 'No Peak Correction'. The 'Correction Limit' is set to 30 pts. There are 'Factory Defaults', 'OK', and 'Cancel' buttons at the bottom.

Figure 5-13 Peak Detector Tab of the Analysis Method Editor – Classic Peak Detection Algorithm

The ranges and peak amplitude thresholds are identical to those for the advanced algorithm. Refer to [“Advanced Peak Detection Algorithm” on page 5-19](#) for details. Refer to [Appendix B, “Software Genotyping Algorithms,”](#) for additional information.

Note: Applied Biosystems recommends that you use the default settings for all other analysis parameters for AmpF $\mathbb{L}$ STR data analysis.

Peak Quality Tab

The Peak Quality tab allows the user to set thresholds that determine the PQVs for certain peak metrics. The PQV flags green if the actual value for that item is above the user defined threshold. A PQV flags yellow if the actual value falls below the user-defined threshold. These PQVs are used in the calculation of the overall Genotype Quality. When you click the Peak Quality tab, the following page opens.

The screenshot shows the 'Analysis Method Editor - HID' dialog box with the 'Peak Quality' tab selected. The dialog has five tabs: General, Allele, Peak Detector, Peak Quality, and Quality Flags. The Peak Quality tab contains several sections with input fields:

- Signal level:**
 - Homozygous min peak height: 200.0
 - Heterozygous min peak height: 100.0
- Heterozygote balance:**
 - Min peak height ratio: 0.7
- Peak morphology:**
 - Max peak width (basepairs): 1.5
- Pull-up peak:**
 - Pull-up ratio: 0.05
- Allele number:**
 - Max expected alleles: 2

At the bottom right of the dialog is a 'Factory Defaults' button. At the very bottom are 'OK' and 'Cancel' buttons.

Figure 5-14 Peak Quality Tab of the Analysis Method Editor

The settings include the following:

- Signal level (in fluorescent units)
 - Homozygous min peak height (default = 200)
 - Heterozygous min peak height (default = 100)

Peaks between the minimum peak height and the signal peak height are sized and called, but the peak height flag is yellow, and the PQV value will be lowered as a result.

- Heterozygote Balance
 - Min peak height ratio (default = 0.5)

In a heterozygous allele, the higher peak height is X and the lower peak height is Y. If the ratio Y/X is less than the heterozygote balance, then the process quality value flag for peak height ratio will be triggered and the GQ (genotype quality) value will be lower.

- Peak Morphology
 - Max peak width (basepairs) (default = 1.5)
- Pull-up peak
 - Pull-up ratio (default = 0.05)

Considers the ratio of any color peak directly under a major peak. Flags spectral pull-up quality value as in the genotypes table.

- Allele Number
 - Max expected allele number (default = 2)

You may want to leave the default setting at 2 so that you are flagged when there is a possible mixture, especially in casework. If analyzing known mixtures or polyploids, enter the maximum number of alleles you expect. In order to display all alleles, you must enter the correct number of alleles into the table profile.

- SNP (for SNP genotyping application only)
 - Double peak (default = 0.5)

(See [“PQV Flags on the Genotypes Page”](#) in Appendix A.)

Quality Flags Tab PQV Thresholds

Analysis Method Editor - HID

General | Allele | Peak Detector | Peak Quality | **Quality Flags**

Quality weights are between 0 and 1.

Quality Flag Settings

Spectral Pull-up	0.8	Control Concordance	1.0
Broad Peak	0.8	Low Peak Height	0.3
Out of Bin Allele	0.8	Off-scale	0.8
Overlap	0.8	Peak Height Ratio	0.3

PQV Thresholds

	Pass Range:	Low Quality Range:
Sizing Quality:	From 0.75 to 1.0	From 0.0 to 0.25
Genotype Quality:	From 0.75 to 1.0	From 0.0 to 0.25

Figure 5-15 Quality Flags Tab of the Analysis Method Editor

The ranges you specify in this section determine the flag assigned to the sizing quality and genotyping quality only. These two flags are independent of one another; Therefore, the thresholds are set independently. The software calculates a numeric value from 0 to 1 to represent the sizing quality and genotype quality based on certain criteria and/or PQVs. You can then determine a numeric low-quality range and pass range. For example, if you use the default threshold settings shown in the figure below, any results over 0.75 pass and any results below 0.25 are of low quality. Any results between 0.26 and 0.74 are marked to check.

Use the Quality Flags tab to set the importance level of the PQVs available for HID and SNP genotyping analysis. The individual PQVs on this page are used in the calculation to determine the overall genotype quality. This tab allows you to determine how much weight is applied to each PQV in this calculation.

Use the options under PQV Thresholds to set Pass/Check/Low Quality thresholds for the Sizing Quality (SQ) parameter for the Samples view and the Genotype Quality (GQ) parameter for the Genotypes view. Anything that is not labeled Pass  or Low Quality  will be labeled as check .

Note: Applied Biosystems recommends that you use the default settings for AmpF[®]STR data analysis.

Options

Data entry restrictions for the PQV thresholds ensure that the lower limit of the Pass range is always greater than the upper limit of the Low Quality range. The Check range is the interval between Pass and Low Quality, if any. For example, if you use the default threshold settings shown in the figure above, any result over 0.75 passes, any result at 0.25 or below is of low quality, and any result from 0.26 to 0.74 will be marked for you to check. (See [Appendix A, “Process Quality Values.”](#))

When the default setting for the Quality Metrics Display (Analysis tab in Preferences window) is “Symbols,” a Pass for the SQ and GQ parameters is indicated by a green square , a Check is indicated by a yellow triangle , and a Low Quality is indicated by a red octagon .

Some quality metrics are either Pass or Low Quality (for example, Background Peak), and some are warning flags with either Pass or Check values (for example, File Not Found). Such values are not adjustable by the Threshold parameter.

Section 5.3 Table Settings Tab

Overview Use the Table Settings tab to perform the following tasks:

- Create new table profiles for viewing and exporting
- Hide and show table columns
- Filter the table entries (rows) in Project windows

The Table Settings tab allows you to determine what columns are viewed in the Samples and Genotypes tab, and set filtering properties.

The Table Settings tab contains a list of Table Settings in the database. To display the Table Settings tab, select **Tools > GeneMapper Manager > Table Settings**.

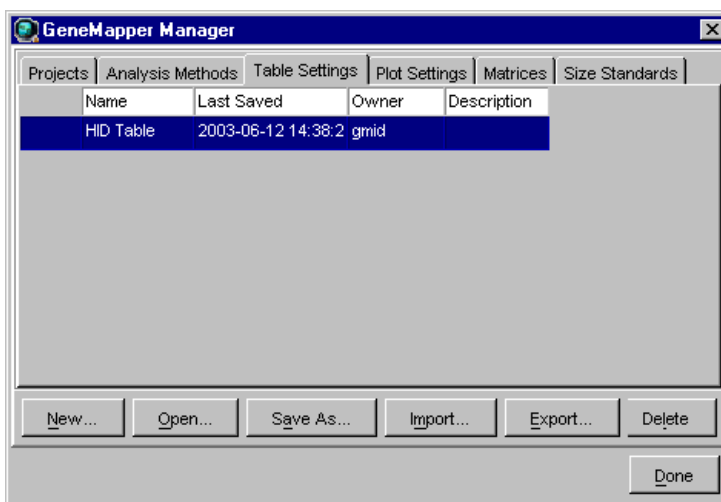


Figure 5-16 Table Settings Tab of the GeneMapper Manager

Table Settings Tab Elements

Table 5-7 Description of elements on the Table Settings Tab

Item	Description
Name column	Table Setting name
Last Saved column	Date/Time stamp displaying when the object was last saved
Owner column	User name
Description column	Description of the object
New button	Opens the Table Setting Editor window Always enabled
Open button	Opens the editor for selected table settings Enabled when a single object is selected
Save As button	Displays the Save As dialog box Enabled when a single object is selected
Import button	Displays a dialog box for Importing objects Always enabled (.xml file)
Export button	Displays a dialog box for Exporting selected objects Enabled when one or more objects are selected (.xml file)
Delete button	Deletes the selected object(s)
Done button	Closes the GeneMapper Manager

Default Table Settings

Applied Biosystems provides three default tables for each application:

- AmpFLSTR Default
- Microsatellite Default
- SNP Default

You need to import these default tables into the database through the Table Settings tab.

To import a default table:

1. Press **Import**. A dialog box opens.
2. Double-click the **Tables Settings** folder to display all of the default tables.
3. Select the default table you wish to import, then click **Import**. The default table appears in the Table Settings tab.

Note: These profiles can be used as models for customizing new profiles.

Table Settings Editor

Introduction When you click New or Open on the Table Settings tab, the Table Settings Editor window opens. Use the Table Settings Editor to show or hide information specific to your sample, add filtering capabilities, and give content information for each column in the project window.

Note: You must have a table highlighted to activate the Open button.

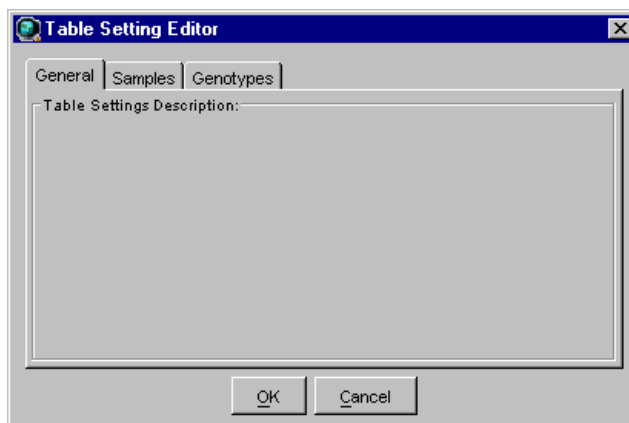


Figure 5-17 Table Settings Editor

The Table Settings Editor window contains the following tabs:

- General
- Samples
- Genotypes

The General Tab The General tab provides the capability to give a name and description to a new profile.

Note: You cannot change the name of an existing profile, only the description.

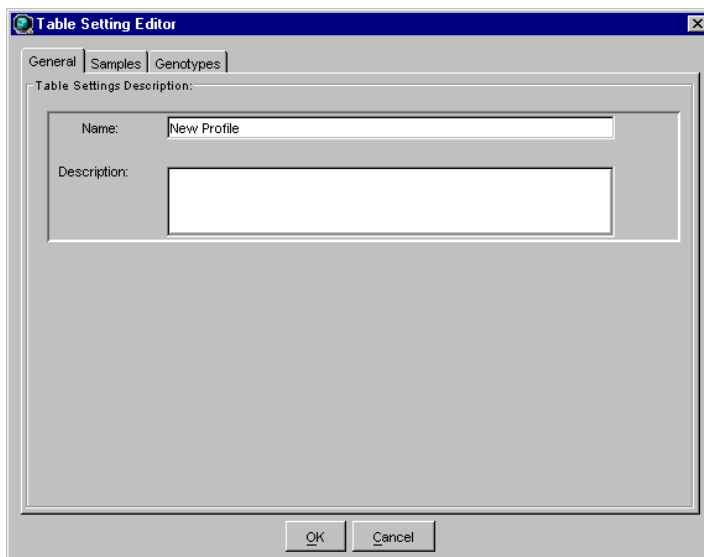


Figure 5-18 General Tab of the Table Settings Editor

The Samples Tab The Samples tab provides the capability to filter columns in the Samples table of the project window.

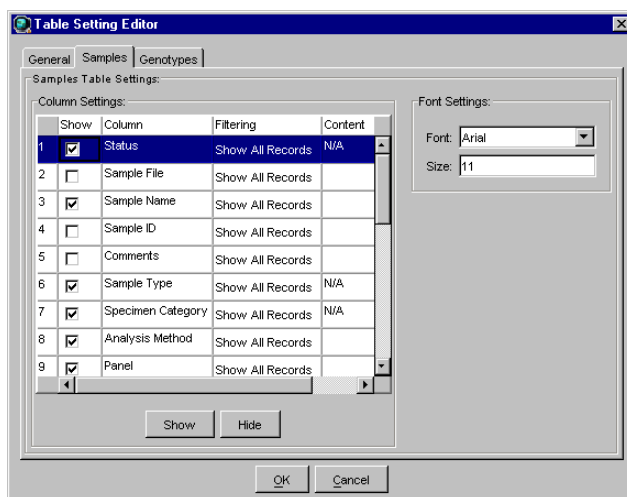


Figure 5-19 Samples Tab of the Table Settings Editor

The Genotypes Tab The Genotypes tab provides the capability to filter columns in the Genotypes table of the project window and the capability to control the allele settings.

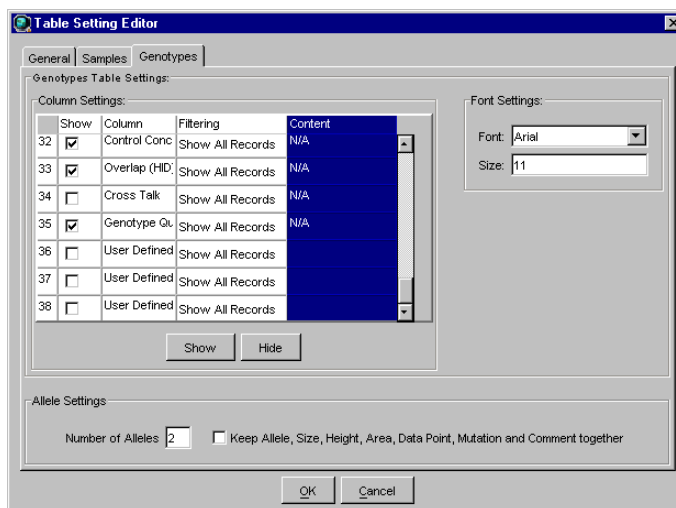


Figure 5-20 Genotypes Tab of the Table Settings Editor

The Sample and Genotypes Tab Elements

The Samples and Genotypes tabs display the Table Setting properties that will be applied to the corresponding tabs in the Project window. The properties or elements are described in the table below.

Samples and Genotypes Settings

Table 5-8 Elements and settings for samples and genotypes data:

Element	Description
Column settings	These settings contain the valid columns for either Samples or Genotypes data. • Show column - List with an editable check box for each Project column, controlling whether the column is shown or hidden • Column column - List of column headings for the Project window • Filtering column - List of filtering properties for each Project window column • Content column - Sets what information is displayed in each Project window column as selected by the appropriate filtering method. "N/A" indicates that no comments can be made for these rows.
Filtering Properties (Summary) (For more specific information, see "Filtering Controls" on page 5-35.)	The filtering properties for a Project table column are controlled in the Filtering and Content columns. Modifying these settings updates the property in the Column/Filter table. For more information, see "Filtering Controls" on page 5-35.
OK	Closes the Table Setting Editor window and applies any pending changes to the Project window. Contents of the Table Setting Editor are saved to the GeneMapper ID database.
Cancel	Closes the Table Setting Editor window without making any pending changes.
Show	Selects all highlighted rows as visible.
Hide	Selects all highlighted rows as hidden.

Allele Settings

The Allele Settings box on the Genotypes page of the Table Setting Editor window controls how the Allele, Size, Height, Area, Mutation and Comment columns are displayed in the Genotypes view.

Table 5-9 Allele settings and descriptions:

Allele Settings	Description
Number of Alleles	The number of alleles per marker that are displayed in the genotypes table. The default value is 2. However, when you are analyzing mixtures or want to have more than two alleles per marker in the table, you need to increase this value.
Keep Allele, Size, Height, Area, Mutation and Comment together	If the box is unchecked, then the columns display as: Allele 1, Allele 2, Size 1, Size 2, Height 1, Height 2, Area 1, Area 2, Mutation 1, Mutation 2, Comment 1, Comment 2 or If the box is checked, then the columns display as: Allele 1, Size 1, Height 1, Area 1, Mutation 1, Comment 1, Allele 2, Size 2, Height 2, Area 2, Mutation 2, Comment 2

Creating a New Table Setting

To create a New Table Setting:

1. Select **GeneMapper Manager > Table Settings** and click **New** to open the Table Setting Editor window.
2. In the **General** tab, enter a name for the new setting and a description if necessary.
3. Open the **Samples** and/or **Genotypes** tabs and select the desired contents for the Samples and Genotypes tables in the corresponding tabs.
4. Click **OK** to save the new setting.

Copying a Table Setting

To copy a Table Setting:

1. Select **Tools > GeneMapper Manager** to open the GeneMapper Manager window.
Note: If the table is selected in the project window drop down menu, then you can click the Table Settings icon or **Tools > Table Setting Editor**.
2. Click the **Table Settings** tab.
3. Select a table setting name, then click **Open**. The selected setting opens.
4. Click **Save As** and give the table setting a new name.
5. Click **OK** to save the new table setting.

Editing a Table Setting

To edit a Table Profile:

1. Select **Tools > GeneMapper Manager** to open the GeneMapper Manager window.
Note: If the table is selected in the project window drop down menu, then you can click the Table Settings icon or **Tools > Table Setting Editor**.
2. Select the **Table Settings** tab.
3. Select a table setting name and click **Open**. The selected setting opens.
4. Perform edits in the table setting.
Note: See [“The Sample and Genotypes Tab Elements” on page 5-32](#).
5. Click **OK** when you have completed editing. The changes you have made are saved to the Table Setting.

Deleting a Table Setting

To delete a Table Setting:

1. Select the **Table Settings** tab in the GeneMapper Manager window.
2. Select a table setting name and click **Delete**.

A warning alert is shown. Click **OK** to remove the setting.

Filtering Controls

The Samples Table Settings column labeled “Filtering,” shown in the figure below, is used to filter or set the type of record or display presented for a Project window column.

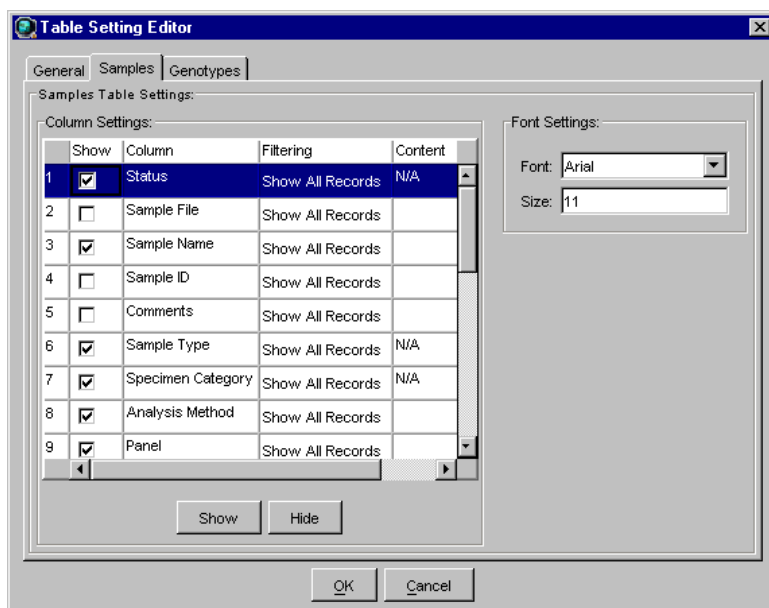


Figure 5-21 Filtering Column of the Table Settings Editor

Samples View Column Filtering Settings

Note: When you apply these settings to filter parameters, be aware that when no instance of the type specified exists, the Sample Table displays blank. Return to the Table Manager and reset the filter.

The following information describes how to use the filter settings for the Samples Column Settings. The default “Show All Records” can be changed to one of the following:

Table 5-10 Samples view filter settings:

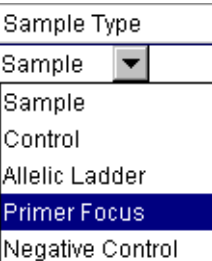
Column Name	Filter Settings
Status Sample File	Analyzed or Not Analyzed 
Sample Name Sample ID Comments Analysis Method Panel Size Standard Matrix Run Name Instrument Type Instrument ID Run Date & Time User Defined Columns	Show Records Containing  Note: This setting allows specification by a string. Enter this information into the Content column.
Sample Type	Sample, Control, Allelic Ladder, Primer Focus, or Negative Control 

Table 5-10 Samples view filter settings: (continued)

Column Name	Filter Settings
Reference Data Off-scale Sizing Quality Invalidated	Yes or No 
Sample File Not Found Matrix Not Found Size Standard Not Found	Show Pass (green squares) or Show Low Quality (red octagons) 
Sizing Quality (SQ)	Show Pass, Show Check, or Show Low Quality  Note: These settings restrict the display to those records meeting these criteria.

Genotypes View Filter Settings

Note: In applying these settings to filter parameters, be aware that when no instance of the type specified exists, the Genotypes table will be displayed blank. Return to the Table Manager and reset the filter.

The following information describes how to use the filter settings for the Genotypes Column Settings. The default “Show All Records” can be changed to one of the following:

Table 5-11 Genotypes view filter settings:

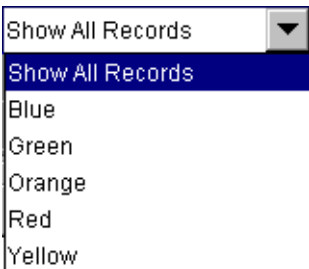
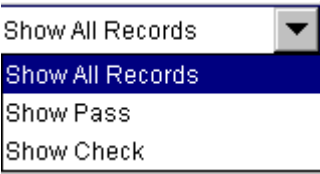
Column Name	Filter Settings
Sample File Sample Name Sample ID Run Name Panel Marker Allele Size Height Peak Area Mutation AE Comment Integration Comments User Defined Columns	Show Records Containing  Note: This setting allows specification by a string. Input this information into the Content column.
Dye	Blue, Green, Orange, Red, or Yellow  Note: This setting can be changed to display either all colors “Show All Records” or the choice of display of an individual color. SNP genotyping data cannot be filtered by this selection.

Table 5-11 Genotypes view filter settings: *(continued)*

Column Name	Filter Settings
Off-scale Sharp Peak (M) One Basepair Allele (M) Single Peak Artifact (M) Split Peak (M) Out of Bin Allele Peak Height Ratio Low Peak Height Spectral Pull-up Allele Number Broad Peak Double Peak (SNP) Narrow Bin (SNP) Control Concordance Overlap (HID)	Show Pass (green squares) or Show Check (yellow triangles) 
Allele Edit Allele Display Overflow	Yes or No 
Genotype Quality (GQ)	Show Pass, Show Check, or Show Low Quality 

Section 5.4 Plot Settings Tab

Overview The Plot Settings tab enables you to create and edit plot setting profiles for viewing your data in the Samples and Genotypes plot views. This section describes how to use the Plot Settings tab in the GeneMapper Manager window of the GeneMapper™ ID Software Version 3.1 and how to use the features of the Plot Settings Editor. To display the Plot Settings Tab, select **Tools > GeneMapper Manager > Plot Settings**.

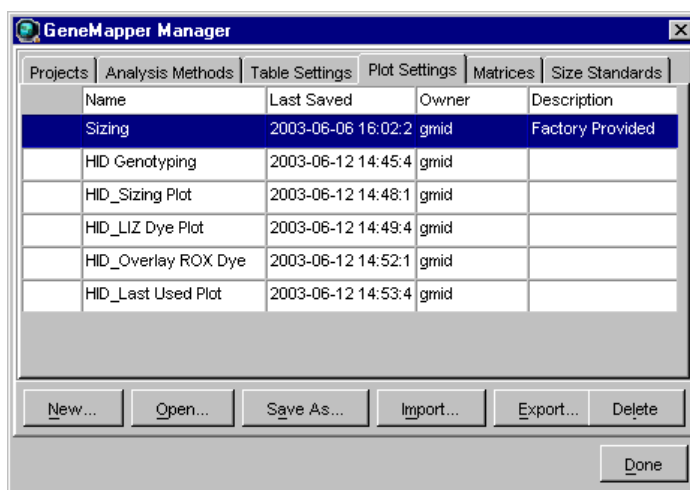


Figure 5-22 Plot Settings Tab of the GeneMapper Manager

Plot Settings Tab Elements

Table 5-12 Description of elements in the Plot Settings Tab

Item	Description
Name column	Plot setting name
Last Saved column	Date/Time stamp showing when the plot setting was last saved
Owner column	User name of the person who created the plot setting
Description column	A description of the plot setting

Table 5-12 Description of elements in the Plot Settings Tab

Item	Description
New button	Opens the Plot Settings editor Always enabled
Open button	Opens the editor for a selected project Enabled when a single plot setting is selected
Save As button	Displays the Save As dialog box Enabled when one or more settings are selected
Import button (.xml file extension)	Displays a dialog box for Importing plot settings Always enabled
Export button (.xml file extension)	Displays a dialog box for Exporting selected plot setting Enabled when one or more plot settings are selected
Delete button	Deletes the selected plot setting(s)
Done button	Closes the GeneMapper Manager

Default Profiles

Applied Biosystems provides five default plot settings for AmpF ℓ STR analysis:

- AmpF ℓ STR Genotyping
- AmpF ℓ STR Sizing
- Last Used Plot
- Overlay LIZ
- Overlay ROX

You need to import these default plots into the database through the Plot Settings tab.

To import a default plot setting:

1. Press **Import**. A dialog box opens.
2. Double-click the **Plot Settings** folder to display all of the default plots.
3. Select the default plot you wish to import, then click **Import**. The default plot appears in the Plot Settings tab.

Plot Settings Editor

Introduction The Plot Settings Editor allows you to customize your plot views by adjusting the following:

- The items displayed in the Sample headers
- The items displayed in the Genotype headers
- The columns displayed in the Sizing table
- How labels are shown
- Display settings

How to Access the Plot Settings Editor

The Plot Settings Editor can be accessed in the following ways:

- Select **Tools > GeneMapper Manager > Plot Settings Tab > New or Open**.
- In the Sample or Genotype Plot views, select a profile to edit and select **Tools > Plot Settings** or click the **Plot Setting Editor** icon on the toolbar.

General Tab

The General tab allows you to enter in the name and description of the plot setting.

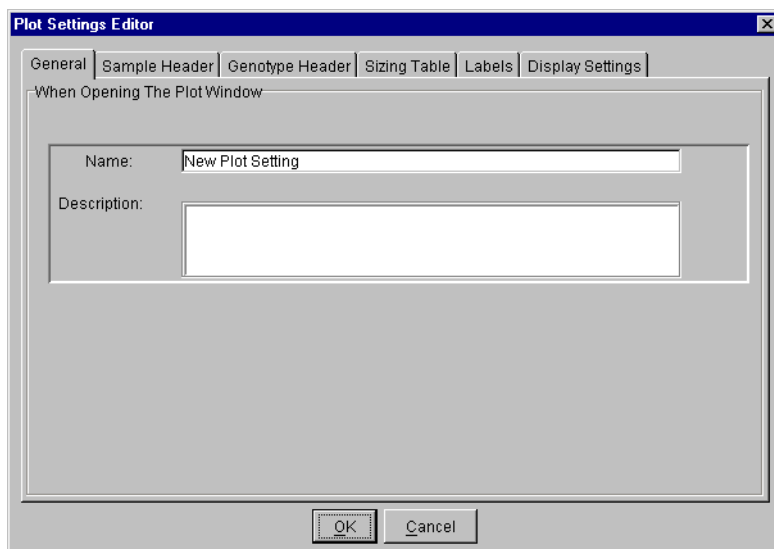


Figure 5-23 General Tab of the Plot Settings Editor

Sample Header Tab

The Sample Header tab controls what is viewed in the header above each sample electropherogram in the Samples Plot view.

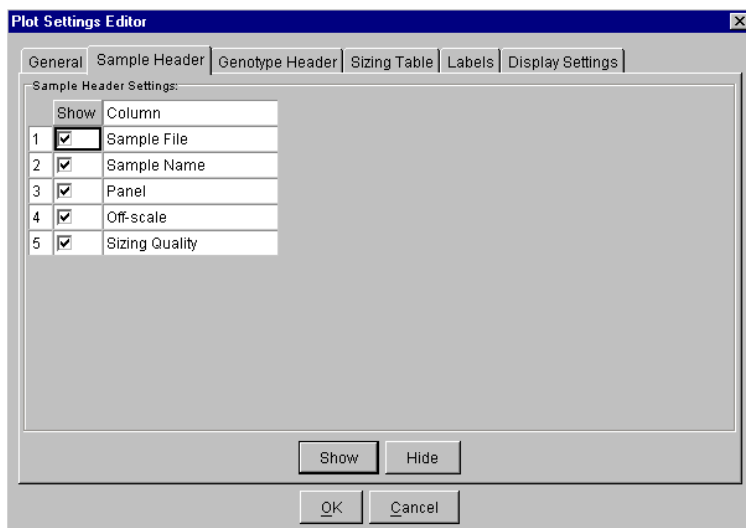


Figure 5-24 Sample Header Tab of the Plot Settings Editor

The Show button displays all selected rows as visible (checked), and the Hide button displays all selected rows as hidden (unchecked).

Genotype Header Tab

The Genotype Header tab controls what is viewed in the header above each Genotype electropherogram in the Genotypes Plot view.

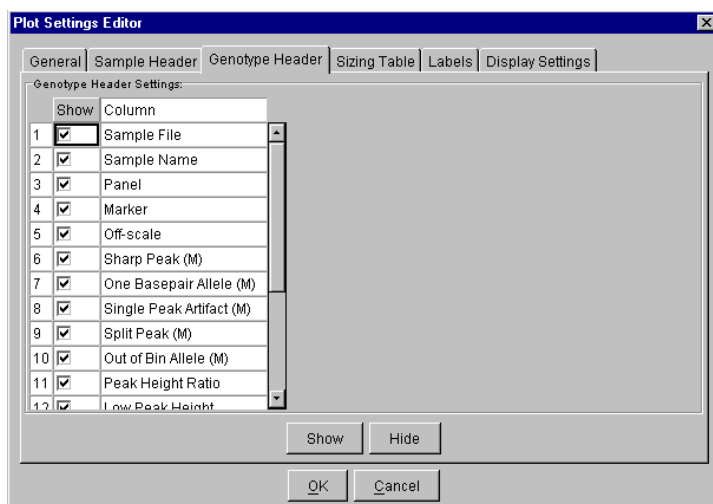


Figure 5-25 Genotype Header Tab of the Plot Settings Editor

The Show button displays all selected rows as visible (checked), and the Hide button displays all selected rows as hidden (unchecked).

Sizing Table Tab The Sizing Table tab controls which columns are displayed in the sizing table as shown in the Samples Plot view.

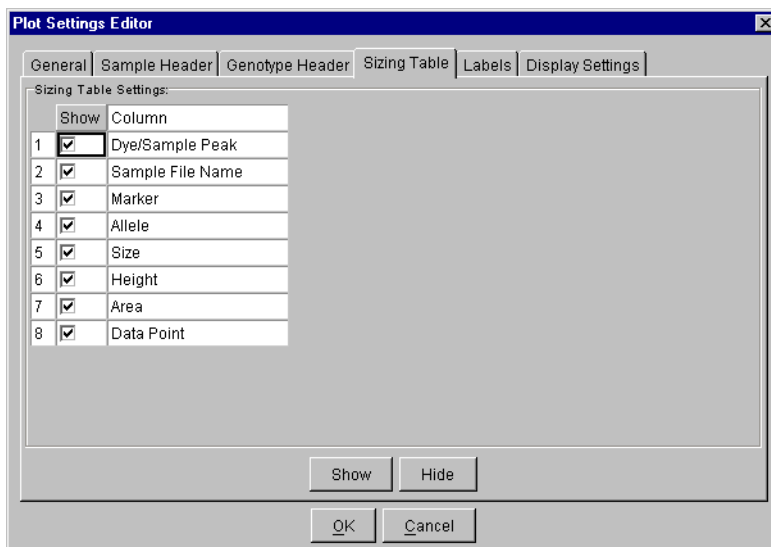


Figure 5-26 Sizing Table Tab of the Plot Settings Editor

The Show button displays all selected rows as visible (checked), and the Hide button displays all selected rows as hidden (unchecked).

Labels Tab The Labels tab controls how labels are viewed for each allele. You can display up to four labels per peak.



Figure 5-27 Labels Tab of the Plot Settings Editor

Use the drop-down menus to display the labels you want to use.

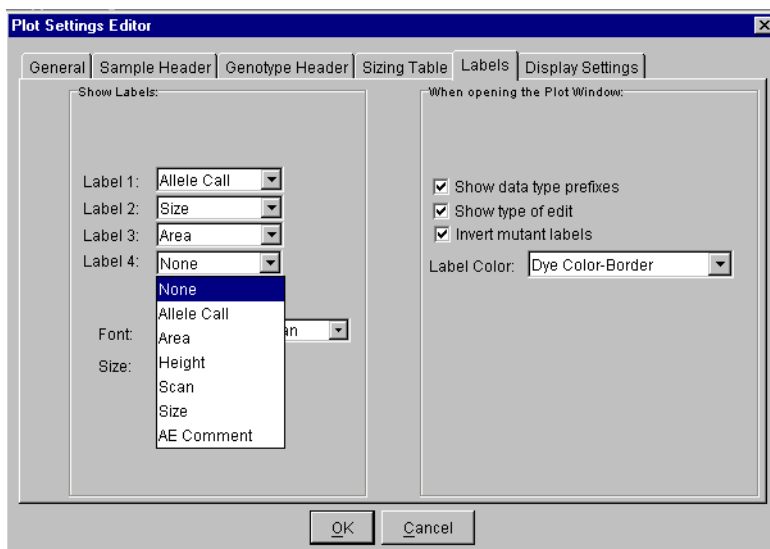


Table 5-13 Labels tab menu items:

Name	Type	Description
Allele Call, Area, Height, Data Point, and Size labels	Drop-down menu items in the label box	Labels determined by the GeneMapper <i>ID</i> software v3.1.
AE Comment	Drop-down menu item in the label box	Shows the last allele edit comment Note: If you delete an allele, the AE Comment is not displayed in the genotype table, is visible in the Allele History. The AE Comment is only displayed for changed alleles.
Show data type prefixes	Check box	Displays label prefixes, such as “a1” for allele call
Show type of edit	Check box	Displays type of edit made to the allele. If an allele is manually edited, the label will display “changed” or “deleted.”
Invert mutant labels	Check box	Identifies mutant bin. If a bin is labeled as a mutant bin in the Panel Manager, then the label color is inverted to distinguish it from normal bins.

Table 5-13 Labels tab menu items: *(continued)*

Name	Type	Description
Label Color	Drop-down menu	Controls the color of the labels. From the drop-down menu select one of the following: Black and White Dye Color - All (text and border in color) Dye Color - Border (text in black, border in color)

Display Settings Tab The Display Settings tab controls how each plot setting will automatically open when a customer chooses that setting.

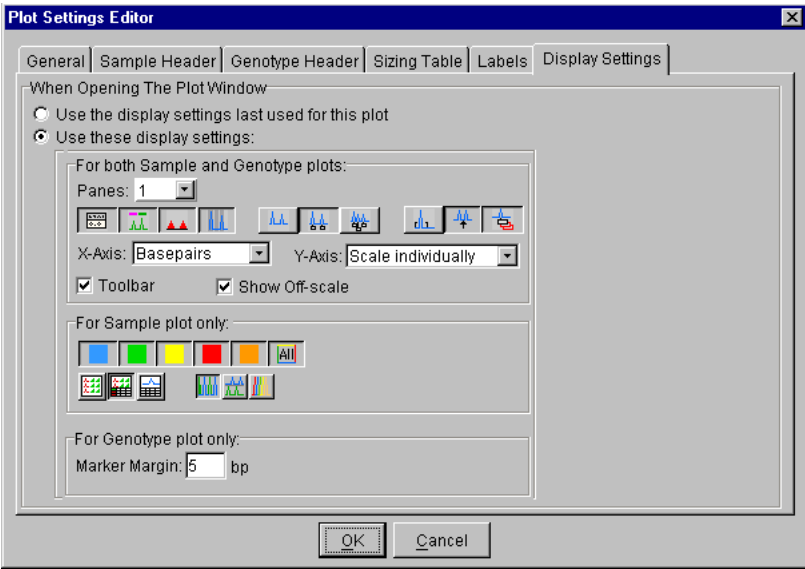


Figure 5-28 Display Settings Tab of the Plot Settings Editor

Icon	Description
	Show/hide plot header
	Show/hide marker range
	Show/hide marker indicators
	Show/hide bins
	Hide labels
	Horizontal labels

Icon	Description
	Vertical labels
	Show/hide peak positions
	Bring/Don't bring controls to top
	Show/hide allele changes
	Show/Hide Individual Dyes or All Dyes
	No table
	Sizing Table
	Genotype table
	Combine Dyes
	Separate Dyes
	Overlay All

You can select **Use the display settings last used for this plot** button to have the Samples and Genotypes plot views display with your previous settings, or select **Use these display settings** to adjust the display with the current settings on this page. These icons are the same as the ones in the Plot window. Move the mouse over each icon to view the tooltip description. The tooltip describes what action occurs if the button is pressed. For example, by default, all colors are shown. If you move your mouse over the All icon , the tooltip says “Hide All Dyes.” If you click the icon, all colors will be hidden.

Section 5.5 Creating and Evaluating a Matrix

Overview This chapter applies only to the ABI PRISM® 310 Genetic Analyzer and the ABI PRISM® 377 DNA Sequencer instruments.

This section describes the process of creating and evaluating a matrix. With the GeneMapper™ *ID* Software Version 3.1, matrices are stored in the GeneMapper *ID* database. They become .mtx files on your hard drive only when you export them. You must export a file in order to run data collection software on the ABI PRISM® 377 DNA Sequencer. You have the option of selecting four or five dyes depending on the application when creating a new matrix for data collection.

Matrices Tab Elements

Table 5-14 Description of elements in the Matrices tab:

Item	Description
Name column	Matrix name
Last Saved column	Date/Time stamp showing when the matrix was last saved
Owner column	User name of the person who created the matrix
# of Dyes column	The number of dyes selected for the matrix
Description	A description of the matrix
New button	Opens the Matrix Editor dialog box Always enabled
Open button	Opens the editor for a selected matrix Enabled when a single matrix is selected
Save As button	Displays the Save As dialog box Enabled when a single matrix is selected

Table 5-14 Description of elements in the Matrices tab: *(continued)*

Item	Description
Import button	Displays a dialog box for Importing matrices Always enabled Note: The extension must be <i>.mtx</i>
Export button	Displays a dialog box for Exporting the selected matrix Enabled when one or more matrices are selected Note: The extension must be <i>.mtx</i>
Delete button	Deletes the selected matrix(s) Always enabled
Done button	Closes the GeneMapper Manager Always enabled

About Matrices

Introduction

There are two dye-labeling chemistries currently available to prepare nucleic acid samples to use the GeneMapper *ID* software on ABI PRISM instruments:

5' end labeling Dye Sets F or G5

Each chemistry has a set of dye labels that fluoresce at different wavelengths when excited by a laser.

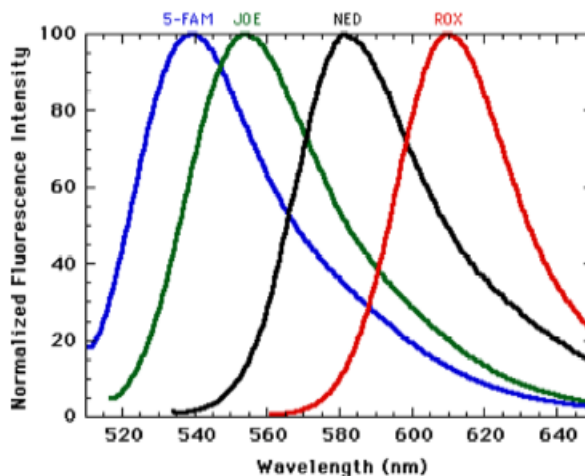
During data collection on the...	The wavelengths are separated...
ABI PRISM® 310 Genetic Analyzer, ABI PRISM® 377 DNA Sequencer, 377XL, or 96-lane upgrade instrument	by a spectrograph into a known spectral pattern across a detection system with the sequencer.

Multicomponent Definition

This process of eliminating spectral overlap is called multicomponenting. Applying a matrix to raw data allows you to generate multicomponented data.

Why Is a Matrix Necessary

A matrix is necessary because the four or five dyes used to label the fragments fluoresce at different wavelengths and may have spectral overlaps:



Assigning a Matrix to a Sample

Normally the matrix is assigned to a sample file automatically upon generation during or after a run. Additionally, a matrix can be manually assigned to a ABI PRISM 377 DNA Sequencer sample file or a ABI PRISM 310 Genetic Analyzer sample file from within the GeneMapper *ID* software.

When to Assign a Matrix

Before you can successfully analyze ABI PRISM 377 DNA Sequencer sample files or ABI PRISM 310 Genetic Analyzer sample files using the GeneMapper *ID* software, you must make a new matrix or assign an existing one to a set of sample files.

Limitations to Matrices

The matrix is instrument-specific. You cannot apply a matrix you made on the ABI PRISM 377 DNA Sequencer to data you collected on an ABI PRISM 310 Genetic Analyzer, nor can you apply a matrix made on one ABI PRISM 377 to a sample or gel file made on another ABI PRISM 377. In other words, you cannot apply matrix created on one instrument to other instruments of the same model.

You can only assign a matrix to sample files generated on the same instrument, under the same electrophoresis, gel matrix and buffer conditions, and using the same dye set.

When to Create a New Matrix

Create a new matrix in the following conditions:

- For each dye set:
F and G5
- When you change the dye set you use to label sample fragments, for example, if you are using the fifth dye.
- When you see multiple unexpected peaks of different colors under an expected peak.
- Anytime a service engineer changes an instrument part, performs preventive maintenance, or realigns the optics.

For information on when to create a new matrix, refer to *Protocols for Processing AmpFLSTR® PCR Amplification Kit Products with the Microsoft® Windows NT® Operating System* (PN 4341742).

Where to Store Exported Matrix Files

Store exported matrix files (.mtx) intended for use by Data Collection software in:

D:\AppliedBio\Shared\Analysis\Sizecaller\Matrix

If Data Collection and Analysis are installed on different computers, the location is the same. Remember to copy the matrix from the analysis computer to the Data Collection computer.

Note: GeneMapper ID software matrices are stored in the Oracle database.

Process of Creating a New Matrix

The following figure shows the procedure for making a new matrix:

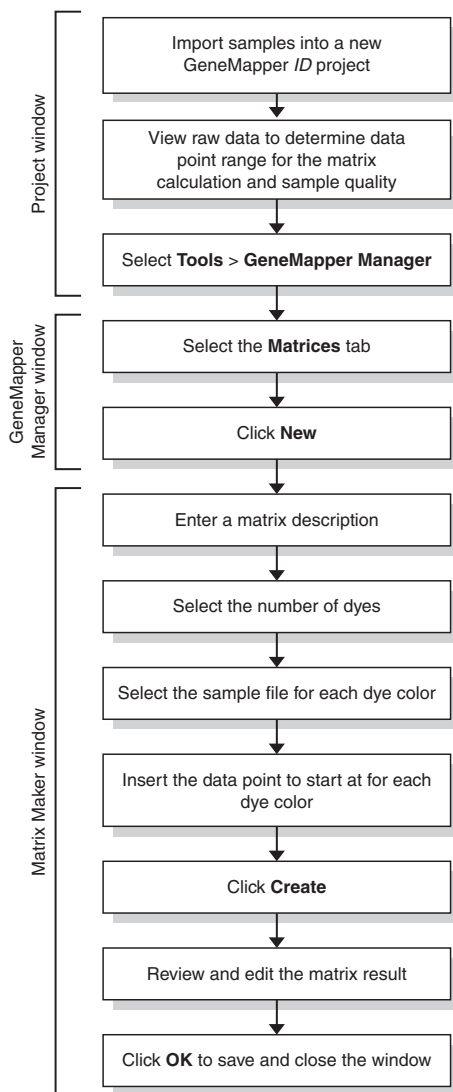


Figure 5-29 Creating a New Matrix

For sample preparation and loading information, refer to the appropriate instrument user guide.

Steps to Create a New Matrix

Table 5-15 Steps to create a new matrix:

Step	Process	See Page
1	Choosing a Data Point Range	5-59
2	Generating a New Matrix	5-60
3	Assigning the Matrix to Samples	5-63
5	Using the Matrix with Data Collection	5-63
6	Causes for Bad Matrices	5-64

Choosing a Data Point Range for the Matrix Calculation

Introduction Depending on how well your Matrix Standards run, it may be necessary for you to choose a specific range of data points to be considered for your matrix calculation.

In order to choose appropriate values for the data point range, you must first view the sample file raw data from each of the matrix standard files, so you can decide where to choose the start point for the data point range.

Viewing the Raw Data

To view raw data:

1. Open a new GeneMapper *ID* Project.
2. Import Samples by selecting **File > Add Samples to Project**.
3. Select the files you want to view:
 - a. Navigate to the disk/directory containing the samples for creating a matrix.
 - b. Select the files you wish to use.
 - c. Click **Add to List**.
4. Click **Add** to import the files into a new project and close the **Add Samples to Project** window.

The Project window reopens with the imported files displayed in the **Samples** tab.
5. Select a sample file.
6. Select **Raw Data** from the File menu.

What to Look For in the Raw Data Display

In the raw data display of the sample files verify the following:

- Data peaks are present in all four or five of the matrix standards.
- There are no anomalies.
- The baseline is stable (flat between peaks).
- Peaks are on-scale— peaks do not exceed 8191 relative fluorescent units— and the peaks of the dye of interest have a value of at least 200.

If peak data does not show these characteristics, refer to [“Causes for Bad Matrices”](#) on [page 5-64](#), for possible interpretations of your peak data.

Choosing a Data Point Range

To choose a data point range:

1. Move the cursor well away from the primer peak, in a region at the beginning of the run and in a flat part of the baseline, and record the data point values for both the start and stop points in the flat part of the baseline of the data point range.

Note: When choosing the start point, do not include primer peaks in the data point range (refer to [“Eliminating Primer Peaks”](#) on [page 5-59](#)). Enter these values in the next step when generating the new matrix (refer to [page 5-60](#)).

2. Return to the Project window by selecting the run folder in the navigation pane.

Eliminating Primer Peaks

Both the primer peaks and the data peaks are displayed when you view the raw data of your matrix standards. Any time you run dye-labeled samples on a (gel) ABI PRISM® 310 Genetic Analyzer or (capillary) ABI PRISM® 377 DNA Sequencer, you have excess dye-labeled primer in the reaction. The primer peak displays as the first peak, usually off-scale because it is in molar excess. Eliminate the primer peak when making a matrix, by choosing the start point after the primer peak in a flat area with a stable baseline.

Note: To create a good matrix, you need at least five fragments in each color.

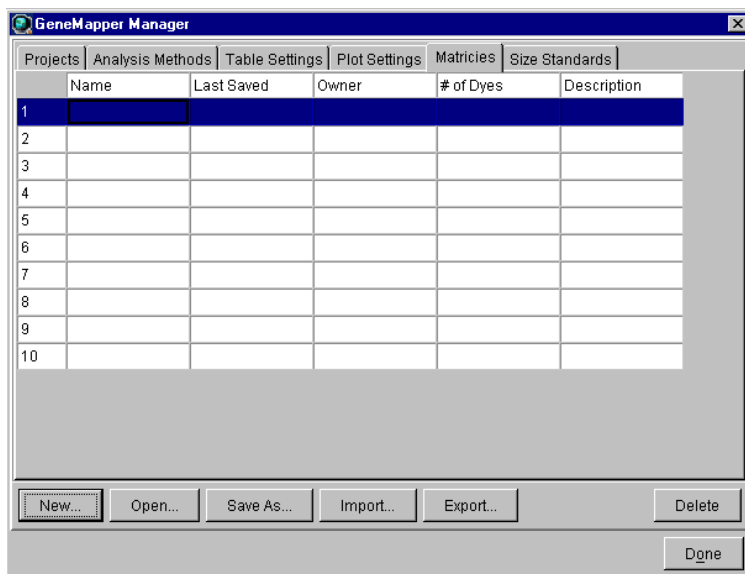
Generating a New Matrix

Creating a GeneMapper *ID* Software Matrix

To create a GeneMapper *ID* software matrix:

1. Select **GeneMapper Manager > Tools**. Then click the **Matrices** tab.

The Matrices page opens.



- Click the **New** button on the Matrices tab. The Matrix Editor dialog box opens.

Matrix Editor

Matrix Description

Matrix Name:

Description:

Matrix Settings

Select the Matrix Standard Sample File:

Number of Dyes: **4**

B No File Selected for "B" Data Start At: 1000

G No File Selected for "G" Data Start At: 1000

Y No File Selected for "Y" Data Start At: 1000

R No File Selected for "R" Data Start At: 1000

Points: 100000

Create

Matrix Result

	B	G	Y	R
B	0.000	0.000	0.000	0.000
G	0.000	0.000	0.000	0.000
Y	0.000	0.000	0.000	0.000
R	0.000	0.000	0.000	0.000

OK Cancel

- Enter in a Matrix name and description, if desired.
- Choose the number of dyes from the **Number of Dyes** drop-down list.

If 5 dyes are selected, a button for "O" data is added to the bottom of the list.

Matrix Settings

Select the Matrix Standard Sample File:

Number of Dyes: **5**

B No File Selected for "B" Data Start At: 1000

G No File Selected for "G" Data Start At: 1000

Y No File Selected for "Y" Data Start At: 1000

R No File Selected for "R" Data Start At: 1000

O No File Selected for "O" Data Start At: 1000

Points: 100000

Create

5. The B, G, Y, R, and O buttons represent dye colors. Choose the file associated with the appropriate dye color.
 - a. Click a button to display a pop-up menu.
 - b. Use the pop-up menu to access a sample file to link to each of the dye-labeled primers.
 - c. Choose the sample file that represents the dye color for that button.

6. Enter the start point that you determined when choosing a data point range in the Start At field.

Refer to [“Choosing a Data Point Range”](#) on page 5-59.

7. Enter the total number of data points to include in order to calculate the matrix in the Points field.

In most cases, leave the default value, unless you must exclude a portion of your data.

Note: You must have at least five peaks to make a matrix.

8. Click **Create** to generate a new matrix.
9. Click **OK** to save and close the Matrix Editor dialog box.

Matrix Example

The following is an example of the matrix results showing the values used to calculate the overlap correction.

Matrix Result					
	B	G	Y	R	O
B	1.0000	0.2009	0.1176	0.0319	0.0385
G	0.9422	1.0000	0.8531	0.4977	0.2718
Y	0.5638	0.6374	1.0000	0.9224	0.6925
R	0.2897	0.3527	0.7938	1.0000	1.175
O	0.0404	0.0962	0.1992	0.304	1.0000

For each dye, the value where the dye fluorescence is read by the appropriate filter is 1.000. The adjacent colors show the amount of overlap for which the system must compensate. The adjacent values, in most cases, should be less than 1.000, but greater than or equal to 0.0000.

Assigning the Matrix to Samples

Introduction After generating the new matrix, assign it to all the samples that you want to analyze.

IMPORTANT! After assigning your matrix to samples, refer to [Appendix C, “Project Window Software Interface,”](#) for information on evaluating the matrix.

- Procedure** To assign a matrix to samples:
1. Open or create a GeneMapper *ID* Project containing the samples you wish to analyze.
 2. Select the Matrix column in the project window.
 3. Select the appropriate matrix from the drop-down list for each sample, or use the **Edit > Fill Down** feature.
 4. Apply additional project settings prior to analyzing the sample files.

Using the Matrix with Data Collection

Using Different Computers for Data Collection and Analysis Be sure to copy matrices generated and exported on the analysis computer (the computer running GeneMapper ID software) to the Data Collection computer, if different. Copying the matrices will ensure that the correct matrix name is stored in the sample file. The proper matrix is required for accurate analysis of ABI PRISM® 310 Genetic Analyzer and the ABI PRISM® 377 DNA Sequencer sample files.

To transfer the matrix from one computer to another:

1. Click the **Matrices** tab in the GeneMapper Manager window.
2. Select **Matrix to Export**.
3. Click **Export**.
4. Select a name and location for the matrix.
5. Transfer the *.mtx* file to the data collection computer.

**Where to Store
Exported Matrix
Files**

Store exported matrix files (.mtx) intended for use by Data Collection software in:

D:\AppliedBio\Shared\Analysis\Sizecaller\Matrix\

If Data Collection and Analysis are installed on different computers, the location is the same. Remember to copy the matrix from the analysis computer to the Data Collection computer.

Note: GeneMapper ID software matrices are stored in the Oracle database.

Causes for Bad Matrices

**If an Error
Message
Displays**

There are two possible causes for the error messages shown in the following table:

If...	Take this action...
you designated the wrong files,	reassign the matrices. Refer to “Assigning the Matrix to Samples” on page 5-63 .
the signal is too weak to make a matrix,	rerun the matrix standards.

Section 5.6 Size Standards Tab

Overview The Size Standards tab in the GeneMapper Manager window contains a table of size standards stored in the database. To display the Size Standards tab, select **Tools > GeneMapper Manager > Size Standards**.

Figure 5-30 displays the four default size standards that are loaded with the GeneMapper *ID* software on an analysis computer only, and Figure 5-31 displays the standards load when GeneMapper *ID* software is loaded on a computer running 3100/3100-*Avant* Data Collection software v2.0.

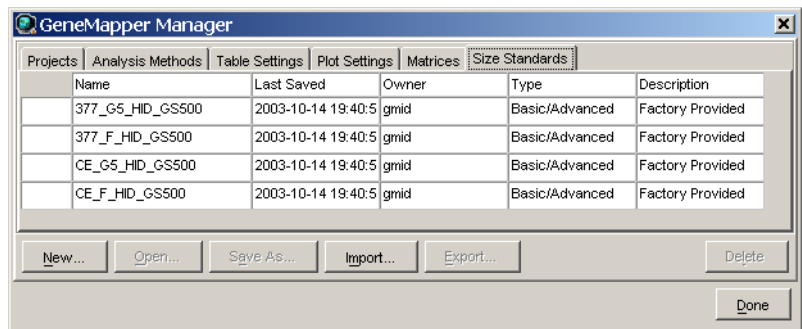


Figure 5-30 Standards for a stand alone analysis computer

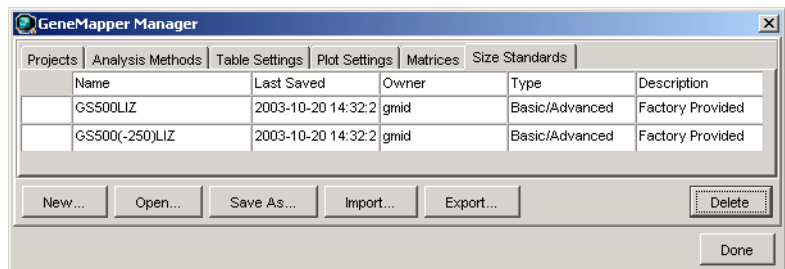


Figure 5-31 Standards for 3100/ 3100-*Avant* system computer using Data Collection software v2.0

**Size Standards
Tab Elements****Table 5-16 Description of elements in the Size Standards tab:**

Item	Description
Name column	Size standard name
Last Saved column	Date/Time stamp showing when the size standard was last saved
Owner column	User name of the person who created the size standard
Type	Classic or Basic/Advanced
Description	A description of the size standard
New button	Opens the Specify parameters and Size Standard Editor dialog boxes to create a new size standard
Open button	Opens the editor for a selected size standard Enabled when a single size standard is selected
Save As button	Displays the Save As dialog box Enabled when a single size standard is selected
Import button	Displays a dialog box for Importing size standards Always enabled
Export button	Displays a dialog box for Exporting selected size standard(s) Enabled when one or more size standards are selected
Delete button	Deletes the selected size standard(s)
Done button	Closes the GeneMapper Manager

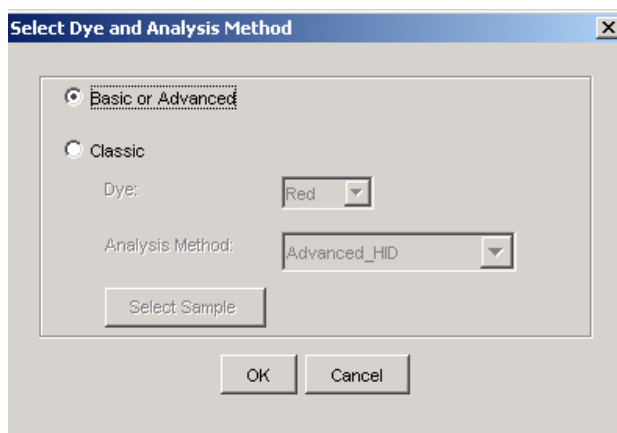
Creating/Editing a Size Standard

Overview To create a size standard, you must first specify the parameters under which the size standard is created and then define the peaks using the Size Standard Editor. When editing a predefined size standard, only the Size Standard Editor dialog box will be shown.

Specifying Size Standard Parameters

To specify size standard parameters:

1. Select **Tools > GeneMapper Manager**. Then click the **Size Standards** tab.
2. Click the **New** button to open the Select Dye and Analysis Method dialog box.



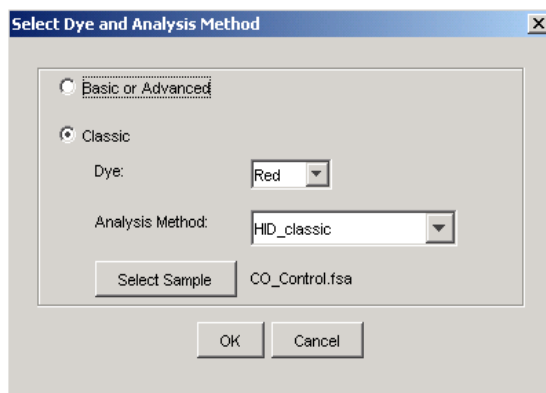
3. Perform the applicable procedure below to use the Size Standard Editor.

Note: The mode you select must be the same as the peak detection algorithm you use to analyze the samples in Analysis Method.

Using the Size Standard Editor in Classic Mode

To use the size standard editor in classic mode:

1. Complete the Select Dye and Analysis Method window:



- a. Select **Classic**.
- b. Select a dye color from the drop-down list.
- c. Select the Classic Analysis Method from the drop-down list.
- d. Select the row for the particular sample file that contains the dye standard you want to use as the template.
- e. Click **OK**.

The Size Standard Editor opens.

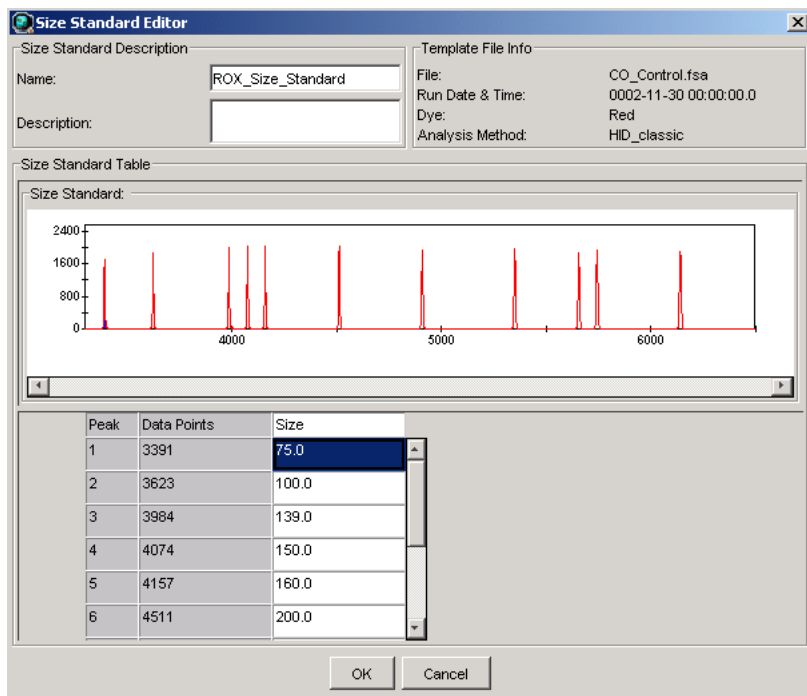


Figure 5-32 The Size Standard Editor in Classic Mode

The Size Standard Editor in Classic mode shows an electropherogram and a table of peaks for the dye color and sample selected. The software assigns a number to each peak found in the electropherogram in order, from left to right. You should be able to recognize the peak pattern of the standard in the electropherogram.

Note: You can only change the peak size value in the right column of the table. You cannot change or rearrange the peak numbers.

Note: If too many peaks display in the electropherogram or the baseline is too high, you might need to adjust the analysis parameters.

2. Enter in a name and description for the size standard.

3. Specify the peaks of the standards and their sizes.

Click the peak you want to define either in the electropherogram or in the table, or click on a row and the corresponding peak is highlighted.

Note: For easier viewing, you can zoom in on the electropherogram by click+drag on the X-axis numbers.

4. Type the peak size in the corresponding column.

Note: When you want to ignore a peak for the standard definition, leave a zero in the Size column.

5. Press the tab key to move to the next size standard peak, then enter its value.

Note: Ensure you tab to the next size standard peak and not to a spike or to baseline noise. See for all GeneScan size standard profiles and definitions.

Note: To skip a fragment, leave the value as zero in the corresponding cell in the table.

6. Click **OK** to save and close the Size Standard Editor, or click **Cancel** to close without saving.

Using the Size Standard Editor in Basic or Advanced Mode

The Size Standard Editor window in Basic or Advanced mode allows you to define the peaks for each size standard dye you use.

Size Standard Editor

Edit

Size Standard Description

Name:

Description:

Size Standard Dye:

Size Standard Table

	Size in Basepairs
1	0.0
2	0.0
3	0.0
4	0.0
5	0.0
6	0.0
7	0.0
8	0.0
9	0.0

OK Cancel

To use the size standard editor in Basic or Advanced mode:

1. Enter a name and description for the size standard you are creating.
2. Choose a Size Standard Dye color from the drop-down list.
3. Enter the base pair values to be defined in your size standard in the table provided.

Note: Use the Edit menu to undo certain actions or Add and Remove Rows as desired.

Edit

Add New Row Ctrl+A

Remove Row Ctrl+R

4. Click **OK** to save and close the Size Standard Editor, or click **Cancel** to close without saving.

Options Dialog Box

This chapter covers:

Introduction	6-2
HID Analysis Options	6-3
Startup Tab	6-4
Add Samples Tab	6-5
Analysis Tab	6-7
Users Tab	6-9

Introduction

The Options dialog box contains several tabs for setting preferences associated with various software features. You can use the tabs in the Option dialog box to:

- Set project preferences
- Set default analysis parameter preferences
- Set default post analysis display preferences
- Create New Users and Passwords

Note: Each registered user has his or her own set of preferences. When you set these options, it affects only the user currently logged in.

HID Analysis Options

Overview In preparing GeneMapper™ *ID* software to analyze data from AmpFℒSTR kits, view and set options for:

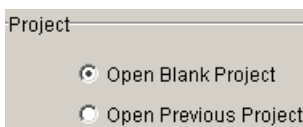
- Startup of GeneMapper *ID* software
- Analysis settings when adding samples to a project
- Analysis displays
- Users of GeneMapper *ID* software

Viewing and Setting Options

Note: These options are active only for the user currently logged into the software.

To view and set options:

1. Select **Tools > Options** to open the Options dialog box.
2. From the Startup tab, view the default startup options:



Note: Later, you may select **Open Previous Project** to open the last project you analyzed using GeneMapper *ID* software.

Startup Tab

The Startup tab contains preferences associated with the startup of the software.



Element	Description
Open Blank Project option button	Sets the software so that upon launch, a blank Project window opens.
Open Previous Project option button	Sets the software so that upon launch, the last Project viewed will open.

Add Samples Tab

The Add Samples tab contains preferences associated with the adding of samples.

Options

Startup | **Add Samples** | Analysis | Users

When adding new samples, automatically...

Set Analysis Method to:

☐ HID_Advanced for all samples.

☒ Read from the Sample.

Set Size Standard to:

☐ CE_F_HID_GS500 for all samples.

☒ Read from the Sample.

Set 310/377 Matrix to:

☐ for all samples.

☒ Read from the Sample.

Set Panel to:

☐ Select a Panel for all samples.

☒ Read from Data collection 'Comment/Panel field'.

Set Sample Type to:

☐ Sample for all samples.

☒ Read from Data collection 'Info' field.

OK Cancel

Element	Description
Set Analysis Method to drop-down menu and Read from the Sample option button	When you add samples, the Analysis Method property is set to the value in the text box, which contains the list of analysis methods from the GeneMapper <i>ID</i> database.
Set Size Standard to drop-down menu and Read from the Sample option button	When you add samples, set the size standard property to the value in the text box, which contains the list of size standards from the GeneMapper <i>ID</i> database.

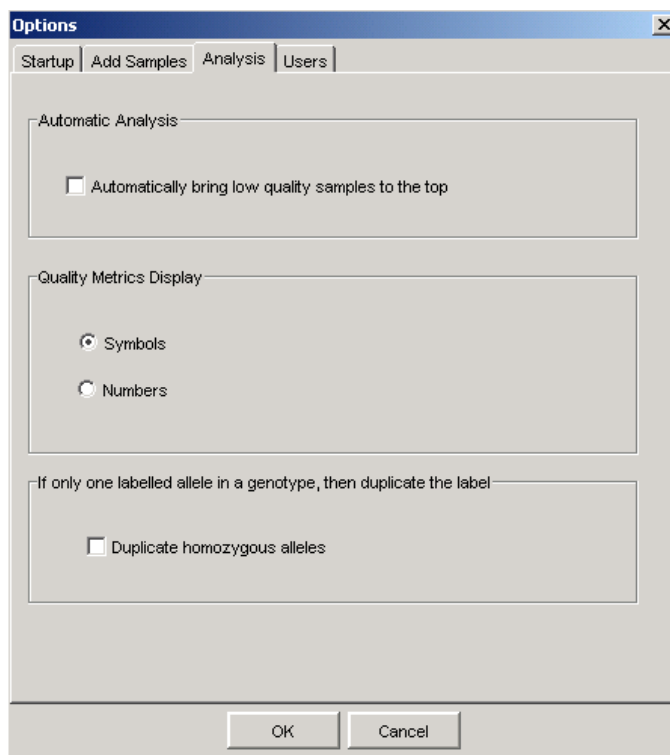
Element	Description
Set 310/377 Matrix to drop down menu Read from the Sample option button	Set the matrix file to the dye you are using or read from the sample file.
Set Panel to • Select a Panel for all samples option button and window • Read from the Data collection 'Comment/Panel field' option button	When you add samples, the panel for all samples is set to the value in the text box. When the option button is selected, the following choices may be made: • Leave the text alone, which selects the existing name, or • Click the field to present the Select a Panel window (to select an existing panel) The Select a Panel window contains the list of panels from the GeneMapper <i>ID</i> database. When you add samples, the panel for all samples is set to the value in the Sample Sheet comment column. Note: For the 3100 instrument, this column is titled "Color Comment." For the 310 and 377 instruments, this column is titled "Comment."
Set Sample Type to • Sample type for all samples option button and drop-down menu • Read from Data collection 'Info' field option button	When you add samples, the Sample Type for all samples is set to the value chosen from the Sample Type drop-down menu, which contains choices for Sample, Control, Allelic Ladder, and Primer Focus. When you add samples, the GeneMapper <i>ID</i> software does a "best guess" match, comparing the contents of the field specified in the Sample type box with the list of sample types (Sample, Control, Allelic Ladder, Negative Control, and Primer Focus). • If a match is found, then the Sample Type property for the sample is set to that sample type. • If a match is not found, then the Sample Type property for the sample is set to "Sample."

Note: The Panel and Sample Type properties accounts for close spellings and abbreviations as shown in the following table.

Case	Result
A sample file field contains either Control, Ctrl, Crl, Cntrl, or a similar variation.	The sample type for the sample is set to Control.

Analysis Tab

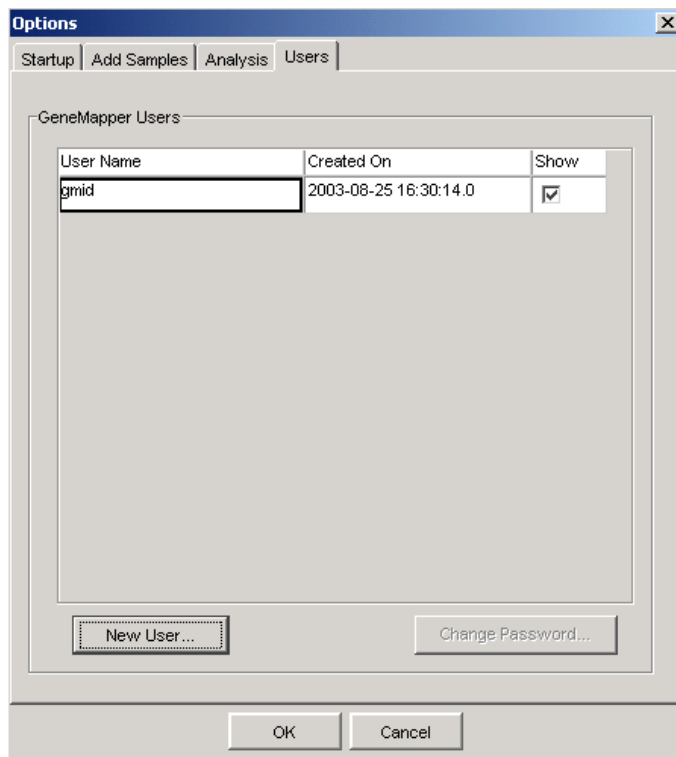
The Analysis tab contains preferences associated with data analysis.



Element	Description
Automatic Analysis group box: Automatically brings errors to the top of the table check box	When samples are analyzed, the software sorts the table automatically to bring samples with errors to the top of the table. Same functionality as the Bring Errors To Top icon in the Project window.
Quality Metrics Display group box: Symbols button Numbers button	Display quality values in project tables as symbols (green square, yellow triangle, red octagon), or numerical values (0.0, ..., 1.0). Note: If Symbols (default) is chosen, then all quality values are shown as symbols. If Numbers is chosen, then only SQ and GQ columns are shown as numbers; the other result columns are symbols.
If only one allele is labeled in a genotype, then duplicate the label group box: Duplicate homozygous alleles checkbox	If this option is checked, single alleles will be displayed in the Genotypes table and in the Plot window as two labels to indicate presumed homozygotes.

Users Tab

The Users tab contains preferences associated with managing registered users.



Note: Once created, user names cannot be deleted.

Element	Description
User Name field	Displays the list of registered users. • User Name column = Your name • Created On column = Date user was created • Show column = Shows your name in User Name drop-down menu of the login window, if checked. Clearing a Show column check box for a user blocks login for that user (can be done by anyone running GeneMapper <i>ID</i> software). Shift-click the column label headers to sort the list by that column.
New User button	Opens the New User dialog box.  To add a user name for GeneMapper <i>ID</i> software, click New User to open a dialog box and then: - Type a user name into the dialog box. - Enter a password using only alphabetic characters. - Confirm the password. - Click OK to assign the user name and close the dialog. When you select this user name on launching GeneMapper <i>ID</i> software, the name is used to identify the preferences set by you. Note: User names must be unique. If the new user name is not unique, an alert message will be displayed. Note: Users added here are for this GeneMapper <i>ID</i> software installation. Users are visible to all other users. The names are converted to lower case and passwords are not case sensitive.

Element	Description
Change Password button	Opens the Change Password dialog box, and allows you to change the password for the selected user. 

Analyzing and Interpreting Data

7

This chapter covers:

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Concordance Checks	7-32

Introduction

The previous chapters described how to customize the software to meet your analysis needs. Once the software is configured to analyze a particular set of samples, such as 3100 four dye AmpF ℓ STR $^{\text{®}}$ sample files, you can use the information in this chapter to analyze your data, view the data, and interpret PQVs.

Considerations for HID Analysis

When using GeneMapper $^{\text{TM}}$ *ID* Software version 3.1 to perform Human Identification (HID) analysis with AmpF ℓ STR $^{\text{®}}$ kits, consider the following:

- HID analysis requires the presence of at least one allelic ladder sample per project. Multiple ladder samples can be used in an analysis, provided individual laboratories conduct the appropriate validation studies.
- For multiple ladder samples, suitable processing of allelic bins is carried out whereby an average of all ladders within a run folder using the same panel is used to calculate allelic bin offsets.
- Allelic ladder samples in a single run folder are considered to be from a single run.
- When multiple run folders are imported into a project, only ladders within a single run folder are used for calculating allelic bin offsets and subsequent genotyping.
- Allelic ladder samples need to be identified as “Allelic Ladder” in the Sample Type column in a project. Failure to make this setting for ladder samples results in failed analysis.
- Allelic bin definitions are stored in the AmpF ℓ STR panels in the Panel Manager.
- Lanes or injections containing the allelic ladder should be analyzed with the same analysis method and parameters used for samples.

- Alleles not found in the AmpF $\mathbb{L}$ STR Allelic Ladders do exist. These off-ladder alleles may contain full and/or partial repeat units. An off-ladder allele is defined as an allele falling outside of the ± 0.5 -bp bin window of any known allelic ladder allele or virtual bin.

Note: If a sample allele peak is called as an off-ladder allele, then the sample needs to be run again to verify the result.

- The marker-specific stutter ratios included in the kit panels and cutoff value used in the tutorial are provided to serve as a tool and a guideline. Final conclusions should be based on careful examination of the STR profiles.

Analyzing Samples Files

To analyze sample files, you can either add .fsa files generated on an Applied Biosystems Genetic Analyzer or you can open an existing project, apply new analysis settings, and re-analyze.

IMPORTANT! In order to be able to re-analyze an existing project, certain criteria must be met. Please see Appendix F for specific information regarding re-analyzing existing projects.

Sample File Format

GeneMapper™ *ID* Software v3.1 supports data (sample files) generated from the following instruments:

- ABI PRISM® 310 Genetic Analyzer
- ABI PRISM® 377 DNA Sequencer
- ABI PRISM® 3100-*Avant* Genetic Analyzer
- ABI PRISM® 3100 Genetic Analyzer

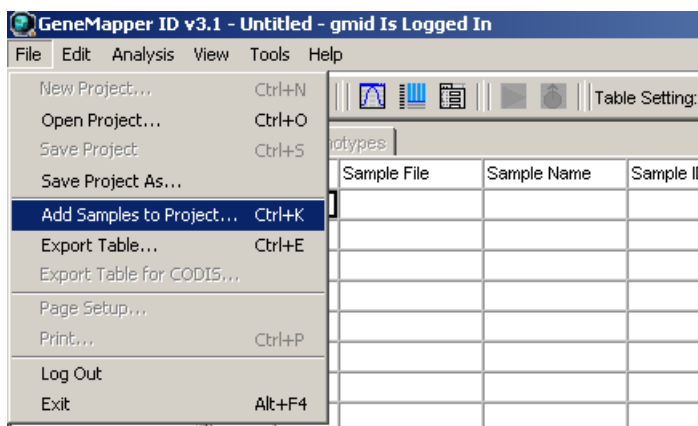
The .fsa file extension is *required* for GeneMapper *ID* software to recognize the file. Sample files created from an instruments using a computer running Microsoft® Windows® NT or 2000 operating system automatically create file with this extension.

Note: Data generated from a 310 Genetic Analyzer or 377 DNA Sequencer using a Macintosh® platform must be converted to a Microsoft® Windows®-based format before it can be used with GeneMapper *ID* software. Refer to [Appendix E, “Converting Macintosh® Sample Files.”](#)

Adding Sample Files to a New Project

To add sample files to a new project:

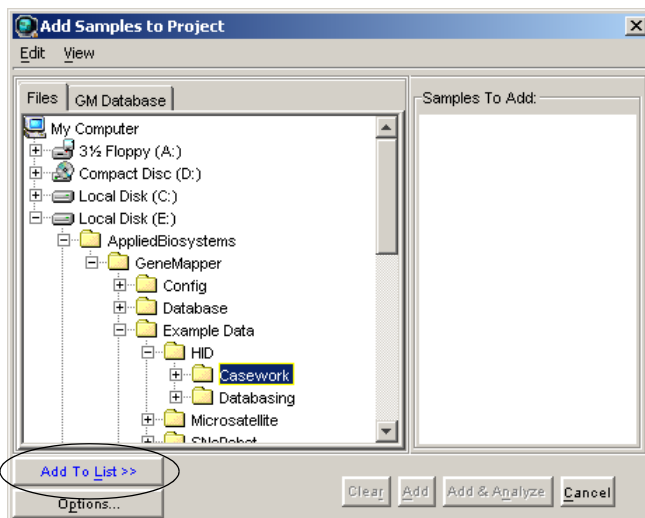
1. Select **File > New Project** to open a new project.
2. Select **File > Add Samples to Project**.



3. In the Add Samples to Project dialog box:
 - a. Locate your sample files on your hard drives or network servers by expanding folders in the navigation pane.
 - b. Use the table below to add folder(s) or samples.

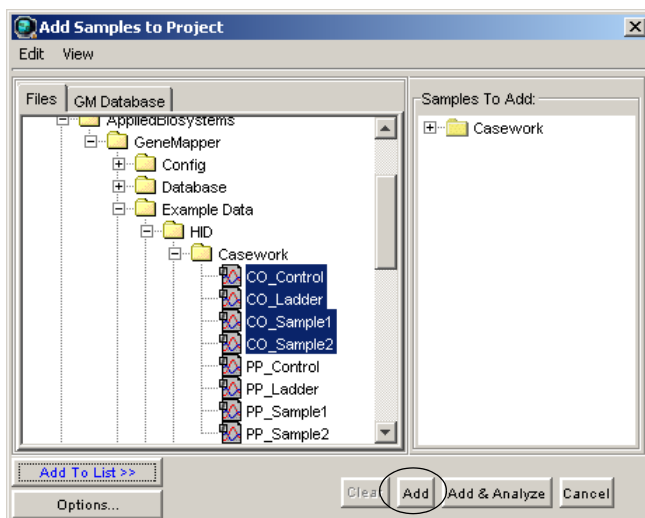
To add ...	Do this ...
A folder containing sample files	Select the folder.
Multiple folders containing sample files. ^a	Use the Shift key to select continuous folders or the Ctrl key to select discontinuous folders.
Some of the files in a folder	Use the Shift key to select continuous samples or the Ctrl key to select discontinuous samples.

a. The data contained in each run folder is analyzed by comparison of the allelic ladder within that particular folder. It is *required* to have at least one allelic ladder per folder.



- c. Click **Add to List**. The folder will then appear in the right pane.

Note: If you select a couple of sample files, the folder from which the samples were selected appears in the right pane.



- d. Click **Add**, the window closes, and the samples files display in the new project window.

Applying Analysis Settings

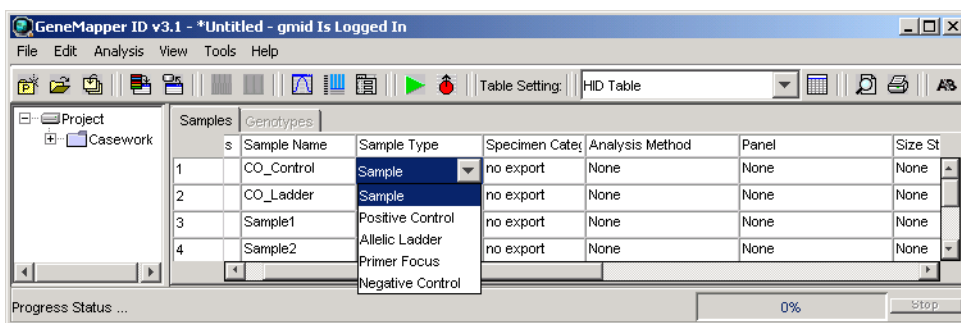
Before you can analyze your sample files, define the analysis settings in the software. The components you must select within the main project window are as follows:

- Sample Type
- Analysis Method
- Panel
- Size Standard
- Matrix (310 and 377 sample files only)

Note: Blank projects can be set up with default analysis settings. These defaults can be selected in the Options window (under the Tools Menu). Refer to [Chapter 6, “Options Dialog Box,”](#) for more information. In addition, these analysis settings can be read from the sample file depending on what information was entered into the sample sheet. (Refer to [Appendix G, “Sample Sheets and Plate Records.”](#))

To select the appropriate analysis settings:

1. In the Sample Type column, use the drop-down list to select one of the following:
 - Sample (DNA evidence or database samples)
 - Positive Control (*e.g.*, Control DNA 9947A or Control DNA 007)
 - Allelic Ladder
 - Negative Control
 - Primer Focus (for SNaPshot kit only)

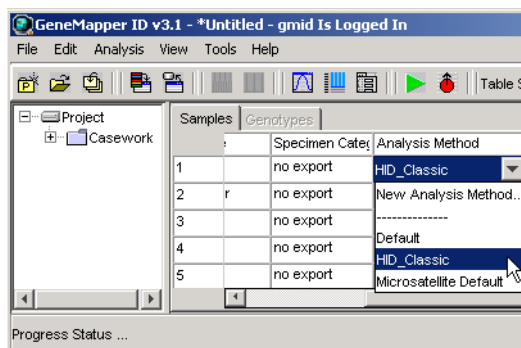


Note: It is recommended that you choose the appropriate sample type. To make it easier, use the data collection sample sheet to make these entries (refer to Appendix G). There is a PQV within the software known as Control Concordance (CC). If you choose the appropriate sample type for your negative and positive controls, the software will automatically check to make sure your negative control does not give any labeled peaks above the peak amplitude threshold and your positive control genotype profile matches the expected profile for 9947A Control DNA. If the software finds that your controls match, you will get a green flag for the CC. You will get a yellow flag for CC if the controls do not match. This is a quick and easy way to check or double-check your controls.

Note: If you use a positive control other than 9947A, you can change the control alleles in the marker table within the panel manager to automatically scan for concordance to your control's profile. (Refer to [Chapter 4, "Using Panel Manager."](#)) However, you can only have one control profile entered at a time per panel.

Note: If a ladder injection is of low quality, you can change the sample type from Allelic Ladder to Sample. This will remove the low quality injection from consideration in calculating the allelic bins.

2. In the Analysis Method column, select a method:

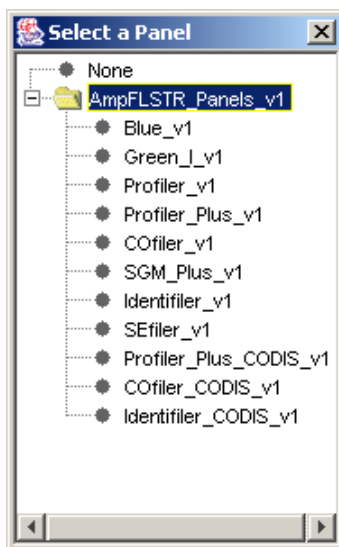


- a. Use the drop-down list to select the appropriate method.

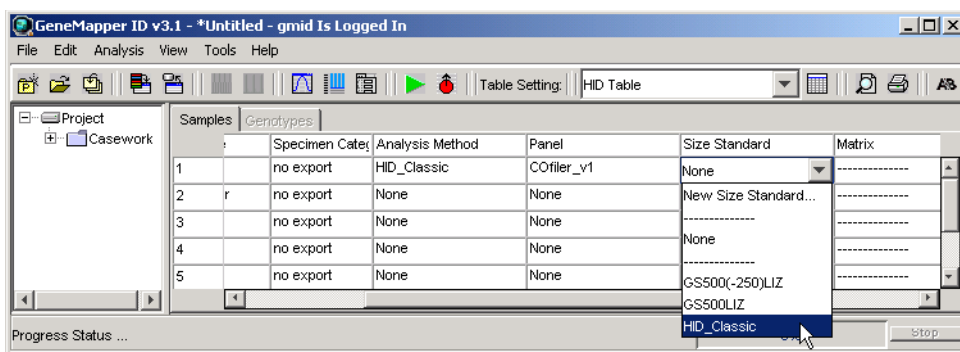
Note: You have the option to define a new analysis method from this drop-down list.

- b. Click the column header cell to highlight the entire column or highlight a portion of the column that you want to apply the same setting to, then select **Edit > Fill Down**.

3. In the Panel column, select a panel:
 - a. Highlight a cell within a sample row by clicking it in the panel column. A Select a Panel window will appear.
 - b. Expand the AmpFLSTR_Panels_v1 folder.



- c. Double-click the appropriate panel to populate the cell within the panel column.
 - d. Click the column header cell to highlight the entire column or highlight a portion of the column that you want to apply the same setting to, then select **Edit > Fill Down**.
4. In the Size Standard column, define the size standard:



- a. Use the drop-down list to select the appropriate size standard.

Note: You have the option to define a new size standard from this drop-down list.

- b. Click the column header cell to highlight the entire column or highlight a portion of the column that you want to apply the same setting to, then select **Edit > Fill Down**.

5. In the Matrix column, select a matrix:

- a. Use the drop-down list to select the appropriate matrix for your 310 or 377 instrument.
- b. Click the column header cell to highlight the entire column or highlight a portion of the column that you want to apply the same setting to, then select **Edit > Fill Down**.

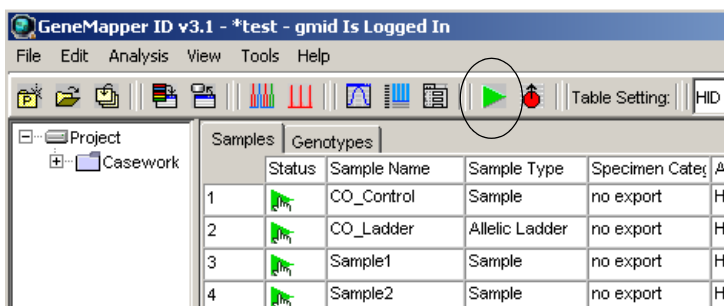
6. Select the desired table setting profile from the drop-down list in the main project window.

Note: You can change this table setting after data analysis at any time to display information in a different table format.

Analyzing the Data

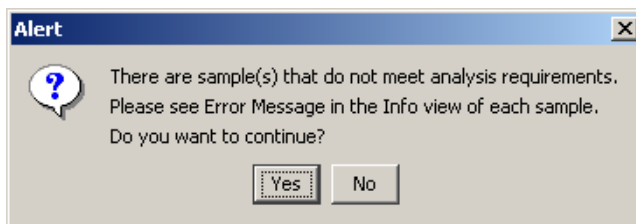
To analyze the data:

1. Click  (Analyze).

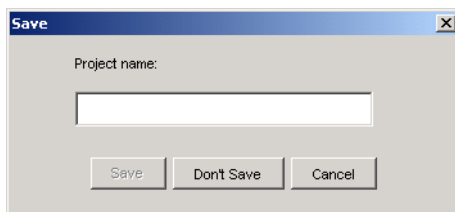


Note: The peak detection algorithm (Classic or Advanced) selected in the Analysis Method must be the same analysis method used to create the size standard definition. The pre-defined size standard definitions that are supplied with the software were created using the Basic/Advanced peak detection

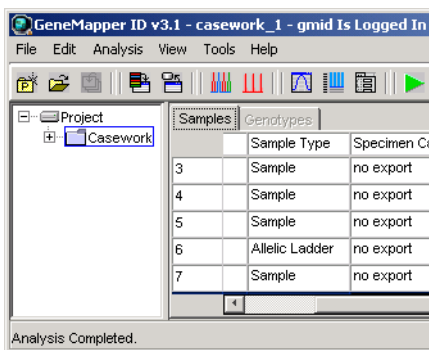
algorithm. If you want to use the Classic peak detection algorithm, define a custom classic size standard definition. If the analysis method and size standard do not match, the software issues the following alert when you press the analyze button:



2. At the Project name prompt, type in the desired name of the project in the Save window, then click **Save**.



During analysis, you can see the progress along the bottom of the project window. Once completed, it will read “Analysis Completed”.



Interpreting Sample PQVs, Sizing Quality, and Precision

Once analysis is complete, check the Sample PQVs (process quality values) and the Sizing Quality (SQ). These flags can be found in the samples tab of the main project window. For an explanation of the sample PQVs, refer to [Appendix A, “Process Quality Values.”](#)

Note: The PQV displayed is determined by the Table Setting you have selected.

Sample PQVS

- If a sample PQV is flagged green, the sample file is considered good quality in respect to that sample PQV.
- If a sample PQV (non SQ) is flagged yellow, look at the raw data to determine the problem. To look at the raw data from the samples tab in the main project window, highlight the sample and select **View > Raw Data**.

If you determine this sample should not be considered for interpretation due to problems within the raw data, you can remove this sample file from the project. Return to the main project window by clicking the “Project” node in the left navigation pane. The sample will still be highlighted. Then select **Edit > Delete from Project**.

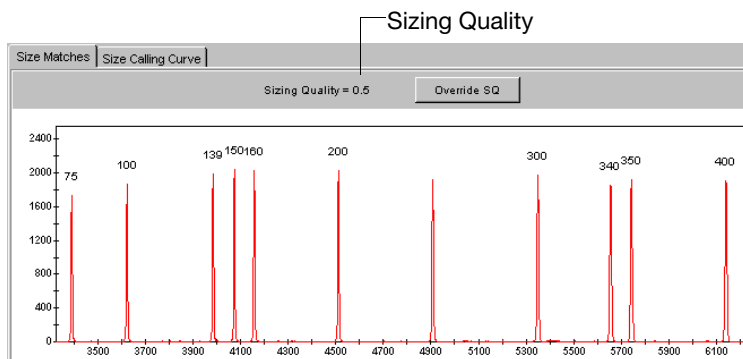
Sizing Quality

Note: Different peak detection algorithms give different Sizing Quality (SQ) results. Therefore, refer to the appropriate section below for instructions on interpreting SQ.

Classic Peak Detection Algorithm

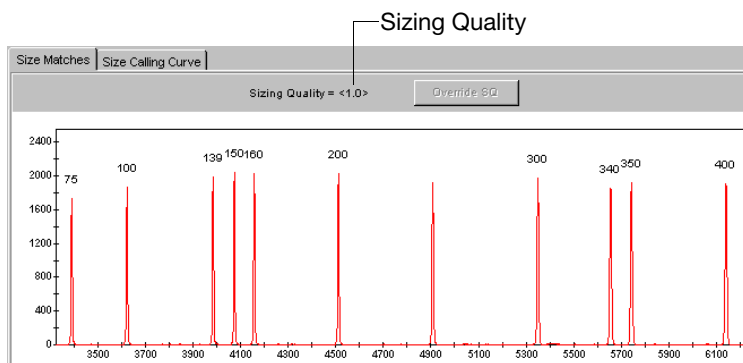
To examine the size standard:

1. Select **Edit > Select All** and click  (Size Match Editor) to view the size standard for all samples.
2. Notice that the Sizing Quality value equals 0.5.



3. View the peak assignments for each sample:
 - a. Press the down arrow key to scroll through the samples on the left side of the Size Match Editor screen.
 - b. Confirm that the size for the peaks in the size standard are correctly assigned for each sample.
4. Select **Edit > Override All SQ** to override the SQ value for all samples.

Notice that the Sizing Quality is changed to <1.0>, which indicates that the size standard was verified by the user.



- Click **Apply**, then click **OK** to close the Size Match Editor and return to the Project window.

Notice that after overriding the SQ values:

- The SQ flags are changed to green squares (Pass).
- The Sizing Quality Override (SQO) column is checked.

SQO	SFNF	SQ
×	■	■
×	■	■
×	■	■
×	■	■
×	■	■
×	■	■
×	■	■
×	■	■

- If one of the following occurs:
 - Size standard peak assignments are incorrect for one or more of the samples in a subsequent analysis.
 - Fewer than three peaks are matched and a red flag is displayed.

Define a new size standard for the affected samples, and then reanalyze the sample.

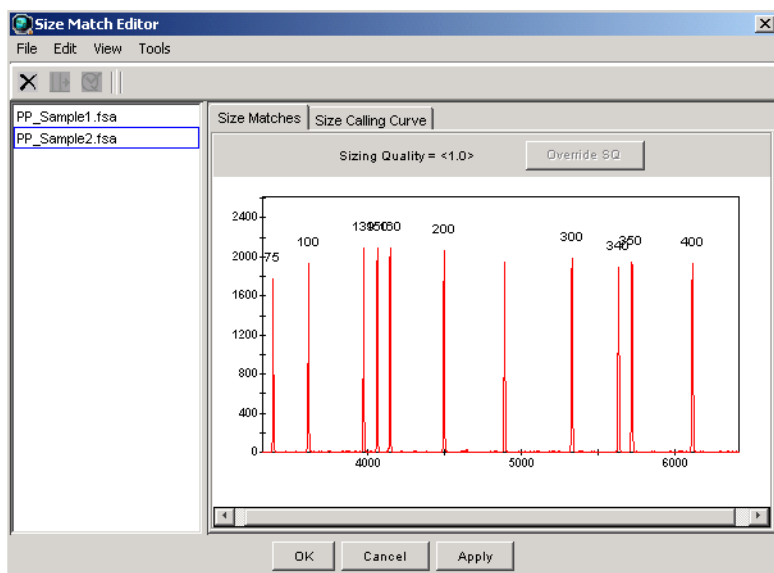
Advanced/Basic Peak Detection Algorithm

To examine the size standard:

- Check the SQ for all samples. Green squares indicate that the samples passed the sizing criteria.
- If the SQ is flagged yellow or red, highlight the row for that particular sample and select **Analysis > Size Match Editor**.
- If the size standard peak assignments are incorrect for one or more of the samples, you can adjust the peak assignments within the size match editor. Refer to the next section for more information about the size match editor.

Size Match Editor

Introduction The Size Match Editor window enables you to examine size standard electropherograms, edit the identification of size standard peaks, and view the size calling curve. To open the Size Match Editor window, click **Analysis** on the toolbar in the GeneMapper *ID* project window and select **Size Match Editor**.



When the Sizing Quality is a low value, you can use the Size Match Editor to adjust the in-lane size standard. Typical problems include the following conditions:

- **Peak shift** - The algorithm does not correctly identify a peak, resulting in invalid positions for other size standard peaks.
- **Missing peak** - The algorithm does not identify a peak.
- **Extra peak** - The algorithm identifies a peak that is not a size standard peak, but a spike or pull-up peak.

Window Elements The Size Match Editor window elements are described in the following table below.

Element	Description
Sample navigation pane	Enables you to select a sample to view by scrolling through the Sample list. The list of samples displayed reflects the list of samples selected in the Project Sample view.
Size Quality indicator	Displays the quality of the currently displayed size standard (name listed above the plot). Value 0.0–1.0.
Size Matches electropherogram	Displays the size standard peaks. Clicking a peak selects it (single selection only).
Size Calling curve	Displays the size calling curve for best fit.
OK button	Closes the Size Match Editor window, accepting the pending changes. For samples whose size standards are modified, the Project window is ready for reanalysis.
Cancel button	Closes the Size Match Editor window, disregarding any pending changes.
Apply button	Same as OK button, except the dialog box remains open.
Override SQ button	Use to override the Sizing Quality value and automatically set the value to 1.0. This button overrides the sizing quality. A check mark displays in the SQ column in the Project window Samples tab.

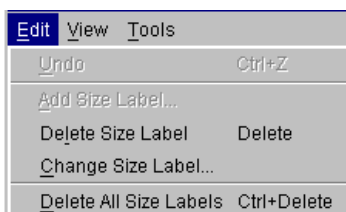
Editor Commands

The following Size Match Editor menus are described in this section:

- Edit
- View
- Tools

Edit Menu

The Edit menu is used to delete or change size labels.

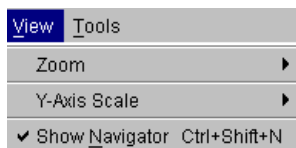


Edit menu items and descriptions:

Item	Description	Enabling
<u>U</u> ndo (Ctrl+Z)	Undoes the last user action.	Enabled after a user action that adds, modifies, or deletes.
<u>A</u> dd Size Label	Adds a size label to the selected peak.	Enabled when an unlabeled peak is selected.
<u>D</u> elete Size Label (Delete)	Deletes the size label of the selected peak.	Enabled when a labeled peak is selected.
<u>C</u> hange Size Label	Opens a dialog box for the selected peak, allowing the selection of a new size label.	Enabled when a labeled peak is selected.
<u>D</u> elete All Size Labels (Ctrl+Delete)	Removes/deletes all size labels for the selected size standard sample.	Always enabled.

View Menu

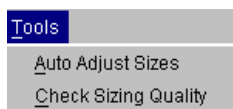
The View menu is used to change the Size Match Editor window.



View menu items and descriptions:

Item	Description	Enabling
Zoom	Submenu used to scale the plot to the following value: Full View (Ctrl+I)	Always enabled.
Y-Axis Scale	- Scale to maximum <u>Y</u> (default) - Scale <u>t</u>o... user defined	Always enabled.
Show Navigator (Ctrl+Shift+N)	Switches the navigator pane (Removes the pane with the first use and restores it on the second use.)	Always enabled.

Tools Menu



Tools menu items and descriptions:

Item	Description	Enabling
Auto Adjust Sizes	Auto adjusts size standard peaks. Tooltip: Auto Adjust	Always enabled.
Check Sizing Quality	Checks the quality of the current size matching. Tooltip: Check Quality	Always enabled.

Toolbar The Size Match Editor toolbar contains the following icons:

Icon	Description	Enabling
	Autoadjusts size standard peaks. Adjusts the size of other peaks automatically to the right of the selected peak. ^a Tooltip: Auto Adjust Sizes	Enabled after a peak is changed.
	Checks the quality of the current size matching. Checks the quality of the revised peak positions and updates the Size Match Quality indicator. ^a Tooltip: Check Sizing Quality	Enabled after a peak label is changed.
	Deletes all size labels. Tooltip: Delete All Size Labels	Always enabled.

a.Only for Basic and Advanced modes.

Using the Size Match Editor

The Size Match Editor window uses the mouse for editing.

Note: All user edited size standard samples automatically have a sizing quality of 1.0, and display a check mark in the SQI column in the Samples window.

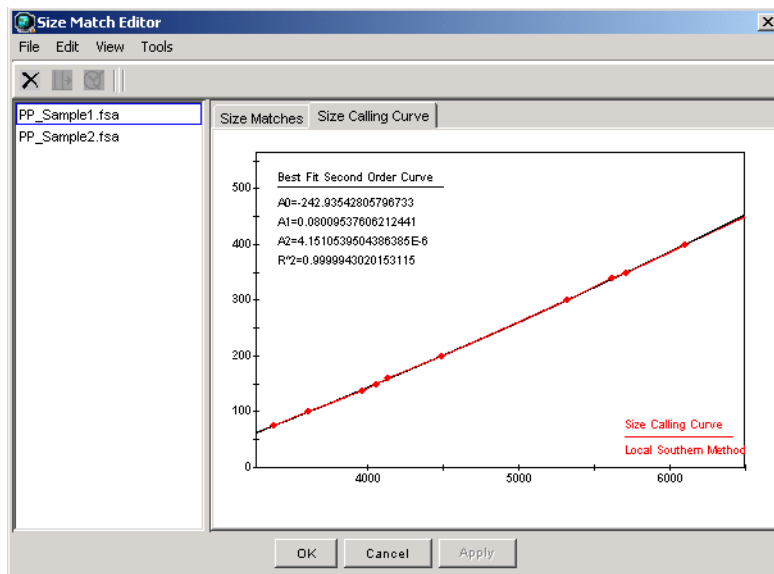
To edit a particular size standard peak:

1. Click the peak to be edited.
2. Right-click the peak to open the editing pop-up window, and Add, Delete, or Change the selected peak.
3. If you chose Add or Change, move the cursor to the right to open the Select Size sub-menu.
Choose the desired height value from this menu.
4. Check the sizing quality by selecting **Check Sizing Quality** under the Tools menu.
If the peaks are sized correctly, but the quality score is still below passing, click the Override SQ button to set the SQ to 1.0.
5. Click **Apply** if you are going to edit a second size standard peak, or click **OK** if you are finished applying your changes to the edited peak.

Size Calling Curve

Overview The Size Calling Curve is used as a measure of how well the internal size standard matches the standard definition, and whether or not it is linear.

The Size Calling Curve displays two curves.



Red Curve If the size calling curve is a red curve, it is based on the size calling method used to analyze the data.

Black Curve If the size calling curve is a black curve, it is a best-fit, least squares curve, which the GeneMapper *ID* software calculates for all samples, regardless of the size calling method used. This curve is provided to help evaluate the linearity of the sizing curve. When the sizing curve and best-fit curve match, they overlap so you see only the size curve.

Note: Sizing problems due to anomalous mobilities may be displayed as non-linear.

**Sizing Precision
(using the 250 bp
peak of GS 500
Size Standard)**

Since the 250 bp peak is not defined when running on capillary electrophoresis platforms, an analyst can use this peak to check the sizing precision (reproducibility).

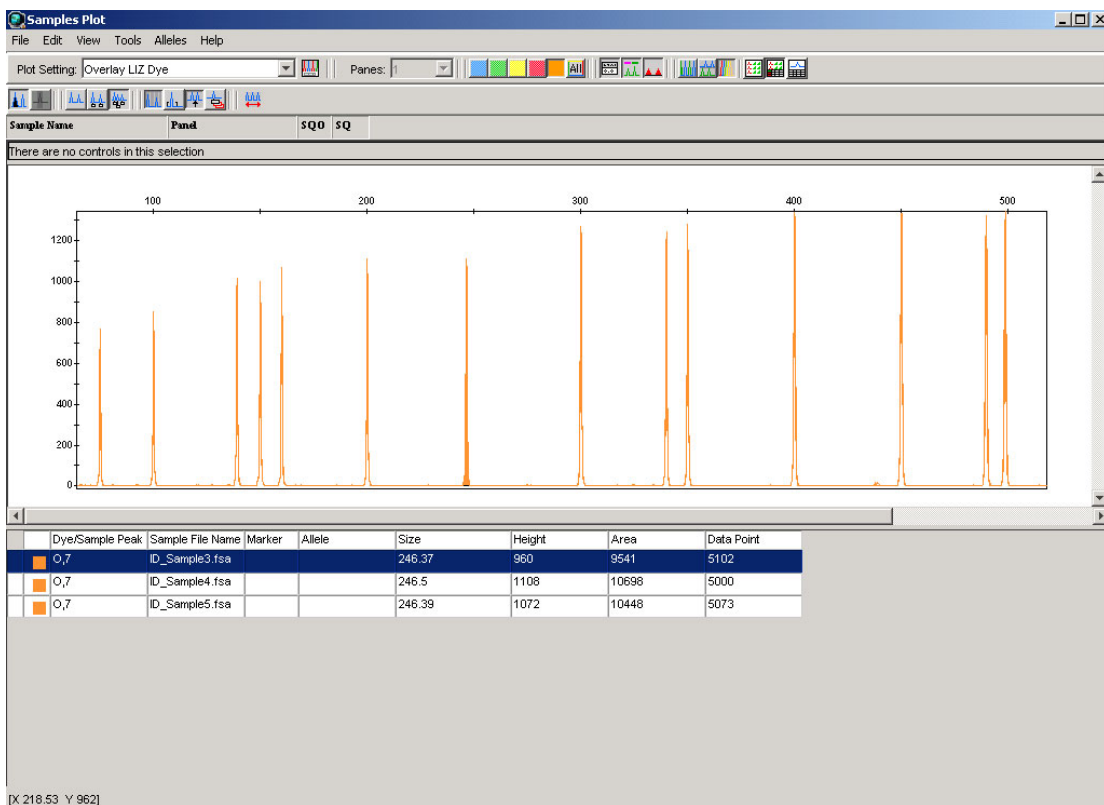
To check size precision:

1. Highlight all the samples in the main project window (**Edit > Select All**)
2. Display the sample plots (**Analysis > Display Plots**).
 - If you have imported the factory provided plot settings (Refer to plot setting sections of Chapter 5), select Overlay LIZ or Overlay ROX depending on the dye color of your size standard from the plot setting drop down box.
 - If you did not import the factory provided plot settings:
 - Select only the color of the size standard in the toolbar
 - Select **View > Plots > Overlay All**
 - select **View > Tables > Sizing**
3. Zoom in on the 250-bp peak:
 - a. Place the cursor to the left of the 250 bp peak, along the top x-axis until the cursor changes to a magnifying glass.
 - b. Click and drag to create a box and release when the box includes the 250-bp peak.
4. Select the 250-bp peak by clicking it.
5. Select **View > Table Filter > Show Selected Rows**.
6. Verify that the values within the size column are within +/- 1 bp

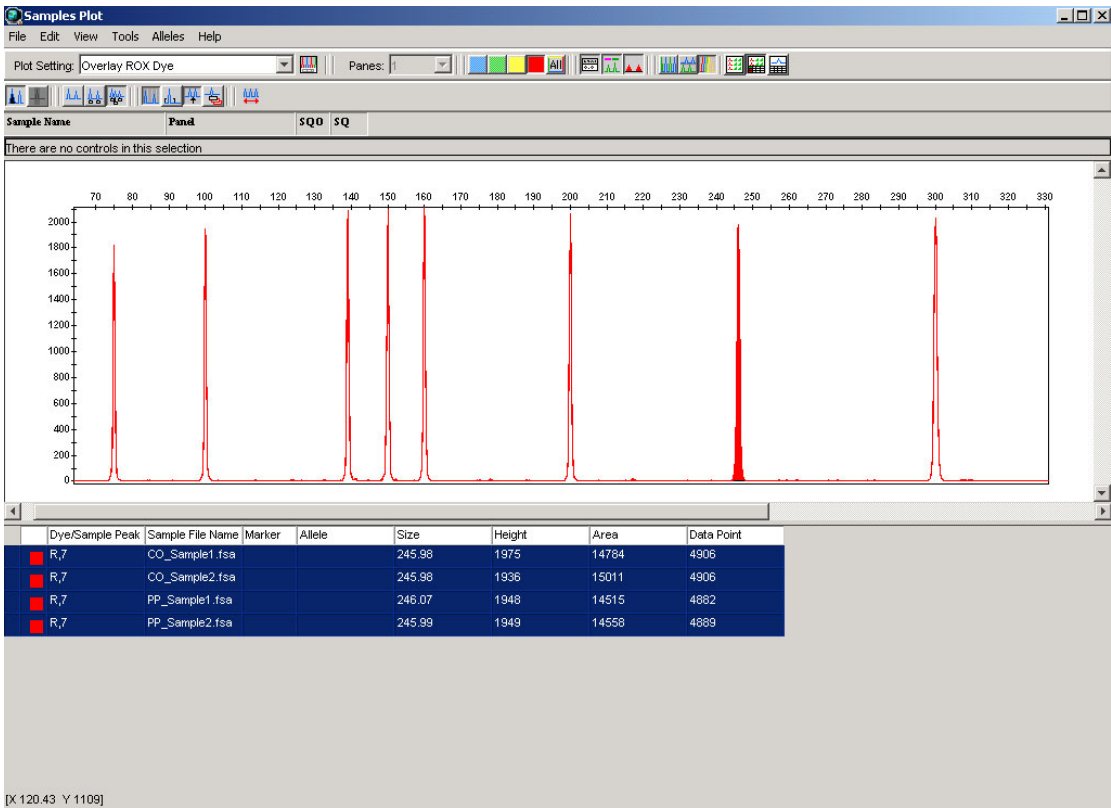
Note: The 250-bp peak should size consistently and overlap. In a typical run, the 250-bp peaks all fall within a size window of approximately 1 bp. Temperature fluctuations in the lab may cause variations > 1 bp.

Overlay LIZ

Use Overlay LIZ to check sizing precision when using a LIZ size standard



Overlay ROX Use Overlay ROX to check sizing precision when using a ROX size standard.



These factory provided defaults should be used as a guideline to customize your plots. Refer to Chapter 5, Section 5.4 for information on creating and saving plot settings, and refer to Chapter 8 for detailed information about plot window toolbars and menus.

Once you have created custom plot settings or imported the factory provided defaults, you can simply switch back and forth between different views by selecting different plot settings from the drop down box in the sample plot window.

Viewing Sample Data

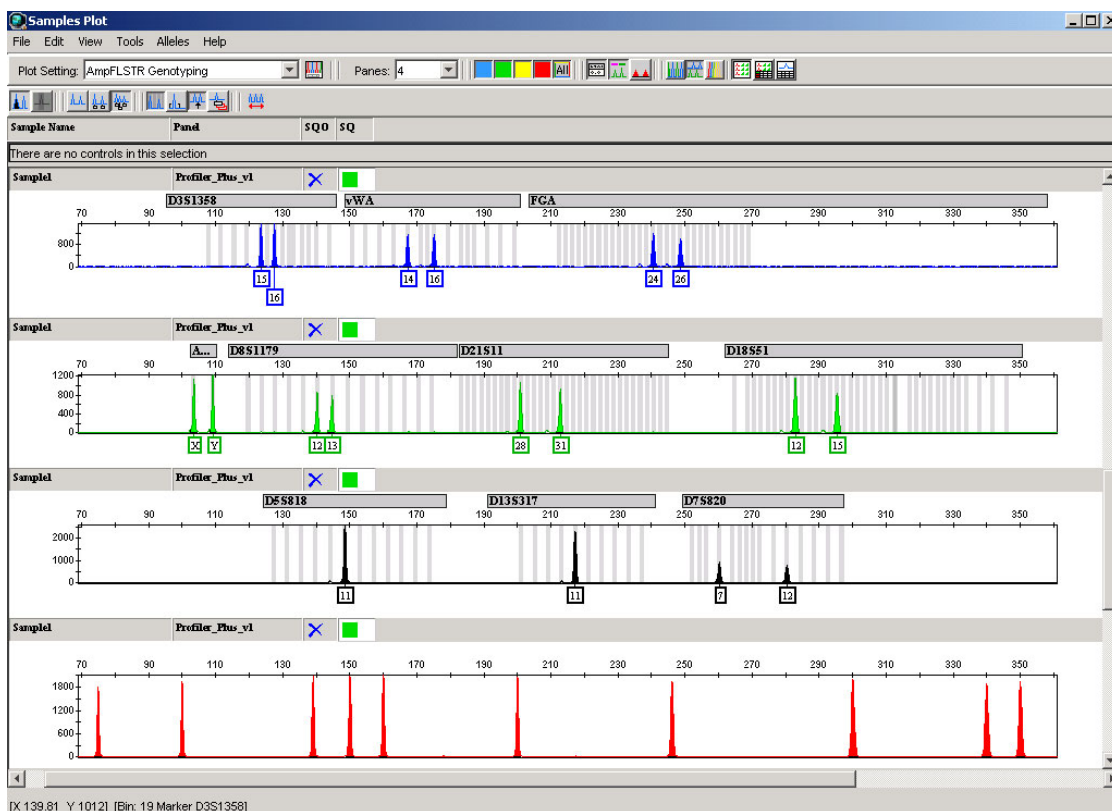
IMPORTANT! If the size quality (SQ) is low, the samples will not be labeled (genotyped).

GeneMapper *ID* v3.1 allows you to fully customize your plot windows. You can create plot settings to re-use, or you can change the plot view as you work.

GeneMapper *ID* v3.1 comes with several default plot settings. These need to be imported into the plot settings tab of the GeneMapper Manager. The plot settings supplied include:

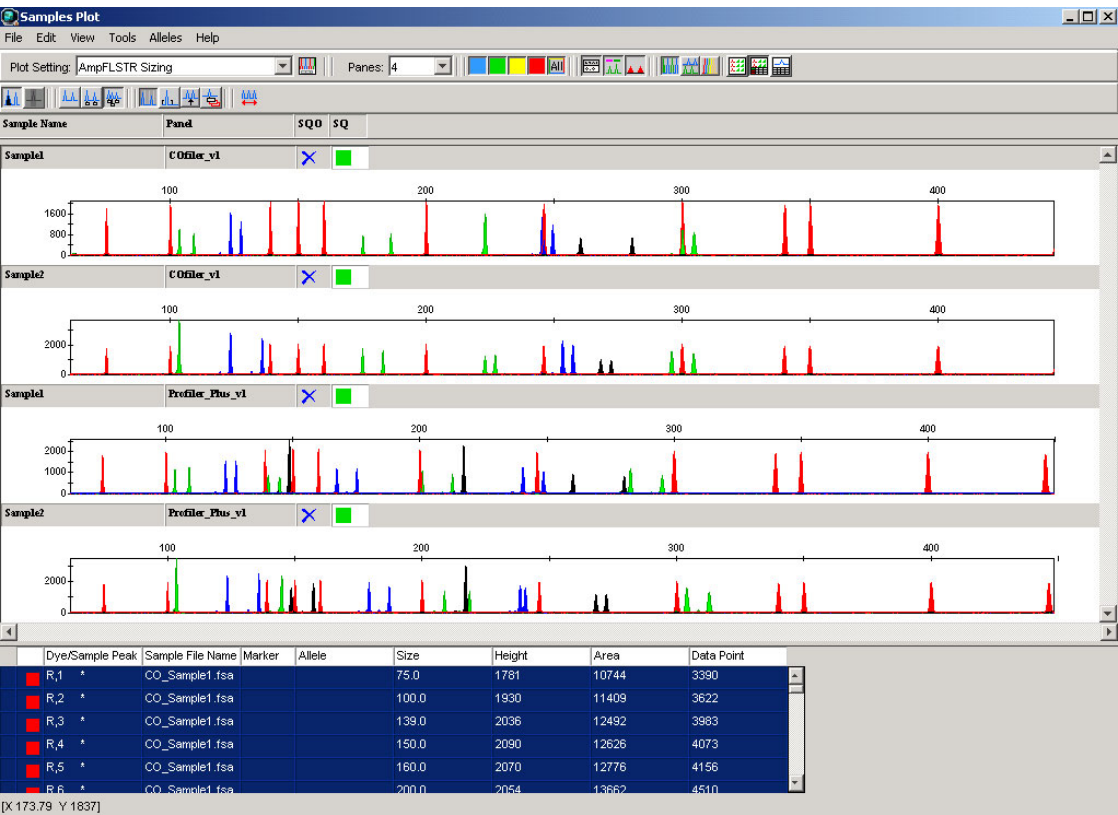
AmpF ℓ STR Genotyping

AmpF ℓ STR Genotyping displays labels on peaks similar to Genotyper plots.



AmpF ℓ STR Sizing

AmpF ℓ STR Sizing displays sizing data similar to GeneScan plots and sizing table.



Editing Allele Calls and Viewing Allele History

If you are going through the labeled plots and you find a mislabeled peak, you can edit these allele calls in the plot windows. When editing alleles, the following rules apply:

- If you delete a label, you will be prompted to enter an allele edit comment, but the comment is not displayed in the genotypes table. To see the allele edit comment, you must display the allele history.
- If you add a label to a previously deleted label, the allele edit comment is displayed in the genotype table, and an “X” is placed in the AE (allele edit) column in the genotypes table.
- If you change the label on a peak, you will be prompted to enter an allele edit comment. This allele edit comment is displayed in the genotypes table and in the allele history, and an “X” is placed in the AE (allele edit) column in the genotypes table.
- When an allele is edited, all PQVs for that marker exhibit a grey triangle to indicate the marker has been modified. The genotype quality (GQ) will show a green square.
- Allele edits made in the plot windows are immediately reflected in the genotypes table.
- When you edit an allele, the software stores a log of the changes that were made in the Allele History.

To edit allele calls:

Note: You must be displaying peaks with labels to edit allele calls.

1. Select the peak by clicking it, and right-click.
 - a. If the peak is already labeled, you have the option to Delete the allele call, Rename Allele, or view the History.
 - b. If the peak is not labeled, you have the option to Add Allele Call, or view History.
2. Choose the appropriate option by clicking on it.

If you are renaming or adding an allele call, choose the allele call or enter a custom name.
3. After you make the necessary change, you will be prompted to enter an allele edit comment. Enter a comment, and press **OK**. The changes are automatically be reflected in the genotypes table.

To view the Allele History:

1. Select the peak by clicking it, and right-click.
2. Select **History** to open the Allele History Window. You can see all the changes made to the allele.

There are 3 types of actions that are displayed:

- Created – is the action in the first row if the peak was labeled by the software automatically. The comment always reads “GeneMapper HID Allele Calling Algorithm.” This action is displayed when you add a label manually to a non-labeled peak
- Deleted – when you remove a label from a labeled peak
- Edited – when you change the label of a labeled peak

Note: If you make multiple edits to an allele, then delete the label. All of the previous edits are NOT listed in the history. Edits are only shown for peaks that have labels. Similarly, allele edit comments are only shown in the genotypes table for peaks that have labels.

Genotypes Table

IMPORTANT! If the overall size quality PQV displayed for all samples is red (low quality), the genotypes table are not accessible.

After viewing plots and editing alleles (as necessary), you can view the genotypes, genotype PQVs, and the overall Genotype Quality in the Genotypes table. This feature provides a tabular format to display the genotypes of your samples and the quality of the samples. Each row corresponds to a locus within a sample. Therefore, if you have 9 loci per sample, you will have 9 rows for that sample within the genotypes tab. This display is also known as a vertical table format. The software is not capable of producing a horizontal table format where each row corresponds to a single sample. To create a horizontal table, export the genotypes table, and make changes using a downstream spreadsheet application, such as Microsoft® Excel. In addition, you can not change the order in which the columns are listed.

What is displayed in the genotypes table is determined by the table setting selected in the Table Setting drop down box in the project window. The table settings are created in the GeneMapper Manager (refer to Chapter 5, Section 5-3). You can simply switch back and forth between table settings by changing the setting in the drop down box. Keep in mind that the table setting change affects both the samples table and the genotypes table.

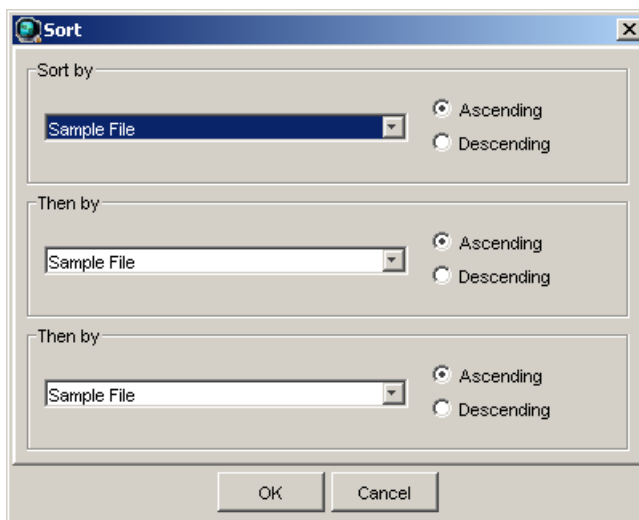
Note: The default settings are designed to display two alleles per marker. Therefore, if you are working with mixtures and need to display more than two alleles per marker, you will need to change the Number of Alleles within the Genotypes Tab of the Table Setting Editor.

Sorting a Genotype Table

You can sort the table by any present column header.

To sort a Genotype table:

1. If the genotypes table is not already displayed, click the genotypes tab in the main project window, or go to **View > Genotypes**.
2. Select **Edit > Sort**. The following window will appear. You can sort up to three criteria.

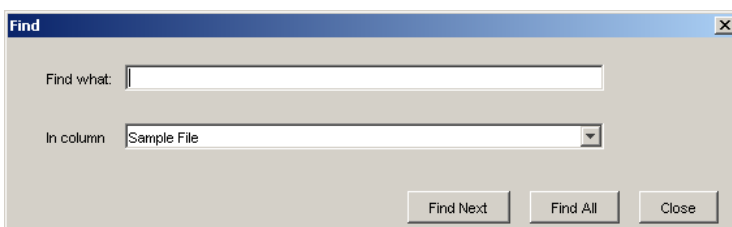


3. Select the criteria to sort by from the drop down boxes and choose ascending (A-Z; 1, 2, 3...) or descending (Z-A; 3, 2, 1).
4. Click **OK**. The table will sort automatically after clicking OK.

Find Option If you are looking for a particular sample, or a particular set of samples, you can use the find option.

To use the Find option:

1. If the genotypes table is not already displayed, click the genotypes tab in the main project window, or select **View > Genotypes**.
2. Select **Edit > Find**.



3. Enter the string of text or numbers you wish to find, and select what column you want the software to look in.
4. Click **Find Next** to find the first instance, or click **Find All** to find all instances at once.

The rows containing this item are highlighted in the genotypes tab.

Genotypes Plots When you have a row(s) highlighted, you can display genotypes plots. Genotypes plots display the peaks for a particular marker (loci). Highlight the desired rows and select **Analysis > Display Plots**. Refer to Chapter 8 for more information about viewing genotype plots.

Interpreting Genotype PQVs and Genotype Quality

Genotype PQVs and Genotype Quality are displayed in the Genotypes Table and Plots. To view the Genotype Table, click the Genotypes Tab from the main project window. Or, if you have selected it to be displayed in your sample plots, highlight samples within the genotype tab and go to **Analysis > Display Plots**.

Genotype PQVs

The PQVs are an easy way to scan through the data and determine which markers (loci) are problematic without going through the electropherograms for every sample file. PQVs are very useful for processing databasing samples or for high-throughput analysis. Refer to Appendix A for a detailed description of each PQV.

To use the Genotype PQVs to evaluate your data:

1. Within the Genotypes tab, select **Analysis > Low Quality to the Top** to bring all samples with low-quality (Red) and questionable (yellow) Genotype Qualities.
2. Highlight all the rows with low-quality and questionable Genotype Qualities.

Note: Green Genotype Qualities may not need any further assessment. Green indicates a high confidence level that the allele calls are accurate.
3. Select **Analysis > Display Plots**.
4. Scroll through the plots using the PQVs to help determine where the problem lies.
5. Decide if you agree or disagree with the software.
 - If you agree, then you might remove the sample from the project, or remove the allele calls for that particular marker.
 - After reviewing a flagged PQV, you can override the GQ by right-clicking the red or yellow GQ flag. You will be asked if you want to override the Genotype Quality of that marker. Click **Yes**. All genotype PQVsturn grey, and the GQ turns green.

Override Genotype Quality

Invalidate Genotype Quality

This feature allows you to quickly reset the Genotype Quality to 1.0 and all PQV icons will be greyed out. Since the allele call has not changed, the AE (allele edit) box will not be checked.

To use the Override Genotype Quality feature in the sample plot:

1. Show the Genotype table.
2. Select the Genotype Quality cell of a genotype.
3. Right-click the selected cell to open the Override Genotype Quality dialog box.
4. Click **Yes**.

The Genotype Quality now passes with a value set to 1.0, and the PQV icons are greyed out in the Genotypes table.

To use the Override Genotype Quality feature in the genotype plot:

1. Select the sample header of a plot.
2. Right-click the header to open the Override Genotype Quality dialog box.
3. Click **Yes**.

The Genotype Quality now passes with a value set to 1.0, and the PQV icons are greyed out in the Genotypes table.

Genotype Quality (GQ)

The Genotype PQVs are used to calculate the overall Genotype Quality (GQ). The user can set level of importance of PQVs in the overall calculation of the Genotype Quality (GQ) in the Quality Flags Tab of the Analysis Method Editor (refer to Chapter 5).

Note: The GQ for a specific marker in a ladder file represents the genotype quality for that marker in that ladder file. One of the components involved in GQ calculation is the ladder matching quality. If the matching quality is not so good (could be caused by various reasons, like high noise/background, significant ladder allele imbalance, or mobility shifts causing significant local nonlinearity), the GQ will be yellow even when all the modulators ("all other flags" in Issue) are green. The Genotype Quality can be displayed as a numerical value instead of a colored symbol. Select this preference in the Analysis Tab within the Options window (under the Tools Menu). Refer to Chapter 6.

Concordance Checks

The software is capable of performing three concordance checks:

- Checking control alleles against expected alleles that are predefined in the panel used.
- Comparing genotypes for overlapping loci within different AmpF $\mathbb{L}$ STR kits. For example, D3S1358 is a locus used in both Profiler Plus and Cofiler. If the same sample was amplified with both kits, the software can check that both amplifications produce the same genotype for the overlapping locus.
- Comparing genotypes for overlapping loci within the same AmpF $\mathbb{L}$ STR kit, *e.g.*, checking the concordance between duplicate amplifications and injections of the same sample.

This section focuses on the second type of concordance check listed above.

Requirements for performing a concordance check include:

- The genotypes must be saved in the local GeneMapper database. Thus, the samples must be analyzed and the project saved.
- The sample files to be compared must have the same name.

Note: If the sample file names are different due to laboratory protocols or automatic naming conventions, you can still perform this procedure by typing a shared name for two or more samples in the UD1 column. You can simply highlight the cell in the UD1 column and type in a common name for the samples to be compared. This information can also be read from a sample sheet. If you type a pipe bar { | } after the panel name in the comment field (Profiler_Plus_v1|sample1), the information after the pipe bar will be displayed in the UD1 column.

To check concordance for shared markers:

1. If the genotypes table is not already displayed, click on the genotypes tab in the main project window, or select **View > Genotypes**.
2. Select **Analysis > Non-concordant Sample to Top**. Any rows that are non-concordant will be brought to the top and highlighted.
3. To view the plots for all non-concordant markers, keep the rows highlighted, and select **Analysis > Display Plots**.

Using Plot Windows - Samples and Genotypes

8

This chapter describes how to examine and interpret electropherograms within the GeneMapper™ ID Software Version 3.1 Samples and Genotypes plot windows, and explains the various electropherogram settings and interactions in the plot window menu items.

This chapter covers:

About the Plot Window	8-2
Plot Window Toolbars	8-4
Plot Window Menus	8-5

About the Plot Window

Introduction The Samples and Genotypes plot windows allow you to visually assess your data. If genotype calls are created, you can view and edit allele calls assigned by the GeneMapper ID software algorithms.

Purpose of the Plot Window

Samples Plot Window

The Samples plot window allows you to:

- View electropherograms on a per sample basis
- View a sizing or genotypes table for sample(s) shown
- Overlay all samples to determine size standard quality
- Edit markers and bins
- View the relationship between controls and samples
- Edit allele calls

Genotypes Plot Window

The Genotypes plot window allows you to:

- View electropherograms on a per genotype basis
- View all genotype calls for a given marker
- Edit markers and bins
- View the relationship between controls and samples
- Edit allele calls

Displaying Plot Windows

Display the plot window from the GeneMapper Project window.

To open the plot window:

1. Highlight any number of samples or genotypes from the project window by selecting the row number or sample file name.
2. Click  (Display Plots) on the toolbar, or **Analysis > Display Plots**.

When you select a sample, all of the corresponding genotypes are automatically selected in the Project Window Genotypes Tab. If a plot window is open, the view refreshes as you switch between tabs.

When you select a genotype, the corresponding sample(s) are automatically selected in the Project Window Samples Tab. If a plot window is open, the view refreshes as you switch between tabs. The genotype selection is maintained until additional samples or genotypes are selected or deselected.

Note: Samples with a low sizing quality (SQ) value cannot be displayed in the Plot window. Only raw data can be viewed for low quality samples.

Note: The Plot Window cannot contain electropherograms from both the Samples and Genotypes tabs at the same time.

Customizing Plot Windows

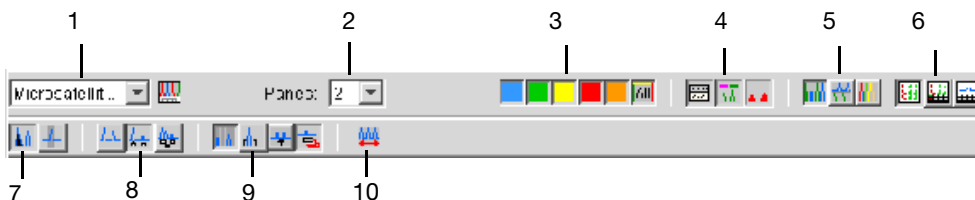
You can customize certain features within the Plot Windows using the pull-down menu items or toolbar icons. Frequently used plot views can be saved as Plot Setting profiles using the Plot Settings Editor accessible from the GeneMapper Manager.

- The last profile used in the Samples Plot is used to open the Genotype plot.
- The last profile used in the Genotypes Plot is used to open the Sample Plot.

Plot Window Toolbars

Toolbar Descriptions

The toolbar icons enable and disable plot features similar to their associated menu items as described throughout this chapter. Position your mouse over an icon to view its tooltip description.



Plot window toolbar icons and descriptions:

Item	Description
1	Pull-down menu that allows selection of a plot profile Note: Click the Plot Settings Editor icon to edit the selected profile. (Tools menu)
2	Pull-down menu that controls the number of electropherogram panes shown
3	Dye color toggle icons (View menu)
4	Header, Marker Range and Marker Indicator toggle icons (View menu)
5	Icons that select the plot display: Combine Dyes, Separate Dyes, Overlay All (View menu)
6	Icons that select the Sample Plot window to be displayed: No Table, Sizing Table, Genotypes Table (View menu)
7	Icons that select either Peak Selection Mode or Binning Mode (Alleles menu)
8	Icons that select the Label display: No Labels, Horizontal Labels, Vertical Labels (View menu)
9	Icons toggle additional view features: Overlay Bins, Show/Hide Peak Position, Controls To Top, Show/Hide Allele History (View menu)
10	Icon switches view to full X-axis and Y-axis scales: Full View from the View menu.

Plot Window Menus

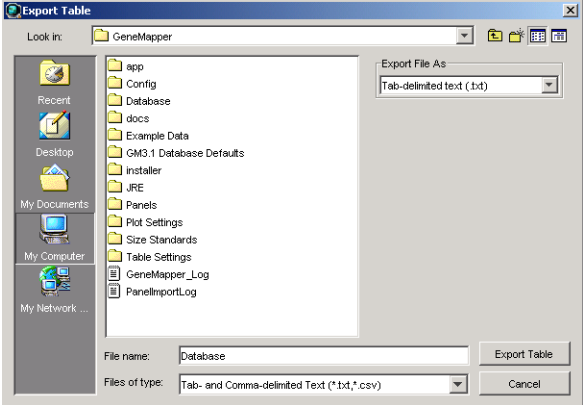
File Menu The File menu commands are used to perform the basic commands.

<u>F</u> ile	<u>E</u> dit	<u>V</u> iew	<u>T</u> ools	<u>A</u> lleles	<u>H</u>
Sa <u>v</u> e Panel					Ctrl+Shift+S
<u>P</u> rint					Ctrl+P
<u>E</u> xport Table					Ctrl+E
<u>C</u> lose Plot Window					Escape

File menu items and descriptions:

Command	Description
Save Panel (Ctrl+Shift+S)	Opens the Save Panel(s) dialog box informing you of what panel(s) have been modified and how many projects currently in the GeneMapper <i>ID</i> database will be affected. The options are: • Save Panel - This option allows you to save any marker and bin changes made to the current panel name, affecting all projects analyzed using this panel. • Save Panel As - This option allows you to save any marker and bin changes made to a new panel name, affecting only the current project being analyzed. Note: When using Save Panel As feature, the application creates a duplicate panel with bins from the current bin set and all bins from associated bin sets. • Discard Changes - This option allows you to discard any changes made to markers and bins.
Print (Ctrl+P)	Automatically prints the contents of the plot window in the following order: • controls • samples/genotypes • table (if any) Note: Customize the view prior to printing.

File menu items and descriptions: *(continued)*

Command	Description
<u>E</u> xport Table (Ctrl+E)	Opens the Export Table dialog box allowing you to export either the sizing or genotypes table shown in the Samples Plot window.  Note: This command is functional only when a table is currently being viewed. The exported table contains the same configuration as the currently viewed table.
<u>C</u> lose Plot Window (Escape)	Prompts to save any pending changes and closes the plot window.

Edit Menu

The Edit menu command provides basic editing for the Plot window menus described in this section.

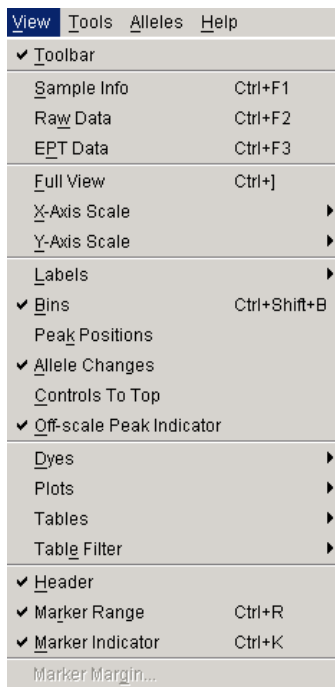


Edit menu item and description:

Item	Description
<u>U</u> ndo (Ctrl+Z)	The Undo command is enabled after a user action that adds, modifies, clears, or reorganizes. It allows you to undo certain actions, such as: • Editing an Allele • Editing a Bin • Resizing Markers • Zooming

View Menu The View menu contains features you can use to control:

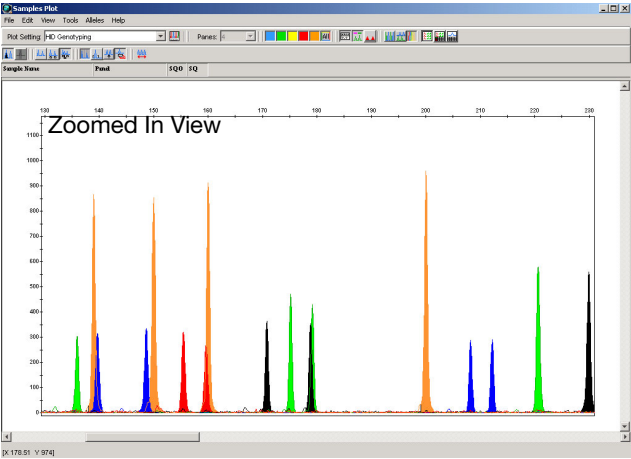
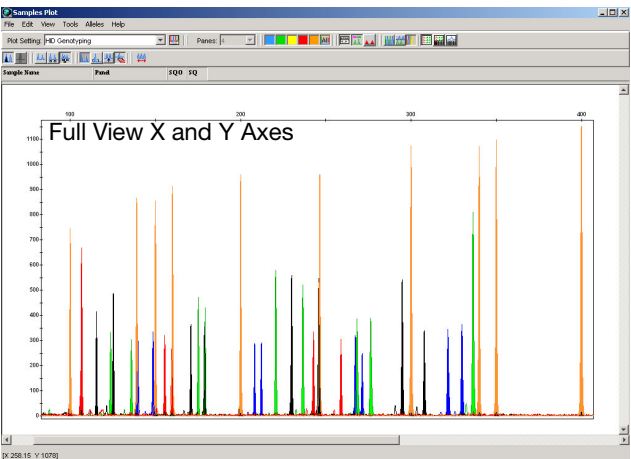
- zooming of electropherograms
- electropherogram lines/dye displays on the view
- a number of display activities



View menu items and descriptions:

Item	Description
Toolbar	Controls whether or not the Plot toolbar is displayed at the top of the Plot window. This item is checked when active.
Sample Info (Ctrl+F1)	Provides quick access to the Sample Info data tab for the selected electropherogram. (See Chapter 3, “Using the Project Window.”)
Raw Data (Ctrl+F2)	Provides quick access to the Raw Data tab for the selected electropherogram. (See Chapter 3, “Using the Project Window.”)

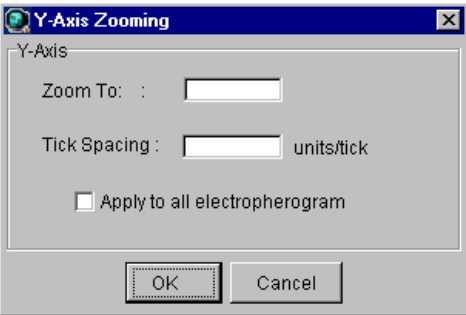
View menu items and descriptions: (continued)

Item	Description
EPT Data (Ctrl+F3)	Provides quick access to the EPT Data tab for the selected electropherogram. (See Chapter 3, "Using the Project Window.")
Full View (Ctrl+J)	Automatically resets the X- and Y-axes of the electropherogram(s) currently viewed to their full scale size.  Note: Double-clicking with the left mouse button on an axis automatically resets that axis to full view. 

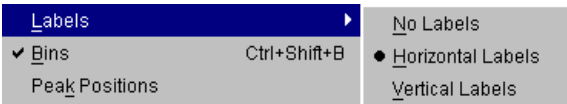
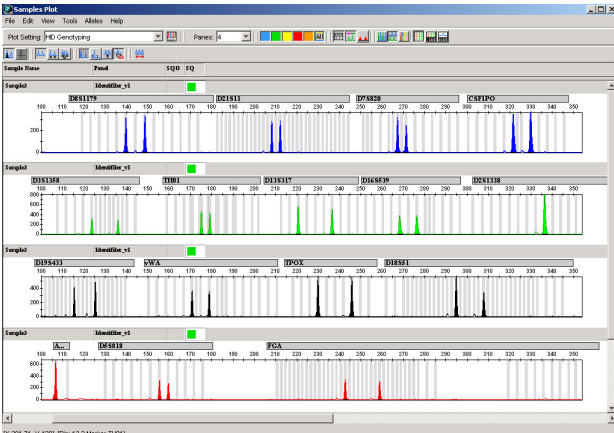
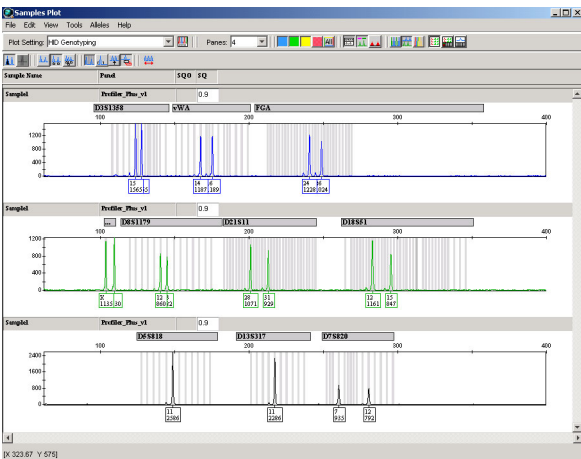
View menu items and descriptions: *(continued)*

Item	Description
X-Axis Scale	Selects the horizontal scale units for the X-axis to be displayed in either base pairs (default) or data points.  Zooms in on a specific region on the X-axis of the electropherogram as follows: - Click and drag, with the left mouse button, the desired region on the X-axis. In the Samples Plot, this action affects all electropherograms. In the Genotypes Plot, this action affects only the selected electropherogram, or - Right-click the X-axis to open the Zoom To dialog box, which enables specific zoom coordinates and tick spacing units to be set. Double-click the X-Axis Sample Plot and all the electropherograms return to Full View. Double-click the X-Axis Genotype Plot and only the selected electropherograms return to Full View. 

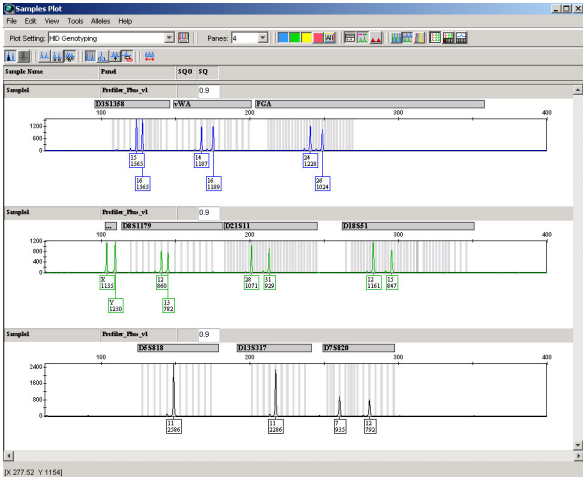
View menu items and descriptions: *(continued)*

Item	Description
Y-Axis Scale	Controls the vertical scale of the electropherograms currently viewed as follows: • Scale Individually - Each electropherogram being viewed is scaled to its maximum height. • Scale To Maximum Y - All plots currently viewed are scaled to a global maximum value. • Scale To - This setting allows a user to set the Y-Axis scale value. If multiple plots are currently viewed, all plots are scaled to this value.  Zooms in on a specific region on the Y-axis of the electropherogram as follows: • Click and drag with the left mouse button the desired region on the Y-axis. This action only affects the selected electropherogram, or • Right-click the Y-axis to open the Zoom To dialog box, which enables specific zoom coordinates and tick spacing units to be set. Use the Apply To checkbox to apply the settings to all electropherograms (panes).  • Double click on the Y-Axis to return selected electropherograms to Full View. • Shift+double click on the Y-Axis to return all electropherograms to Full View.

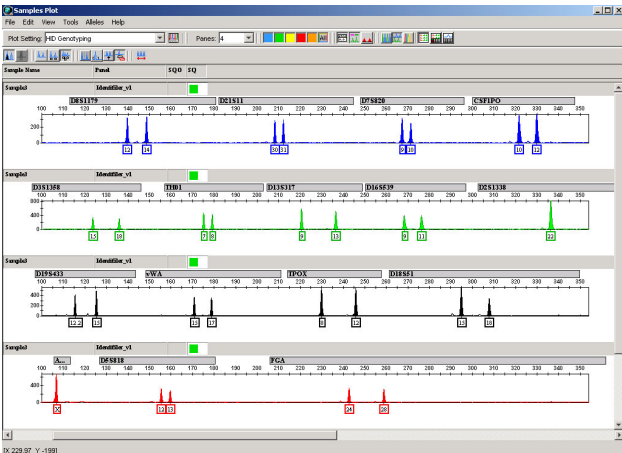
View menu items and descriptions: (continued)

Item	Description
Labels	Controls how labels are viewed beneath an electropherogram.  No Labels - Labels are off.  Horizontal Labels - Labels are positioned horizontally across the screen. Labels in close proximity overlap with the left most label displayed on top. Click any label to bring it to the top for viewing. 

View menu items and descriptions: (continued)

Item	Description
<u>L</u> abels (continued)	Vertical Labels - Labels are positioned vertically across the screen to prevent overlap. This option is recommended for printing.  The screenshot shows the 'Samples Plot' window with three stacked chromatograms. Each plot has a y-axis representing signal intensity (0 to 1200 or 2400) and an x-axis representing genomic position (0 to 400). The top plot is for 'D3S1328' and 'FCA', showing peaks labeled with '12' and '14'. The middle plot is for 'D13S317' and 'D13S317', showing peaks labeled with '12' and '14'. The bottom plot is for 'D13S317' and 'D13S317', showing peaks labeled with '12' and '14'. The labels are positioned vertically to prevent overlap. Note: Labels are enabled in Separate Dyes mode only for Microsatellite samples, and are enabled in Combine Dyes mode only for SNaPshot® kit samples.

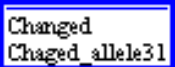
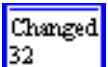
View menu items and descriptions: *(continued)*

Item	Description
Bins (Ctrl+shift+B)	Controls whether or not transparent bins are shown or hidden on an electropherogram. In the Binning mode, bins are automatically displayed and the Bins menu item is checked. Separate Dyes mode  Note: Bins are enabled in Separate Dyes mode only for Microsatellite samples, and are enabled in Combine Dyes mode only for SNaPshot kit samples. When you turn off a specific dye color for SNaPshot kit samples, the bins for that color turn off also.

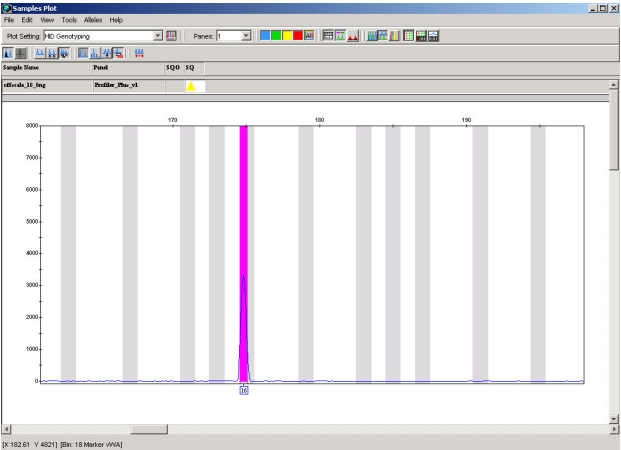
View menu items and descriptions: (continued)

Item	Description
Bins (continued)	Combine Dyes Mode Note: Bins are enabled in Separate Dyes mode only for microsatellite samples, and are enabled in Combine Dyes mode only for SNaPshot kit samples. When you turn off a specific dye color for SNaPshot kit samples, the bins for that color turn off also.
Peak Positions	Shows/hides the peak position indicators on the electropherogram currently viewed. Checked when active. Disabled for overlaid samples.

View menu items and descriptions: *(continued)*

Item	Description
Allele Changes	Shows/hides label editing effects. • Turned Off – Deleted label disappears – Edited/Added label looks normal • Turned On, labels manually edited are shown with descriptors and the allele call. Label deleted - Allele label is shown with a diagonal slash through it.  Label changed - Allele label is shown with a double upper bar.  Label added - Allele label is shown with a double upper bar. 
Controls To Top	Controls whether or not the control samples are displayed at the top of the Plot window. Checked when active. When active, a new pane is displayed at the top of the Plot window. Only samples and their corresponding genotypes designated as Control or Allelic Ladder in the Sample Type column of the Project window display in this pane. If multiple controls are shown, use the scroll bar to scroll through the different controls. Note: Any samples designated as Positive Control, Negative Control, and/or Allelic Ladder in the Sample Type column are affected. If only the controls are selected, only one panel is displayed with a scroll bar function.

View menu items and descriptions: (continued)

Item	Description
<u>O</u> ff-scale Peak Indicator	Controls whether or not offscale peaks are highlighted in an electropherogram. Checked when active. Note: When highlighting of offscale peaks is enabled, a magenta bar overlays any offscale peak in an electropherogram as shown in the figure below. An offscale peak is a peak whose height exceeds the dynamic range of the collection instrument.  The screenshot shows the 'Samples Plot' window. The plot area displays an electropherogram with several vertical grey bars representing peaks. One peak is highlighted with a magenta bar, indicating it is offscale. The y-axis ranges from 0 to 8000, and the x-axis shows positions 170, 180, and 190. The status bar at the bottom indicates 'X:182.61 Y:45211 Bin:10 Marker:VWA'.
<u>D</u> yes	Controls the dye colors shown in the Plot window. Checked when active. Multiple dye colors may be selected. Note: This feature is not available when viewing microsatellite genotypes in the Genotype Plot window.  The screenshot shows the 'Dyes' menu. It lists several options with checkboxes and keyboard shortcuts: ☒ <u>B</u>lue Dye Ctrl+1 ☒ <u>G</u>reen Dye Ctrl+2 ☒ <u>Y</u>ellow Dye Ctrl+3 ☒ <u>R</u>ed Dye Ctrl+4 ☒ <u>O</u>range Dye Ctrl+5 ☒ <u>A</u>ll <u>D</u>yes Ctrl+6 Other menu items include 'Plots', 'Tables', 'Table Filter', 'Header', and 'Marker Range' (Ctrl+R).

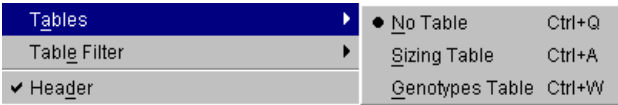
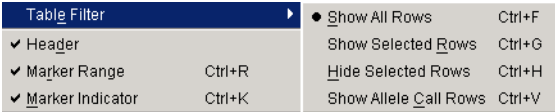
View menu items and descriptions: *(continued)*

Item	Description
Plots	Controls the way electropherograms of samples are shown in the Sample Plot window. • Combine Dyes - All dye colors from a sample are combined into a single source electropherogram.

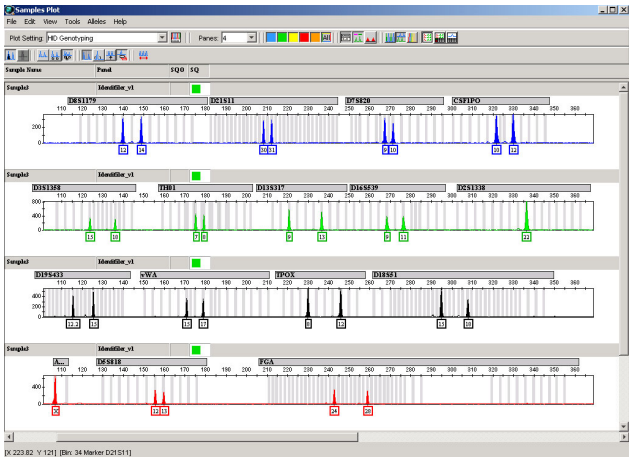
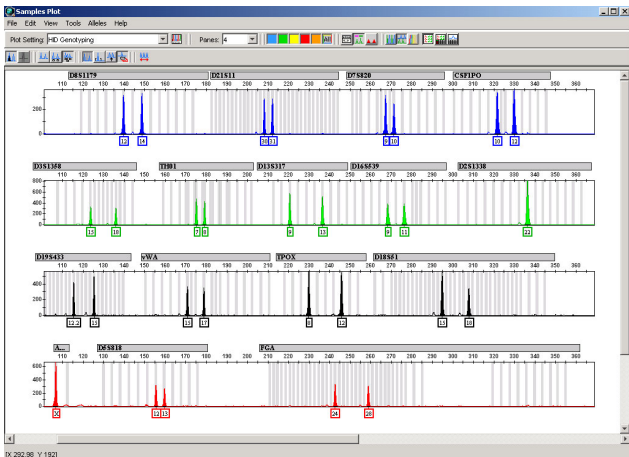
View menu items and descriptions: (continued)

Item	Description
Plots (continued)	Separate Dyes - Each dye color from a sample is separated into an individual electropherogram. Overlay All - Multiple samples are combined into a single electropherogram. The Overlay All option can be used to verify size standards as follows: 1. Select any number of Samples in the Project window. 2. Click (Display Plots). 3. Turn off all dye color icons except the dye color used for the size standard. 4. Click (Overlay All) to display size standard electropherograms, allowing you to check for outliers.

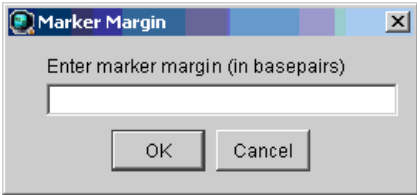
View menu items and descriptions: *(continued)*

Item	Description
Tables	Controls which table, if any, is viewed at the bottom of the Sample Plot window.  • No Table - Turns off Table. • Sizing Table - Shows sizing table, which displays information for all detected peaks in the selected samples. Use the Plot Settings Editor to configure the columns shown in the table. Use the Table Filter menu item command to control which rows of data are shown. • Genotypes Table - The genotypes table, as displayed in the Project window Genotypes tab, is shown for the selected samples. Use the Table Setting Editor to configure the columns shown in the table.
Table Filter	Controls which data rows are displayed in the Sizing or Genotypes Table viewed at the bottom of the Sample Plot window.  • Show All Rows - Shows all available data rows. • Show Selected Rows - Shows only peaks that are selected in the electropherogram(s). • Hide Selected Rows - Hides all peaks that are selected in the electropherogram(s). • Show Allele Call Rows - Shows only peaks that have allele calls assigned. This control is applicable to the Sizing Table only.

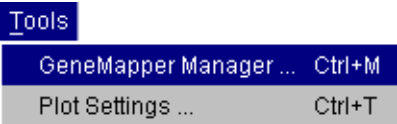
View menu items and descriptions: (continued)

Item	Description
Header	Controls whether the individual Sample or Genotype Header is shown above an electropherogram. Checked when active. Note: The information shown in the Header can be configured through the Plot Setting Editor. • Headers Turned On  • Headers Turned Off 
Marker Range (Ctrl+R)	Controls whether or not the colored marker bar is displayed at the top of an electropherogram.

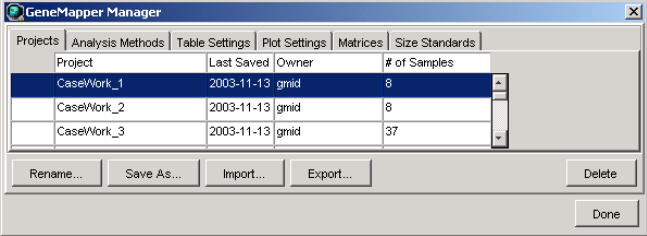
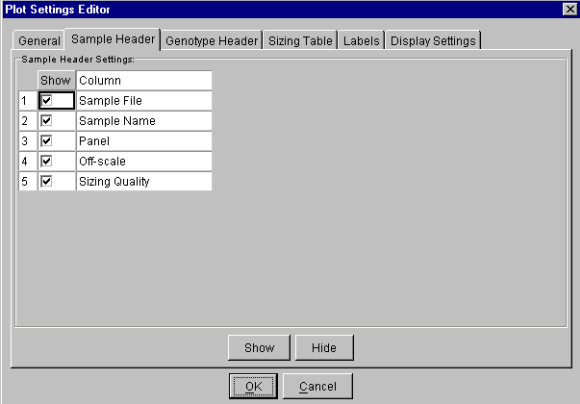
View menu items and descriptions: *(continued)*

Item	Description
<u>M</u> arker Indicator (Ctrl+K)	Controls whether or not the red triangles at the bottom of the electropherogram, which indicate the marker allele size range, are displayed.
Marker Margin	Opens the Marker Margin dialog box to allow you to determine how many base pairs beyond the allele size range are displayed in the electropherogram. Note: This option is only enabled when viewing the Genotype plots. The value can be pre-configured through the Plot Setting Editor. 

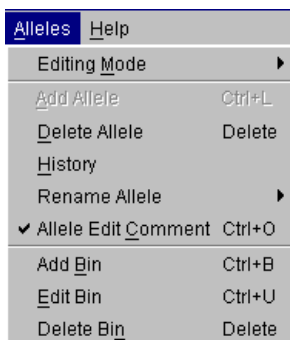
Tools Menu The Tools menu provides access to the GeneMapper Manager and the Plot Settings Editor.



Tools menu items and descriptions:

Item	Description
<u>G</u> eneMapper Manager (Ctrl+M)	Opens the GeneMapper Manager to allow creation or editing of various project settings. Used to create a new Plot Setting profile. See Chapter 2, “Getting Started with GeneMapper ID Software,” for more information.  Note: New profiles can only be created through the GeneMapper Manager.
<u>P</u> lot Settings (Ctrl+T)	Opens the Plot Settings Editor to allow the currently selected Plot Setting profile to be edited. 

Alleles Menu The Alleles menu, used to edit allele call labels on selected peaks, is only enabled when labels are displayed in the Plot window and one or more peaks are selected.



Alleles menu items and descriptions:

Item	Description
Editing Mode	Controls whether the action of the mouse within the electropherogram is in Peak Selection or Binning mode. In Peak Selection mode you can: • Select peaks • Edit allele calls Clicking on a peak automatically highlights that peak's data row within the Sizing Table or (Genotype Table, if an allele call has been made). • Select peaks continuously Use the Shift key or Click+Drag inside the plot area. • Select discontinuous peak selection Use the CTRL key. The selected peak is maintained when switching between the Samples and Genotypes tabs in the Project window with the Plot view still visible.

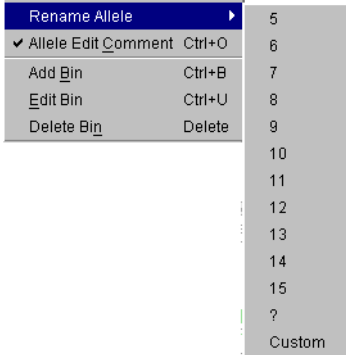
Alleles menu items and descriptions: *(continued)*

Item	Description
Editing <u>M</u> ode (continued)	In Binning mode you can: - Edit marker size ranges - Edit bins When the Binning mode is selected, the Plot window automatically switches the display configuration to an appropriate view for editing bins. - Plots shown in Separate Dyes mode for microsatellite samples - Plots shown in Combine Dyes mode for SNP genotyping samples - X-axis displayed in base pairs - Marker Indicator and Marker Range displayed - Bins displayed To edit marker size ranges (for microsatellites only): - Select Binning mode. - Click the marker indicator of the marker to be edited. A red indicator displays. - Drag the red indicator line to the desired size range. Note: To edit allele calls, refer to Add Allele, Delete Allele, and Rename Allele rows in this table. To edit Bins, refer to Add Bin, Edit Bin, and Delete Bin rows in this table. To edit marker size ranges for SNPs, the bins need to be edited. Binning Mode is disabled when: - There is no bin set for the panel. - The bin set was deleted.

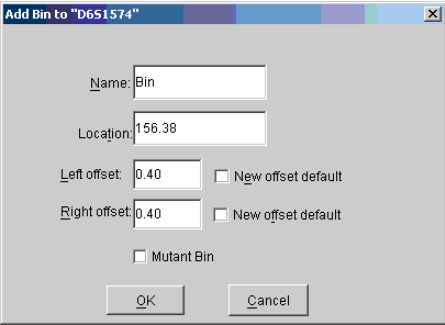
Alleles menu items and descriptions: *(continued)*

Item	Description
<u>A</u> dd Allele (Ctrl+L)	Allows you to add a new allele call to an unlabeled peak as follows: 1. Select Peak Selection mode. 2. Select an unlabeled peak using the left mouse button. 3. Select the Add Allele menu item or right-click mouse button to open the Add Allele dialog box. If Allele Edit Comment is selected, the Add Allele Comment dialog box opens, allowing you to enter an allele comment for the selected peak. Labels must be turned on. A bin name is assigned to the label if the selected peak falls within a bin defined for the marker. Otherwise, the allele is designated with a question mark (“?”) until you rename the allele. If the Show type of edit checkbox is selected in the plot profile, the word “Added” displays inside the box. Note: For quick access to “Add Allele Call,” double-click an unlabeled peak.
<u>D</u> elete Allele (Delete)	Allows you to delete an allele call from a labeled peak as follows: 1. Select Peak Selection mode. 2. Select a labeled peak using the left mouse button. 3. Select the Delete Allele menu item or right-click mouse button to open the allele edit options and click Delete. If Show Allele Edit Comment is selected, the Delete Allele Comment dialog box opens allowing you to enter an allele comment for the selected peak. The label is then deleted. If the Show type of edit checkbox is selected in the plot profile, the word “Removed” displays inside the label box (if Allele Changes is selected from the View menu). Note: Multiple alleles may be deleted at the same time by selecting multiple peaks. This is constrained to the peaks in the plots currently shown.
<u>H</u> istory	Allows you to view the allele history in columns labeled Basepair, Allele Name, User Name, Modification Date, Action, and Comments.

Alleles menu items and descriptions: *(continued)*

Item	Description
Rename Allele	 Allows you to change the allele call on a labeled peak. 1. Select Peak Selection mode. 2. Select a labeled peak using the left mouse button. 3. Select the Rename Allele menu item or click with the right mouse button to open the allele edit options and click Rename Allele. 4. Select an allele call from the menu to rename the allele. If you select Allele Edit Comment, the Edit Allele Comment dialog box opens allowing you to enter an allele comment for the selected peak. The allele is then renamed to the selected allele call. If Show type of edit prefix is turned on in the plot profile, the word “Changed” displays inside the label box.
Allele Edit Comment (Ctrl+O)	Controls whether or not the Allele Comment dialog box opens when you make label changes. Checked when active. To view the comments entered: • Show the allele edit (AE) comment column in the Project Window Genotypes Table, or • Select the History option when right-clicking a selected labeled peak, or • Double click a label.

Alleles menu items and descriptions: *(continued)*

Item	Description
Add <u>B</u> in (Ctrl+B)	Allows you to add a marker bin as follows: 1. Select the Binning mode. 2. Click the Marker Range for the bin. A line and a cross hair opens on the electropherogram. a. Select the Add Bin menu item or right-click to open the Add Bin option. b. Add a bin by either dropping the cross hair at the center of the desired bin location or by dragging across the desired bin range. The Edit Bin dialog box opens allowing you to add bin information. 
Edit <u>B</u> in (Ctrl+U)	Allows you to edit a marker bin as follows: 1. Select Binning mode. 2. Select the bin to be edited. 3. Select the Edit Bin menu item, or right-click the selected bin to open the bin edit options, and select Edit Bin. The Edit Bin dialog box opens allowing you to edit bin information. To quickly resize a bin, select a bin and adjust the right and left handles to the desired size range. To relocate or move the bin (mouse action only): 1. Select Binning mode. 2. Select the bin to be moved. 3. Click and drag the center of the bin to a new location.

Alleles menu items and descriptions: (continued)

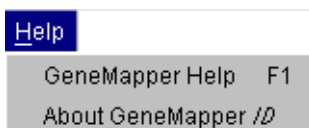
Item	Description
Delete Bin (Delete)	Allows you to delete a marker bin as follows: 1. Select Binning mode. 2. Select the bin to be deleted. 3. Select the Delete Bin menu item, or right-click the selected peak to open the bin edit options and then select Delete Bin. The bin will then be deleted.

Move Allele Using the mouse, you can move an allele label to a new peak.

To move an allele:

1. Left click a label to select it.
 2. Hold down the Ctrl key and drag the label to the new peak.
- The old label will be marked “removed.” The new label will be marked “added.”

Help Menu The Help menu provides access to this user guide in PDF and to the About GeneMapper *ID* software window.

**Help menu items and descriptions:**

Item	Description
GeneMapper Help (F1)	Opens the GeneMapper <i>ID</i> User Guide PDF file using Adobe Acrobat Reader.
About GeneMapper <i>ID</i>	Opens the About GeneMapper <i>ID</i> window that indicates registration information and software version number.

Exporting Table Data

This chapter describes how to export table data.

This chapter covers:

Introduction	9-2
Exporting Samples and Genotypes Table	9-2
About CODIS	9-5
CODIS Export Manager	9-6
CODIS Table Export	9-8

Introduction

You can export the samples table or the genotypes table. These tables can then be imported into a spreadsheet application, such as Microsoft® Excel. In addition, the data can be exported in a compatible FBI Laboratory Combined DNA Index System (CODIS) format.

Exporting Samples and Genotypes Table

The exported table reflects what is displayed in the samples or genotypes tabs. Therefore, the exported table depends on what table setting you have selected when you export the table. In addition, the table is organized in a vertical format, displayed one marker per row.

Viewing the Exported Table in a Spreadsheet Application

The sample and genotype PQVs do not display as color symbols when you import the table into a spreadsheet application. The Sizing Quality (SQ) and Genotype Quality (GQ) display as a numeric value from 0 to 1. The lowest quality is 0 and 1 is the highest value. All other PQVS have one of the following values:

Value in Spreadsheet	Definition
TRUE	Yes, there is an “X” in this cell
FALSE	No, there is not an “X” in this cell
0	Green (pass)
1	Yellow (check)
-1	Grey (Edited)
-2	N/A (Not Applicable)

Casework Project																						
	C	D	E	F	G	H	I	J	K	L	M	N	O	P	Q	R	S	T	U			
1	Panel	Marker	Dye	Allele 1	Allele 2	AE C _t	AE C _t	ADO	AE	OS	BIN	PHR	LPH	SPU	AN	BD	CC	OVL	GQ			
2	Cofiler_v1	D3S1358	B	14	15			FALSE	FALSE	0	0	0	0	0	0	0	0	0	0	0.5		
3	Cofiler_v1	D16S539	B	11	12			FALSE	FALSE	0	0	0	0	0	0	0	0	0	0	0.5		
4	Cofiler_v1	AMEL	G	X				FALSE	FALSE	0	0	-2	0	0	0	0	0	0	0	0.5		
5	Cofiler_v1	TH01	G	8	9.3			FALSE	FALSE	0	0	0	0	0	0	0	0	0	0	0.5		
6	Cofiler_v1	TPOX	G	8				FALSE	FALSE	0	0	-2	0	0	0	0	0	0	0	0.5		
7	Cofiler_v1	CSF1PO	G	10	12			FALSE	FALSE	0	0	0	0	0	0	0	0	0	0	0.5		
8	Cofiler_v1	D7S820	Y	10	11			FALSE	FALSE	0	0	0	0	0	0	0	0	0	0	0.5		
9	Cofiler_v1	D3S1358	B	12	13			TRUE	FALSE	0	-2	-2	0	0	0	0	-2	0	0.78			
10	Cofiler_v1	D16S539	B	5	8			TRUE	FALSE	0	-2	-2	0	0	0	0	-2	0	0.78			
11	Cofiler_v1	AMEL	G	X	Y			FALSE	FALSE	0	-2	-2	0	0	0	0	-2	0	0.92			
12	Cofiler_v1	TH01	G	5	6			TRUE	FALSE	0	-2	-2	0	0	0	0	-2	0	0.9			
13	Cofiler_v1	TPOX	G	6	7			TRUE	FALSE	0	-2	-2	0	0	0	0	-2	0	0.92			
14	Cofiler_v1	CSF1PO	G	6	7			TRUE	FALSE	0	-2	-2	0	0	0	0	-2	0	0.86			
15	Cofiler_v1	D7S820	Y	6	7			TRUE	FALSE	0	-2	-2	0	0	0	0	-2	0	0.88			
16	Profiler_Pl	D3S1358	B	14	15			FALSE	FALSE	0	0	0	0	0	0	0	0	0	0	0.5		

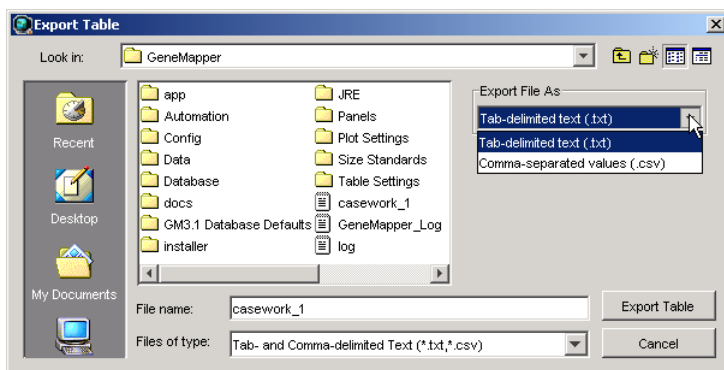
Figure 9-1 Example of an exported table with PQVs displayed in an Excel spreadsheet

It is recommended that you create a separate table setting to use just for exporting if you do not want to have all of these PQVs within the exported table. Or, you can manipulate the table once it is imported into a spreadsheet to remove these columns.

To export a table:

1. Click the tab of the table you want to export (Samples or Genotypes).
2. Select **File > Export Table**.

3. Complete the Export Table dialog box.



- Navigate to the location you wish to export the file.
- Select to export the file as Tab-delimited text (.txt) or Comma-separated values (.csv) from the Export File as drop-down list.
- Type in the desired File Name. The default file name is the name of the project.
- Click **Export Table**.

Opening Exported Samples or Project Tables in Microsoft Excel

Note: You need to adjust column widths in Microsoft Excel, even if you adjusted them appropriately in the GeneMapper *ID* software.

If you exported the files as a Tab-delimited text (.txt) file

- Find the exported table file on your hard drive.
- Right-click on the file and select **Open With > Choose Program**.
- From the list of applications, choose Microsoft Excel for Windows. The table opens within a spreadsheet.

If you exported the files as a Comma-separated values (.csv):

- Find the exported table file on your hard drive.
- Double-click to open the file. The table opens within a spreadsheet.

About CODIS

GeneMapper *ID* Software Features GeneMapper *ID* software can export data from the analysis in a format suitable for the FBI Laboratory Combined DNA Index System (CODIS). For more information about CODIS, see:

<http://www.fbi.gov/hq/lab/codis/index1.htm>

CODIS Requirements Creation of CODIS CMF files from GeneMapper *ID* software requires that:

- A specimen category is selected for each sample to be exported.
- Samples designated as a positive control, negative control, or allelic ladder cannot be exported.
- Allele calls contain acceptable characters: numbers, decimal points, letters X and Y for Amelogenin allele calls, and the symbols less than (<) and greater than (>).

CODIS Export Manager

The CODIS Export Manager is used to view and set values for three fields required for exporting samples to CODIS:

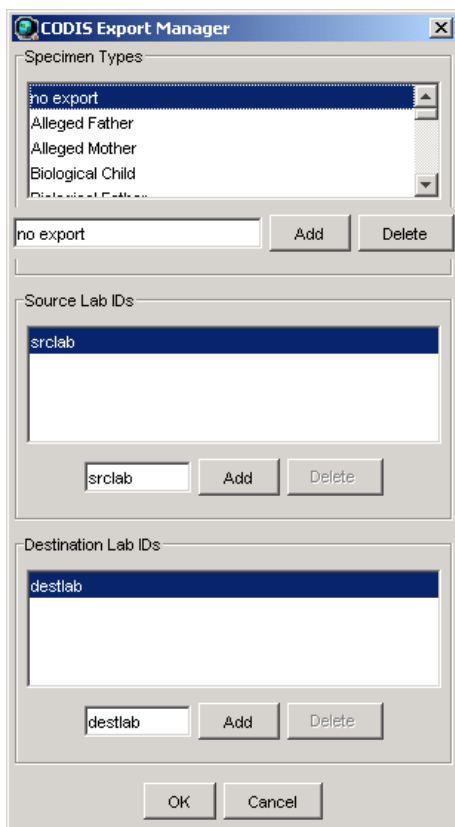
- Specimen Types
- Source Lab ID
- Destination Lab ID

Setting CODIS Export Fields

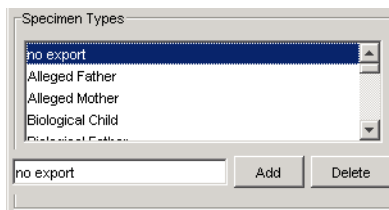
CODIS laboratories should use their assigned Source Lab ID and Destination Lab ID.

To set CODIS export fields:

1. Select **Tools > CODIS Export Manager**.



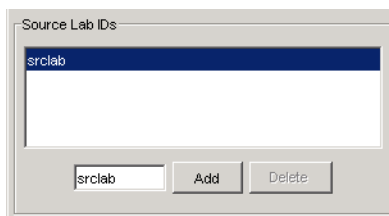
2. View the Specimen Types.



The specimen types included in GeneMapper *ID* Software version 3.1 are currently accepted by CODIS.

When CODIS accepts a new specimen type, you can add the specimen type to the software by typing it in the text box and then clicking **Add**.

3. Add a Source Lab ID.

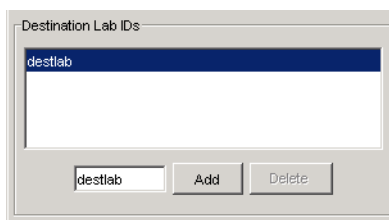


- a. Enter your assigned source lab ID in the text box.

Note: The source lab ID cannot exceed nine characters.

- b. Click **Add**.

4. Add a Destination Lab ID.



- a. Enter your assigned destination lab ID in the text box.

Note: The destination lab ID cannot exceed nine characters.

- b. Click **Add**.

5. Click **OK** to save the changes and close the CODIS Export Manager.

CODIS Table Export

CODIS Specimen Number

The CODIS Specimen Number (up to 24 characters) is accessed from:

- The Sample Name field, if the UD1 (User Defined 1) column is not used
- The UD1 column, if the UD1 column is used

Assigning specimen Types

Before you can export a table in the CODIS format, a specimen type must be assigned to all samples.

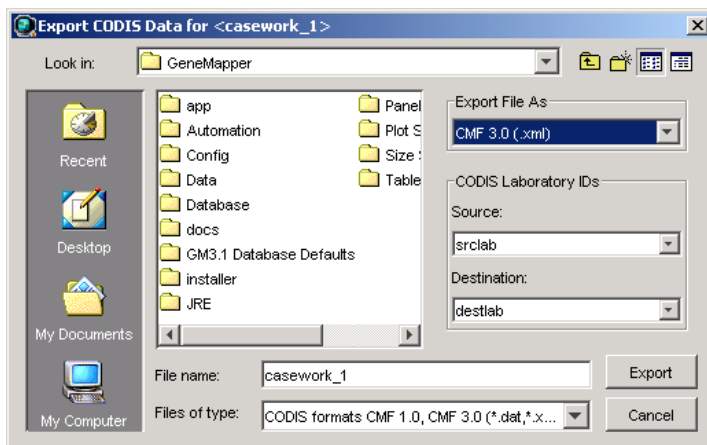
To assign specimen types:

1. Open the project.
2. For each row, select the appropriate setting in the Specimen Category column:
 - a. Use the default setting of **no export** for allelic ladders and controls.

Note: Exporting sample types designated as allelic ladders or controls generates an error message.
 - b. Select the appropriate specimen type for each sample.
3. Save the project.

To export the CODIS table:

1. Select **File > Export Table for CODIS**.



2. Complete the Export CODIS dialog box:
 - a. Navigate to the location you wish to export the file.
 - b. In the Export File As section, select **CMF 3.0 (.xml)** from the drop-down list.
 - Note:** GeneMapper *ID* software supports CMF 1.0 (.dat) file types for export.
 - c. In the CODIS Laboratory IDs section, select a source lab from the Source drop-down list and a destination lab from the Destination drop-down list.
 - d. Type in the desired File Name. The default file name is the name of the project.
 - e. Click **Export**.

This chapter covers:

Printing Data	10-2
Page Set Up Options	10-4
Printing Sizing Plots Similar to Genescan Using the Predefined AmpF Φ STR Sizing Plot Setting	10-7
Printing Genotype Plots Similar to Genotyper Using the Predefined AmpF Φ STR Genotyping Setting	10-8

Printing Data

Printable Views In the GeneMapper™ ID software you can print the following views:

- Samples table
- Genotypes table
- Samples plots
- Genotypes plots
- Raw data
- Sample info
- EPT data

To print one of the views listed above, display the view on your screen, then select **File > Print**. For example, if you want to print the raw data of a particular sample, select the sample in the Samples tab, then select **View > Raw Data** to display the raw data. Then select **File > Print**.

Note: You cannot print any displays from within the Panel Manager. Hard copies from the Panel Manager can be obtained by taking a screen shot of the desired view, then printing the screen-shot.

Printing Considerations

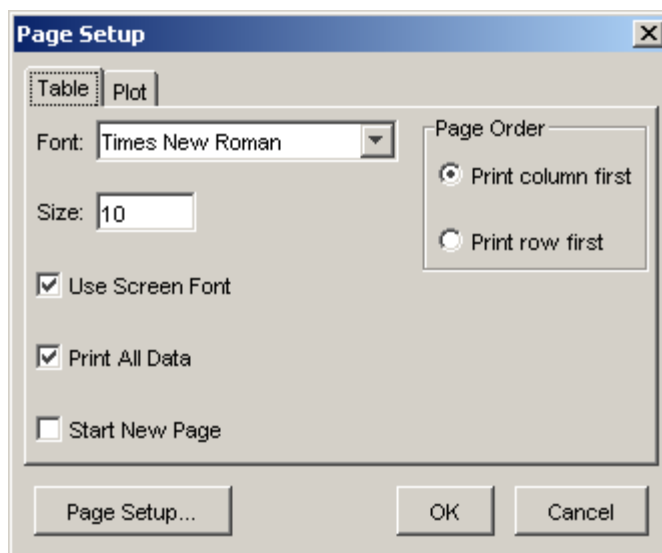
Things to consider when printing:

- The more information displayed in a printout, the slower the page(s) print. To maximize printing speed, configure your displays with the minimal amount of data needed. For example, the printing speed is faster if you label your peaks with one label each, instead of four labels each.
- If you do not need printouts to contain as much information as when you are editing and interpreting the data, you can create plot settings specifically for printing within the GeneMapper Manager.
- The printout resembles the display on the screen. If you zoom, the printout shows the zoomed in area. However, if you are printing the sample or genotypes table, and you have adjusted the column widths, the adjusted widths are not displayed in the printouts.
- When printing the samples or genotypes tables, usually all columns do not fit on one page. Refer [“Page Set Up Options” on page 10-4](#) for more information.

- You can export these tables and adjust the column widths within a spreadsheet application such as Microsoft Excel to fit on one page.
- The speed of printing depends on the memory specifications of your printer.
- In GeneMapper *ID* software, various print performance tests were conducted. These tests measured the total time (mm:ss) needed to print a one-page document under various plot configurations. Print time was measured by time taken to spool document plus time for spool to leave printer queue. Time did not include the time taken for the printed page to exit the printer. Differences in time can result if the document is printed on a slower CPU (1000 MHz) versus a faster CPU (2386 MHz) and if the GeneMapper *ID* CPU had less RAM. The printer used for comparisons in-house was an HP 4500.
- Applied Biosystems cannot test all available printers on the market, and therefore, cannot guarantee printing will be successful with each and every printer available.

Page Set Up Options

Table Tab



Options

- Page Order – When tables are printed, the number of columns and rows can exceed the limit so that they cannot be printed on one page.

Example:

A table has 50 columns and 10 rows. A page can accommodate 10 columns only, so five pages are required to print 50 columns horizontally. Similarly, if the number of rows are more than what can be accommodated on a page then the printing goes beyond a page (vertically). Now if the table has both the situations, meaning number of columns are 50 (5 pages horizontally) and number of rows are 1000 (10 pages vertically), the printing can be done in 2 ways (total 50 pages). Either the pages can be printed horizontally first (5 pages with all columns and 100 rows) or the pages can be printed vertically first (10 pages with all rows and 10 columns).

- **Prints Columns First** – This option completes printing all columns for each page of samples before moving on the next page of samples (see order below):

	Columns 1-5	Columns 6-10	Columns 11-15
Samples 1-20	Page 1	Page 2	Page 3
Samples 21-40	Page 4	Page 5	Page 6

- **Print Rows First** – This option completes printing all Samples (rows) first for each page before moving on to the columns (see order below):

	Columns 1-5	Columns 6-10	Columns 11-15
Samples 1-20	Page 1	Page 3	Page 5
Samples 21-40	Page 2	Page 4	Page 6

- **Use Screen Font** – The users can change the fonts of system via the operating system. If the fonts are changed and this option is selected, during printing system fonts and not application fonts are used.
- **Print All Data** – There is special handling for large data in a column. For example, if the sample file name is too big, ellipses are introduced in the middle of the name, then the length of the name is decreased from the middle to a certain length. This way, the data can still be printed on one page. When this option is selected, the whole name is printed and there is no special handling to introduce ellipses and truncate data.

- **Start New Page** – When this option is selected, if the table is to be printed after plots, the table prints on a new page. If not selected, then table prints immediately after plots are printed (starting on same page as plots, if space left) and may not fit on that page.

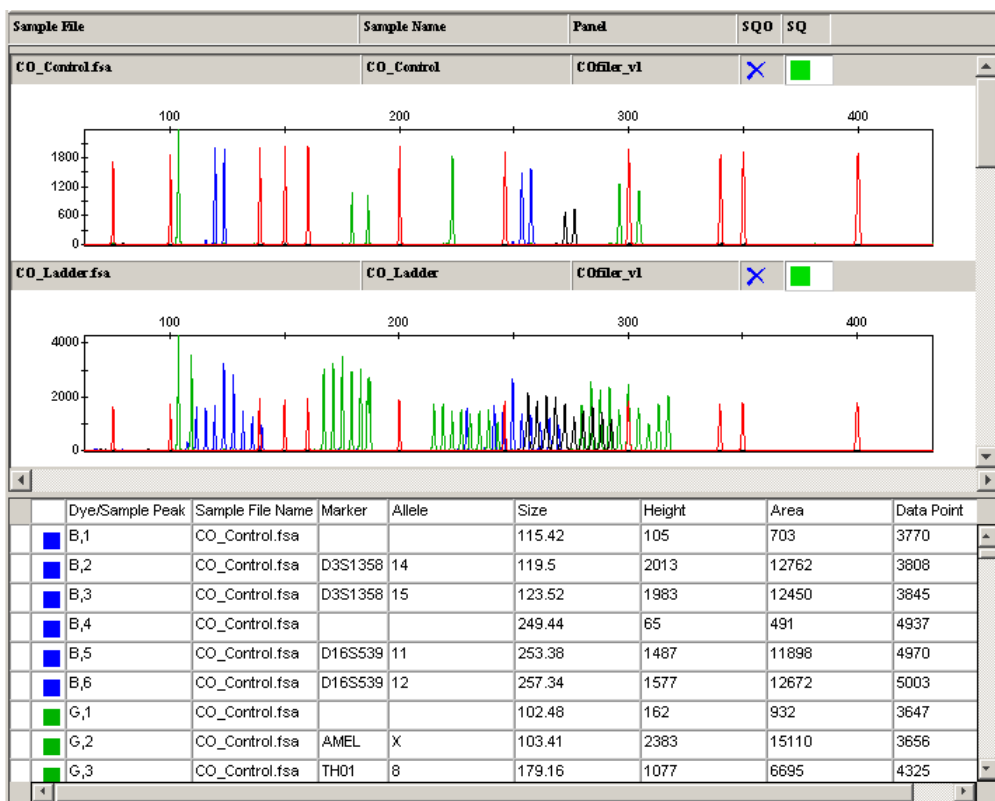
Plot Tab

Options

- **Honor plots per pane** – When this option is selected, the number of plots selected to be displayed per pane (1 - 8) prints per page.
- **Small** – When this option is selected, the number of plots per pane selected in user interface is ignored and a preset size of the plot (small) prints.
- **Medium** – When this option is selected, the number of plots per pane selected in user interface is ignored and a preset size of the plot (medium) prints.
- **Large** – When this option is selected, the number of plots per pane selected in user interface is ignored and a preset size of the plot (large) prints.

Printing Sizing Plots Similar to Genescan Using the Predefined AmpF $\mathcal{L}$ STR Sizing Plot Setting

Below is a representation of how the data will print when using this plot setting:

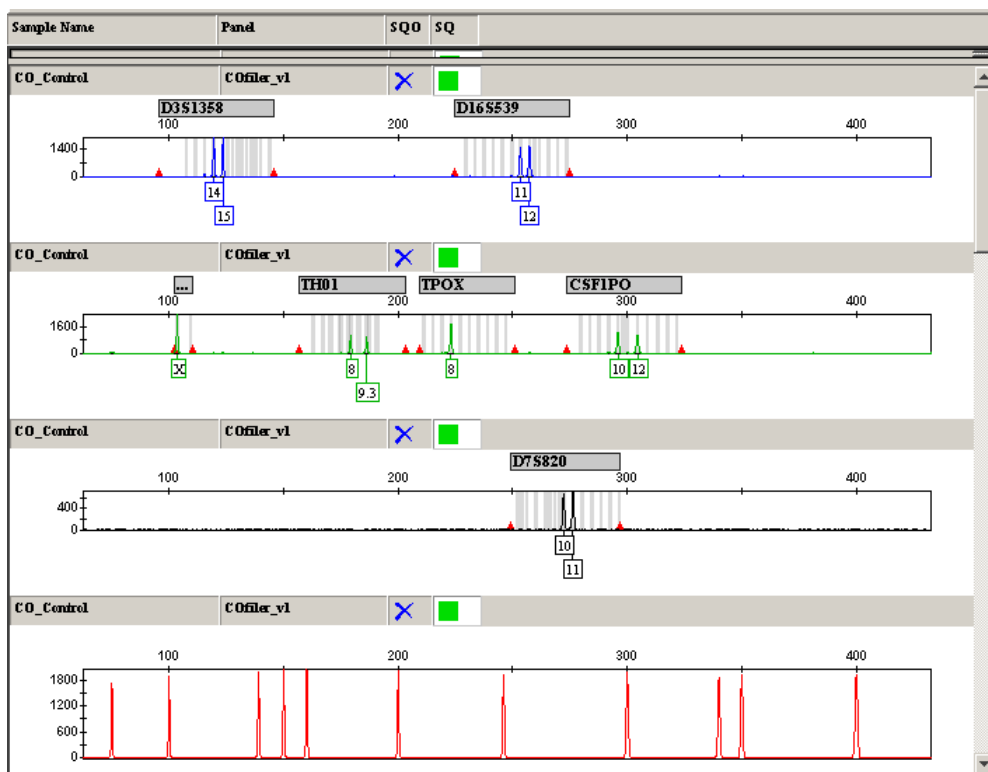


To print this view:

1. Within the samples tab of the main project window, select the samples in the table you want to display in the printouts.
2. Select **Analysis > Display Plots**.
3. Select **AmpF $\mathcal{L}$ STR Sizing** from the Plot Setting drop-down list. (Refer to Chapter 5 for information on importing plot settings into the GeneMapper Manager.)
4. Select **File > Print** (leaving all page set-up options as default).

Printing Genotype Plots Similar to Genotyper Using the Predefined AmpF ℓ STR Genotyping Setting

The allelic ladder and control samples as defined in the sample type will be printed first if multiple samples are shown. Below is a representation of how the data for one sample will be printed when using this plot setting:



To print this view:

1. Within the samples tab of the main project window, select the samples in the table you want to display in the printouts.
2. Select **Analysis > Display Plots**.

3. Select **AmpFSTR Genotyping** from the Plot Setting drop-down list. (Refer to Chapter 5 for information on importing plot settings into the GeneMapper Manager.)
4. Select **File > Print** (leaving all page set-up options as default).

Process Quality Values

This appendix describes how users can optimize the quality assurance level of their results by using the Process (component-based) Quality Values (PQV) in the GeneMapper™ ID Software Version 3.1 to troubleshoot the data analysis process.

This appendix covers:

Overview of	
Process (Component-Based) Quality Values (PQV)	A-2
Using Process (Component-Based) Quality Values	A-3

Overview of Process (Component-Based) Quality Values (PQV)

What are Process Quality Values

Process Quality Values (PQVs), values reported by data analysis, aid in finding and fixing problems in sample preparation and analysis. These values are the end results reported by the PQV system.

The PQV Process

The PQV process includes the following:

- At the end of the process, quality values (also known as PQV or Process Component-Based Quality Values) are assigned to the size calling process and the allele calling process.
- On the Samples page and Genotypes page, the columns presenting PQV result data, other than SQ and GQ results, display the following samples after analysis:
 - Pass  (green square) symbol when no problem exists,
 - Check  (yellow triangle) symbol when there are problematic components such as missing size standards, or missing matrices
 - Low Quality  (red octagon) symbol when the result falls below the defined threshold

What are PQVs

PQVs reported by data analysis are an aid to finding and fixing problems in sample preparation and analysis. These values are the end results reported by the PQV system.

Editing PQVs

When you edit PQVs you must create an Analysis Setting and select the application mode to control which PQVs are used and available to you for editing.

Using Process (Component-Based) Quality Values

The PQV System Quality values reported by the GeneMapper *ID* software are an aid to finding and fixing problems in sample preparation and analysis. These values are the end results reported by the PQV system. For more information on the PQV system, see [“Unique Features of GeneMapper ID Software v3.1” on page 1-4](#).

The color-coded, sortable PQV values on the Samples and Genotypes pages allow you to isolate problem samples quickly and examine the electropherograms.

Two types of PQV values are presented on the Samples and Genotypes pages.

On the Samples page:

- The SFNF, MNF, SNF, and OS parameters report results as Pass  or Check .
- The second type, the Sizing Quality (SQ) parameter, reports results as Pass , Check , or Low Quality .

On the Genotypes page:

- The SHP, AN, AE, OBA, SPA, Bin, PHR, LPH, SPU, BD, SP, OS, CC, OVL, NB, and DP parameters report results as Pass  or Check .
- The second type, the Genotype Quality (GQ) parameter, reports results as Pass , Check , or Low Quality .

Note: On the Genotypes page, the ADO and AE parameters are also PQV parameters, but they report results as checks  instead of colored flags.

A red octagon for the Sample SQ indicates that the sizing standard is of low quality. A red octagon for the Sample GQ parameter means the analysis of that sample is of low quality. You can set the threshold for the GQ PQV values in the Analysis Method Editor on the Quality Flags page.

PQV Values for Specific Applications

The PQV values are used for specific analyses. The following table shows which values are used for each of the following analysis.

HID = Human Identification, and
SNP = SNP Genotyping (SNaPshot® Kit)
Microsatellite

✓ = Used with this analysis
No check mark = Not applicable.

Analysis					ADO	AE	SHP	AN	OBA	SPA	BIN	PHR	LPH	SPU	BD	SP	OS	CC	OVL	NB	DP	SFNF	MNF	SSNF	SQ
HID					✓	✓		✓			✓	✓	✓	✓	✓		✓	✓	✓	✓		✓	✓	✓	✓
SNP					✓	✓		✓				✓	✓	✓	✓		✓	✓	✓		✓	✓	✓	✓	✓
Microsatellite (di-nucleotide)					✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓				✓	✓	✓
Microsatellite (other)					✓	✓		✓			✓	✓	✓	✓	✓		✓	✓	✓			✓	✓	✓	✓

**Not for ladder files

Rules for PQV Columns

In gaining familiarity with the PQV columns on the Samples page and Genotypes page, keep the following rules in mind:

- Quality metrics with Pass/Check values and no Low Quality value are warning flags. Analysis does not stop if problems are detected with these properties, but you should examine results flagged with Check values.
- Holding the cursor over a column header displays a ToolTip identifying the full name of the column (the default names are often acronyms).
- PQV results are reported in the column labeled SQ on the Samples page and GQ on the Genotypes page.
- The SQ and GQ quality indicators (displayed in either symbols or in numerical values depending upon the Quality Metrics Display setting in the Tools-Options Analysis tab) have the meanings listed below:
 - Green = good data (default: numerical range of 0.75 to 1.0)
 - Yellow = questionable data (default: numerical range of 0.25 to 0.75)
 - Red = low quality data (default: numerical range of 0.0 to 0.25)

PQV Flags on the Samples Page

The PQV flags on the Samples page.

PQV Column	Description
SFNF	Sample File Not Found • A flag (Pass  or Check ) is automatically displayed when no sample file is found. • Not editable.
MNF	Matrix Not Found • A flag (Pass  or Check ) is automatically displayed when no matrix file is found attached to the sample file. • Not editable.
SNF	Size Standard Not Found • A flag (Pass  or Check ) is automatically displayed when no size standard is found. • Not editable.

PQV Column	Description
OS	Offscale - A flag (Pass  or Check ) is automatically displayed when there is offscale data in the signal. - Not editable. - Indicates an offscale result in the size standard region of the signal.
SQ	Sizing Quality - A value (Pass , Check , or Low Quality ) is automatically displayed to indicate the sizing process quality. - The sizing quality is calculated based on the similarity between the size standard fragment pattern and the actual size standard peak distribution pattern in the sample. - Not editable. - This feature performs the following functions: - Identifies and eliminates the primer peaks based on peak shape - Performs size matching (ratio matching) - Makes a size calling curve using the Local Southern Method

PQV Flags on the Genotypes Page

The PQV flags on the Genotypes page:

Column	Description
AE	Allele Edit (check box) - The check box displays unchecked immediately after analysis. - The check box is checked when the marker allele calls have been edited by you. Note: The marker allele calls are edited in the Plot View page. The user can enter comments in the Allele History Comments column on the same page.

The PQV flags on the Genotypes page: *(continued)*

Column	Description
ADO	Allele Display Overflow (check box) - The check box is checked when the number of alleles the marker calls exceeds the number to display previously set by you. - The user specifies how many alleles to display in the Table Settings Editor parameters. The default is two. - There are six columns for each allele to indicate name, size, height, area, mutation and comments.
SHP	Sharp Peak - A flag (Pass  or Check ) is automatically displayed when there is a sharp peak present in the marker signal. - Analysis does not stop if problems are detected with these properties, but you should examine results flagged with Check values. - The PQV system gives a label of SHP to indicate a cluster of peaks with a large, narrow peak in the middle whose width is 50% less than the neighboring peak. - This flag is used for Microsatellite (dinucleotide only) analysis. - Not editable.
AN	Allele Number - A flag (Pass  or Check ) is automatically displayed when the number of alleles exceeds the maximum legal number for the organism, or no alleles are found. That number is specified in the analysis method. - Triggering this flag reduces the final PQV Genotype value (GQ parameter) to zero ("0" multiplier). - Not editable.
OBA	One Basepair Allele - A flag (Pass  or Check ) is automatically displayed when there is a one-base pair allele (a microvariant peak) present in the marker signal. - Two allele peaks that are one base pair apart are flagged. This may indicate the presence of a microvariant and/or an invalid allele call. - This flag is used only for Microsatellite (dinucleotide markers only). - Not editable.

The PQV flags on the Genotypes page: *(continued)*

Column	Description
SPA	Single Peak Artifact - A flag (Pass  or Check ) is automatically displayed when the marker signal contains single peaks due to some problem in electrophoresis. - The flag is triggered when there are no peaks present within a two-base pair range prior to an allele peak. - This feature detects the absence of stutter peaks, which indicates nonmicrosatellite peaks. - This flag is used only for microsatellite (dinucleotide markers only). - Not editable.
Bin	Out of bin allele - A flag (Pass  or Check ) is automatically displayed when the called alleles' peak apex is out of the bin boundary. - Triggering this flag reduces the final PQV Genotype value. The default is 20% (0.2 multiplier) of what it would be otherwise. - For HID, this is labeled OL (off ladder alleles). - This flag is used for microsatellite analysis. - Not editable.
PHR	Peak Height Ratio - A flag (Pass  or Check ) is automatically displayed when there are two alleles present and the ratio between the lower allele height and the higher allele height is below a certain level. - If there are more than two alleles present, the calculation iterates through all the peak pairs. - PHR can be set in the Peak Quality tab of the analysis method (Analysis Manager). The default is 50%. - For microsatellite markers, the ratio is calculated based on peak heights of the called allele peaks. - For SNP, the ratios are calculated the same as for microsatellite markers except it goes across two different colors and only two peaks are used in the calculation. - Not editable.

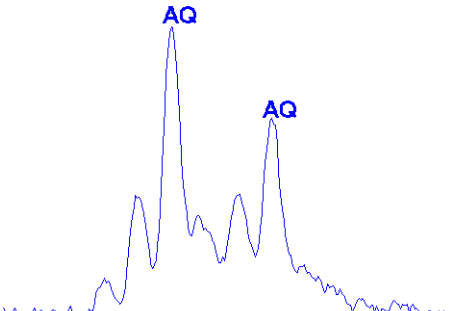
The PQV flags on the Genotypes page: *(continued)*

Column	Description
LPH	Low Peak Height - A flag (Pass  or Check ) is automatically displayed when the alleles are lower than the specified values and do not result in the proper intensity. - The homozygous (default is 200) and heterozygous (default is 100) values can be set in the Peak Quality tab of the Analysis method (Analysis Manager). - Setting this flag reduces the final PQV Genotype value (GQ parameter). The default is 50% (0.5 multiplier) of what it would be otherwise. - Not editable.
SPU	Spectral Pull-Up - A flag (Pass  or Check ) is automatically displayed when the marker signal contains bleed-through peaks (pull-up peaks). - Pull-up is when the peak height of the called allele peak is less than X% of the larger peak within ± 1 data point. - The default is a conservative 10% to check all colors. The user can set this value. - This flag is used for HID, SNP, and Microsatellite. - Not editable.
BD	Broad Peak - A flag (Pass  or Check ) is automatically displayed when the called alleles' peak width is wider than a certain value. The default of this value is 1.5 basepair. - This flag is used for HID, SNP, and Microsatellite (all markers). - Not editable.
SP	Split Peak - A flag (Pass  or Check ) is automatically displayed when a split peak is found by the GeneMapper <i>ID</i> software. - A split peak is defined as overlapping peaks that are < .25 basepairs apart (the horizontal distance from two peak apexes). - Not editable.

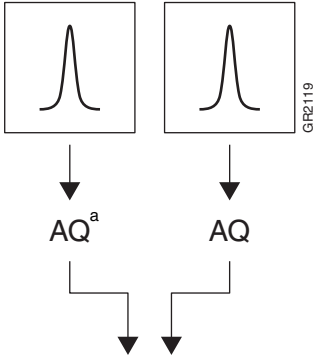
The PQV flags on the Genotypes page: *(continued)*

Column	Description
OS	Offscale - A flag (Pass  or Check ) is automatically displayed when there are offscale peaks present within the marker size range. - This flag is used for all applications. - Not editable.
CC	Control Concordance - A flag (Pass  or Check ) is automatically displayed when the designated control sample does not exactly match the defined alleles for this marker in the panel being scored. - This feature serves as an internal control for quality assurance and allows you to see deviations in your controls. - This flag is used for all applications. - We recommend that you run the control sample at least once for every panel. - Not editable.
OVL	Overlap - A flag (Pass  or Check ) is automatically displayed when a peak in the overlapped region is called twice. (It is possible to have two allele size ranges that overlap.) - This feature serves as a warning for an allele calling error. - This is used for HID only.
NB	Narrow Bin - A flag (Pass  or Check ) is automatically displayed when a peak is present at a position within 0.5 basepairs from a bin and no peak is present inside that bin. - The benefit is to capture peaks that fall outside of bin boundaries due to experimental variations. - This flag is used for SNP only.

The PQV flags on the Genotypes page: *(continued)*

Column	Description
DP	Double Peak - A flag (Pass ■ or Check ▲) is automatically displayed when two peaks of the same color in the same bin have a ratio ≥ 0.5. (minor peak height/major peak height) - This flag is used for SNP only. - Not editable.
GQ	Genotype Quality - A red octagon ⬡ is shown when the analysis for a sample is of low quality. - Pass ■, Check ▲, or Low Quality ⬡ flags are set by the Analysis Method. Not editable. Below is a representation of sample peaks with the allele quality (AQ) assigned.  See the following page for information about the mathematical formula used to calculate the Genotype Quality from a sample represented by the image above.

The PQV flags on the Genotypes page: *(continued)*

Column	Description
GQ (continued)	Genotype Quality - Mathematical Formula - The mathematical formula used is:  $\bar{x} (AQ) = MQ$ $GQ = MQ^b \times (PQV_1 \times PQV_2 \times \dots \times PQV_N)$ a AQ (Allele Quality) is a function of quality value assignments for the following: sizing quality, allele calling quality, bin assignment quality, and bin quality. b MQ (Marker Quality) is modified by user defined PQVs to generate the final GQ value. - PQVs are weighted from 0 to 1. - The actual value of a PQV in the equation is: 1 minus the weight 0 (weight) = no effect on the final GQ calculation (1 minus 0 = 1, therefore no change to GQ value) 1 (weight) = full effect (1 minus 1 = 0) (If you multiply GQ by 0 and you get a GQ value of 0, then the sample analysis is of low quality.) - Between 0 and 1, the higher the value, the greater the impact on GQ. - PQV filtering is controlled by the threshold set by GeneMapper <i>ID</i> software users, and remains fully functional irrespective of what weight is chosen.

Software Genotyping Algorithms

B

This appendix provides a description of the new genotyping algorithms used in the GeneMapper™ *ID* Software version 3.1 and a detailed description of the Peak Detection Basic, Classic, and Advanced algorithm settings.

This appendix covers:

GeneMapper <i>ID</i> Software Genotyping Algorithms	B-2
Basic Mode - Peak Detection Algorithm Settings	B-6
Classic Mode - Peak Detection Algorithm Settings	B-7
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Peak Detection: Polynomial Degree and Peak Window Size Parameters	B-17
Optimizing Peak Detection Sensitivity: Example 1	B-20
Optimizing Peak Detection Sensitivity: Example 2	B-22
Optimizing Peak Detection Sensitivity: Example 3	B-24
Peak Detection: Slope Threshold for Peak Start and Slope Threshold for Peak End Parameters	B-25
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Size Calling Methods for Classic and Advanced Modes	B-29

GeneMapper *ID* Software Genotyping Algorithms

Overview Five algorithms used in the GeneMapper *ID* software are discussed in this appendix:

- **Peak Detection** – uses Basic, Advanced, or Classic mode to detect peaks and process data
- **Size-matching/calling** – matches found peaks to size standards
- **Binning** – determines bin centers for genotyping
- **Allele calling** – produces a consensus call based on several allele-calling algorithms
- **Quality value determination** – assigns a quality value to size- and allele-calling algorithms (See [Appendix A, “Process Quality Values,”](#) for more information.)

Peak Detection Algorithm This algorithm uses Basic, Advanced, or Classic mode to detect peaks and process data. (See [“Basic Mode - Peak Detection Algorithm Settings”](#) on page B-6, [“Classic Mode - Peak Detection Algorithm Settings”](#) on page B-7, and [“Advanced Mode - Peak Detection Algorithm Settings”](#) on page B-12.)

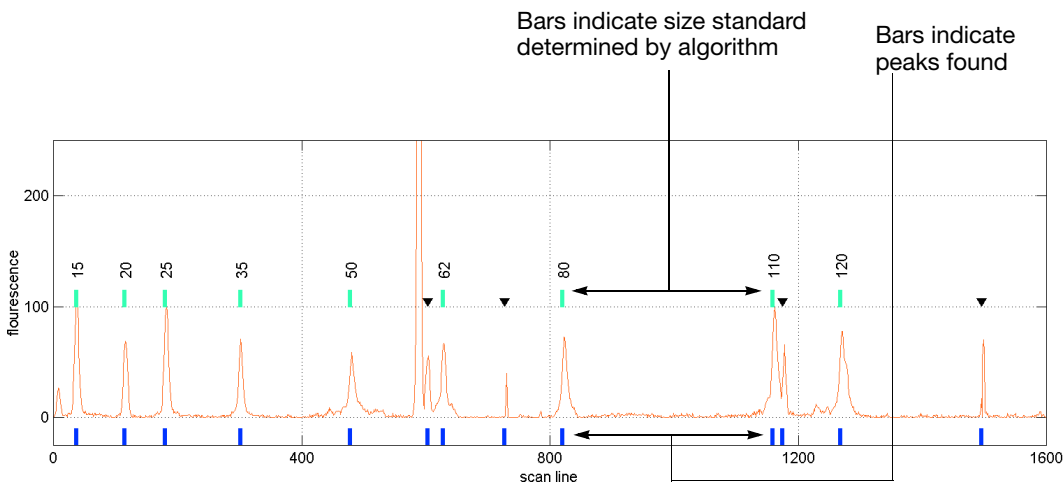
Size-Matching Size-Calling Algorithm This algorithm uses a dynamic programming approach that is efficient (runs in low polynomial time and space) and guarantees an optimal solution. It first matches a list of peaks from the electropherogram to a list of fragment sizes from the size standard. It then statistically derives quality values determined by examining the similarity between the theoretical and actual distance between the fragments.

Size-Matching Algorithm Example

An example of how the size-matching/calling algorithm works is shown below using a contaminated GeneScan™ 120 size standard data.

Peaks found (standard and contamination) are indicated by blue lower bars along the x-axis. The size standard fragments as determined by the algorithm (and their corresponding lengths in base pairs) are designated by the upper green bars. Note that there are more peaks than size standard locations because the standard was purposely contaminated to test the algorithm. The algorithm correctly

identifies all of the size standard peaks and removes the contamination peaks (denoted by the black triangles) from consideration. The large peak is excluded from the candidate list by a filter that has identified it as being atypical with respect to the others.



Binning Algorithm

This algorithm estimates the bin centers using all found alleles. To determine true bin positions, the algorithm uses an iterative clustering technique and then reports a quality value proportional to the binning noise. To enhance flexibility, bins may also be edited manually.

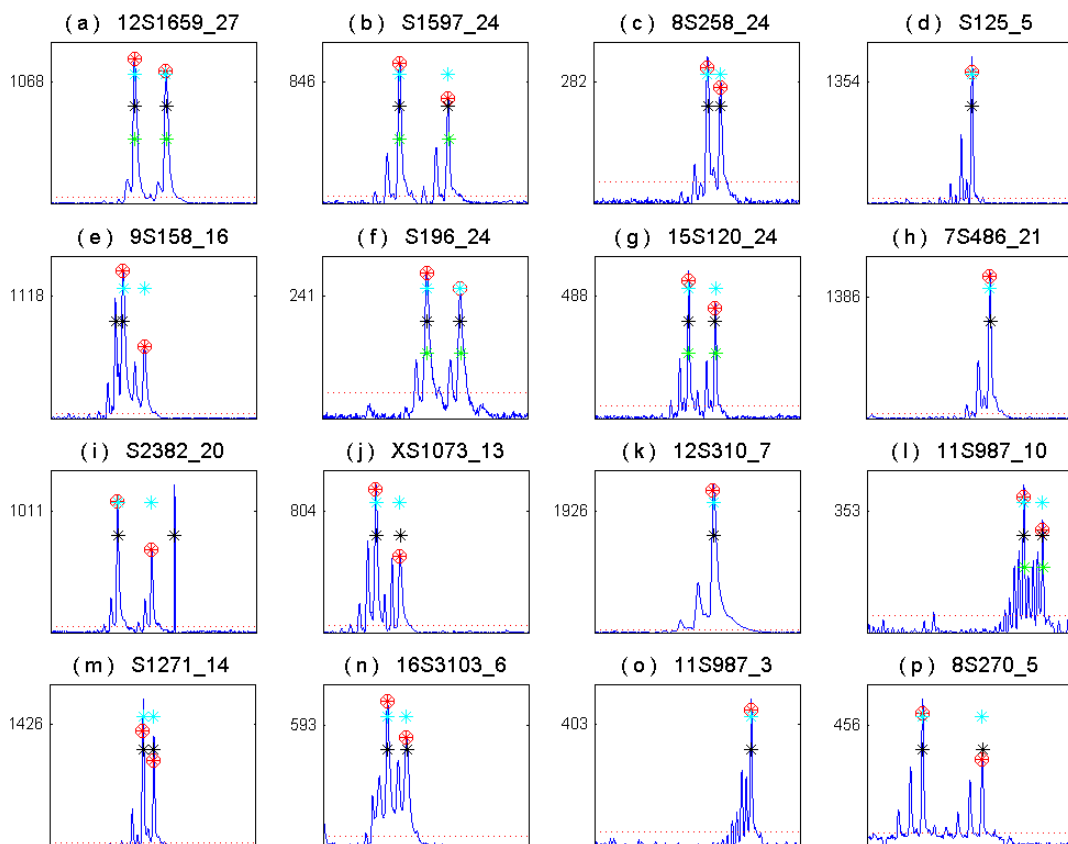
Note: When you create bins manually, all alleles, regardless of their quality values, are treated as reference alleles as long as the alleles reside within the created bins' boundaries.

Allele-Calling Algorithm

Final allele calls are based on a consensus between several different allele-calling algorithms. Each caller has a different design philosophy such that it excels in a particular data regime. A variety of allele-calling algorithms are used. Allele-calling algorithms involve envelope detection, optimization of parametric models, and rule-based systems.

Example Output of Different Allele-Calling Algorithms

The following is an example of three different allele-calling algorithms for 16 samples. User annotations are denoted by (red) circles, allele caller outputs are denoted by (green, black, and blue) asterisks. Note that consensus between multiple callers virtually assures that the calls are correct. In certain cases some algorithms have not made a call since they have determined that the data is too complex to act on, as in (i) and (p). Here the blue asterisks show the calls transmitted to you. Low quality values are reported because in both cases the first algorithm did not call and in (i) the black caller is not in agreement with the blue. However, despite these conditions, the calls are correct. The low quality values alert you to potential problems such as the spurious peak in (i) and the high background in (p).



Quality Value Determination Algorithm

The Quality Value Determination algorithm is used to create Process Quality Values (PQVs). PQVs are reported by data analysis and are an aid to finding and fixing problems in sample preparation and analysis. These values are the end results reported by the PQV system

The quality value determination algorithm equation is the following:

$$MQ^1 = \bar{x} \text{ (AQ}^2 \text{ GQ} = MQ \times (PQV_1 \times PQV_2 \times \dots \times PQV_N)$$

- PQVs are weighted from 0 to 1.
- The actual value of a PQV in the equation is: 1 minus the weight
- 0 (weight) = no effect on the final GQ calculation (1 minus 0 = 1, therefore no change to GQ value)
- 1 (weight) = full effect (1 minus 1 = 0) (If you multiply GQ by 0 and you get a GQ value of 0, then the sample analysis is of low quality.)
- Between 0 and 1, the higher the value, the greater the impact on GQ.

PQV filtering is controlled by the threshold set by GeneMapper ID software users, and remains fully functional irrespective of what weight is chosen.

- 1.MQ (Marker Quality) is modified by user defined PQVs to generate the final GQ value.
- 2.AQ (Allele Quality) is a function of quality value assignments for the following: sizing quality, allele calling quality, bin assignment quality, and bin quality.

Basic Mode - Peak Detection Algorithm Settings

Overview The Basic peak detection algorithm is used as the default algorithm by GeneMapper *ID* software v3.1. Basic Mode uses the Local Southern size calling method which determines the sizes of fragments by using the reciprocal relationship between fragment length and mobility.

In Basic mode there are two analysis parameter options.

- Minimum Peak Height - Automatic
- Minimum Peak Height - User specified (rfu)

Minimum Peak Height

The Minimum Peak Height group box provides two parameter options:

Item	Description
Automatic option button	This option button sets the level automatically for the five dye colors (blue, green, yellow, red, and orange). This level represents the minimum signal strength that will be identified as a peak for each dye (equivalent to 10 times the noise).
User specified (rfu) option button	This option button enables the number entry fields for the five dye colors (blue, green, yellow, red, and orange). These numbers are the minimum signal strength that will be identified as a peak in relative fluorescent units (rfu). For all dyes, the default is 50, the minimum is 1, and the maximum is any number of 10 digits.

Classic Mode - Peak Detection Algorithm Settings

Overview In Classic mode there are five analysis parameter options. They are as follows:

- Ranges
- Data Processing
- Size Calling Method
- Peak Detection
- Split Peak Correction\

Ranges Parameter Options

The following are the Ranges parameter options:

Item	Description
Analysis drop-down menu: Full Range Partial Range	Used to analyze all the data collected on the genetic analysis instrument for each sample. Enter Start and Stop data point numbers in the entry fields in order to specify only a limited range analyzed for each sample. The data point numbers affect what is displayed in the results display. Normally, set the analysis range to start after the primer peak. Note: Sample files generated from the ABI PRISM® 377 DNA Sequencer may have already removed the primer peak by setting the data point range for gel image generation to exclude the primer peak.
Sizing drop-down menu: All Sizes Partial Sizes	Enter Start and Stop size numbers in the entry fields in order to specify only a limited range analyzed for each sample.

Data Processing Parameter Options

The Data Processing parameter options specify how the raw data is processed before peak detection and size calling.

Item	Description								
Baseline checkbox	Used to automatically adjust the baselines of all detected dye colors to the same level for a better comparison of relative signal intensity.								
Multicomponent checkbox Note: Do not check this box for multicapillary instruments. This user function was performed in Data Collection.	Used to specify that the GeneMapper <i>ID</i> software applies a predefined matrix to adjust for spectral overlap when it performs analysis. Although the dyes used to label DNA fluoresce at different wavelengths, the spectra overlap to some extent. Create a matrix file containing a mathematical matrix to correct for this overlap. For a description of matrix files and how to create them, see Section 5.5, “Creating and Evaluating a Matrix,” on page 5-53.								
Smoothing option buttons	Used to help reduce the number of false peaks detected by the GeneMapper <i>ID</i> software. You have the following options: <table border="1"> <thead> <tr> <th>Select...</th><th>To...</th></tr> </thead> <tbody> <tr> <td>None</td><td>apply no smoothing. Select this option if the data has very sharp, narrow peaks of interest.</td></tr> <tr> <td>Light</td><td>provide the best results for typical data.</td></tr> <tr> <td>Heavy</td><td>apply to data from slower runs that has very broad peaks, or to avoid the detection of sharp edges. Choosing this option might reduce peak size or eliminate narrow peaks.</td></tr> </tbody> </table>	Select...	To...	None	apply no smoothing. Select this option if the data has very sharp, narrow peaks of interest.	Light	provide the best results for typical data.	Heavy	apply to data from slower runs that has very broad peaks, or to avoid the detection of sharp edges. Choosing this option might reduce peak size or eliminate narrow peaks.
Select...	To...								
None	apply no smoothing. Select this option if the data has very sharp, narrow peaks of interest.								
Light	provide the best results for typical data.								
Heavy	apply to data from slower runs that has very broad peaks, or to avoid the detection of sharp edges. Choosing this option might reduce peak size or eliminate narrow peaks.								

Size Calling Method Parameter Options

Click a radio button to select the desired size calling method. The GeneMapper *ID* software uses these methods to determine the molecular length of an unknown fragment.

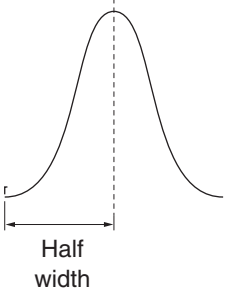
Description of the size calling method parameter options:

Item	Description
2nd Order Least Squares and 3rd Order Least Squares	Both Least Squares methods use regression analysis to build a best-fit size calling curve.
Cubic Spline Interpolation	Forces the sizing curve through all the known points of the selected GeneScan size standard.
Local Southern method	Determines the sizes of fragments by using the reciprocal relationship between fragment length and mobility. (Default method)
Global Southern method	Similar to the Least Squares method in that it compensates for standard fragments that may run anomalously.

Peak Detection Parameter Options

Use the Peak Detection parameter options to specify the minimum peak height to be detected for analysis. This, in turn, controls the number of peaks analyzed. Peaks falling below the parameters specified display in the electropherogram, but are not analyzed, and no values display for them in the tabular data.

Item	Description	For example
Peak Amplitude Thresholds	Set the dye amplitude threshold at a level that allows the software to detect peaks, but eliminate noise. For each dye, the GeneMapper <i>ID</i> software detects peaks above the threshold entered in the entry field.	If you leave the default value of 50, peaks with amplitude above 50 are analyzed and display in the tabular data. Lower amplitude peaks still display in the electropherogram, but are not analyzed and do not display in the tabular data.

Item	Description	For example
Minimum Peak Half Width 	Defines what constitutes a peak. Use to specify the smallest half peak width for peak detection. The range is 2–99. A typical number might be 3 for microsatellites, or 10 for SSCPs.	If this number is large, the software ignores noise spikes. If the peaks in the data are narrow, set the value to a low number. Experiment with this value to determine the best number for the data.

Split Peak Correction Parameter Options

Under denaturing conditions, certain fragments in the GeneScan 2500 standard display as doublets, or split peaks. This standard has labels on both strands of the DNA. Under poor denaturing conditions you see split peaks. One of the two fragments typically has normal mobility, while the other does not. The Split Peak Correction feature allows the software to correctly call each of the splits.

If you select split peak correction, you will also need to verify or change the correction limits. After you decide on a split peak correction method, use the same method for all projects to keep size calling consistent.

Note: Split Peak correction is not used with AmpF Φ STR or SNaPshot applications.

Item	Description
No Peak Correction	No correction for doublets.
GS2500 Peak Correction	Makes the following peak size assignments for GeneScan 2500: • The right peak for all fragments 222, 233, 238, 286, and 490. • The left peak for all other splits.

Item	Description
Left Most Peak Correction	Chooses the left peak for every doublet.
Right Most Peak Correction	Chooses the right peak for every doublet.
Correction Limit	Set a correction limit if correcting for doublets. Set this value slightly larger than the largest split observed. This value (set in data points) specifies the maximum width of split that should be corrected (the difference in data points of the positions of the two peaks).

Advanced Mode - Peak Detection Algorithm Settings

Overview In Advanced mode, there are four main analysis parameter options. They are as follows:

- Ranges
- Smoothing and Baselining
- Size Calling Method
- Peak Detection

Ranges Options The following are the Analysis Range options:

Item	Description
Full Range button	Use to analyze all the data collected on the genetic analysis instrument for each sample.
Partial Range (Data Points) button	Enter Start and Stop data point numbers in the entry fields in order to specify only a limited range to be analyzed for each sample. Data points affect what is displayed in the results. Normally, you set the analysis range to start after the primer peak.
Sizing drop-down menu: All Sizes Partial Sizes	Enter Start and Stop size numbers in the entry fields in order to specify only a limited range analyzed for each sample.

Smoothing and Baseline Options

The Smoothing and Baseline parameter options help to optimize peak size and eliminate noise from the baseline.

Item	Description								
Smoothing option buttons	Used to help reduce the number of false peaks detected by the GeneMapper <i>ID</i> software. You have the following options: <table> <tr> <th>Select...</th><th>To...</th></tr> <tr> <td>None</td><td> apply no smoothing. Select this option if the data has very sharp, narrow peaks of interest. </td></tr> <tr> <td>Light</td><td> provide the best results for typical data. </td></tr> <tr> <td>Heavy</td><td> apply to data from slower runs that has very broad peaks, or to avoid the detection of sharp edges. Choosing this option might reduce peak size or eliminate narrow peaks. </td></tr> </table>	Select...	To...	None	apply no smoothing. Select this option if the data has very sharp, narrow peaks of interest.	Light	provide the best results for typical data.	Heavy	apply to data from slower runs that has very broad peaks, or to avoid the detection of sharp edges. Choosing this option might reduce peak size or eliminate narrow peaks.
Select...	To...								
None	apply no smoothing. Select this option if the data has very sharp, narrow peaks of interest.								
Light	provide the best results for typical data.								
Heavy	apply to data from slower runs that has very broad peaks, or to avoid the detection of sharp edges. Choosing this option might reduce peak size or eliminate narrow peaks.								
Baseline Window text field	Used to automatically adjust the baselines of all detected dye colors to the same level for a better comparison of relative signal intensity.								

Baselining Option

The Baselining option controls the scope of the baseliner. Use this option to set the size *Beta* of the Baseline Window. GeneMapper *ID* software computes a baseline for the electropherogram of each dye independently.

How the Baselining Option Works

A baseline comprises a value at each data point i . The baseline value at each data point i is the lowest electropherogram value in a window whose width *Beta* is set using the Baselining option, and centered at each data point i .

More accurately, the baseline computed in this manner is intermediate. The real baseline value at each data point i , is the highest intermediate value, again in a window whose width $Beta$ is set using the Baselining options and centered at each data point i . GeneMapper *ID* software baselines an electropherogram by subtracting the baseline from the raw electropherogram

Troubleshooting the Baseline Window

The following table describes what happens if the baseline window is either too small or too large.

Using...	Causes...
a small baseline window size	the baseline to creep into the peaks, resulting in shorter peaks in the analyzed data.
a large baseline window size	the baseline to ride too low, resulting in elevated and possibly not baseline-resolved peaks.

Size Calling Method Options

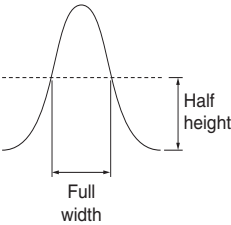
GeneMapper *ID* software uses these methods to determine the molecular length of an unknown fragment.

Description of the size calling method parameter options:

Item	Description
2nd Order Least Squares and 3rd Order Least Squares	Both Least Squares Methods use regression analysis to build a best-fit sizecalling curve.
Cubic Spline Interpolation	Forces the sizing curve through all the known points of the selected size standard.
Local Southern Method	Determines the sizes of fragments by using the reciprocal relationship between fragment length and mobility.
Global Southern Method	Similar to the Least Squares Method in that it compensates for standard fragments that may run anomalously.

Peak Detection Options

The Peak Detection options locate peaks at the positive-to-negative zero crossings of the first derivative of the baselined electropherogram. The peak detector computes the first derivative at a data point i by fitting a polynomial to a window centered on i .

Item	Description	For example
Peak Amplitude Thresholds	GeneMapper <i>ID</i> software reports to you only those peaks whose heights are at least the Peak Amplitude Threshold for that dye. Set the dye amplitude threshold at a level that allows the software to detect peaks, but eliminate noise.	If you leave the default value of 50, peaks with amplitude above 50 are analyzed and display in the tabular data. Lower amplitude peaks still display in the electropherogram, but are not analyzed and do not display in the tabular data.
Full Width Half Maximum 	Defines what constitutes a peak. Use to specify the smallest full width at half maximum for peak detection. The range is 2–99. A typical number might be 3 for microsatellites, or 10 for SSCPs.	If this number is large, the software ignores noise spikes. If the peaks in the data are narrow, set the value to a low number. Experiment with this value to determine the best number for the data.

Item	Description	For example
Polynomial Degree	Sets the degree of the polynomial.	These parameters control the sensitivity of this process. Sensitivity increases with the polynomial degree and decreases with the window size. Use polynomials of degree 2 or 3 for well-isolated peaks, such as those from a size standard, and a degree 4 for finer control. For degree 4, the Peak Window Size should be 1 to 2 times the full width at half maximum of the peaks that you wish to detect. These parameters cannot be set for each color independently.
	Min. setting	
	Max. setting	
	2	5
Peak Window Size	Sets the width of the window	
Min. setting	Max. setting	
1 above the Degree of Polynomial differentiation setting.	Number of data points between peaks	
Slope Threshold for Peak Start Slope Threshold for Peak End	Determines where a peak starts and stops	A peak ends when the first derivative again exceeds the Slope Threshold for Peak End. Slope Threshold for peak start must be non-negative and Slope Threshold for peak end must be nonpositive. Values other than 0 will move the extent of the peak toward its center.

Peak Detection: Polynomial Degree and Peak Window Size Parameters

About These Parameters

Use the Polynomial Degree and the Peak Window Size settings to adjust the sensitivity of the peak detection. You can adjust these parameters to detect a single base pair difference while minimizing the detection of shoulder effects or noise.

Sensitivity increases with larger polynomial degree values and smaller window size values. Conversely, sensitivity decreases with smaller polynomial degree values and larger window size values.

How These Parameters Work

The peak window size functions with the polynomial degree to set the sensitivity of peak detection.

The peak detector computes the first derivative of a polynomial curve fitted to the data within a window that is centered on each data point in the analysis range.

Using curves with larger polynomial degree values allows the curve to more closely approximate the signal and, therefore, the peak detector captures more peak structure in the electropherogram.

The peak window size sets the width (in data points) of the window to which the polynomial curve is fitted to data. Higher peak window size values smooth out the polynomial curve, which limits the structure being detected. Smaller window size values allow a curve to better fit the underlying data.

How to Use These Parameters

Use the table below to adjust the sensitivity of detection.

To...	Polynomial Degree Value	Window Size Value
Increase sensitivity use...	Higher	Lower
Decrease sensitivity use...	Lower	Higher

Guidelines for Using These Parameters

To detect well-isolated, baseline-resolved peaks, use polynomial degree values of 2 or 3. For finer control, use a degree value of 4 or greater.

As a guideline, set the peak window size (in data points) to be about 1 to 2 times the full width at half maximum height of the peaks that you want to detect.

Examining Peak Definitions

To examine how GeneMapper *ID* software has defined a peak, select **View > Show Peak Positions**. The peak positions, including the beginning, apex, and end of each peak, are tick-marked in the electropherogram.

Effects of Varying the Polynomial Degree

The figure below depicts peaks detected with a window size of 15 data points and a polynomial curve of degree 2 (green); 3 (red); and 4 (black). The diamonds represent a detected peak using the respective polynomial curves.

Note that the smaller trailing peak is not detected using a degree of 2 (green). As the peak detection window is applied to each data point across the displayed region, a polynomial curve of degree 2 could not be fitted to the underlying data to detect its structure.

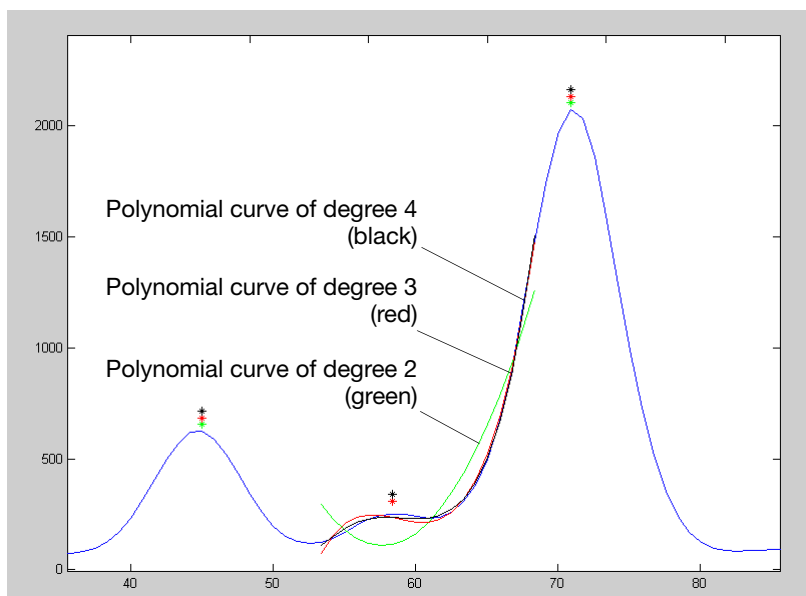


Figure B-1 Electropherogram showing peaks detected with three different polynomial degrees

Effects of Increasing the Window Size Value

In the figure below, both polynomial curves have a degree of 3 and the window size value was increased from 15 (red) to 31 (black) data points.

As the cubic polynomial is stretched to fit the data in the larger window size, the polynomial curve becomes smoother. Note that the structure of the smaller trailing peak is no longer detected as a distinct peak from the adjacent larger peak to the right.

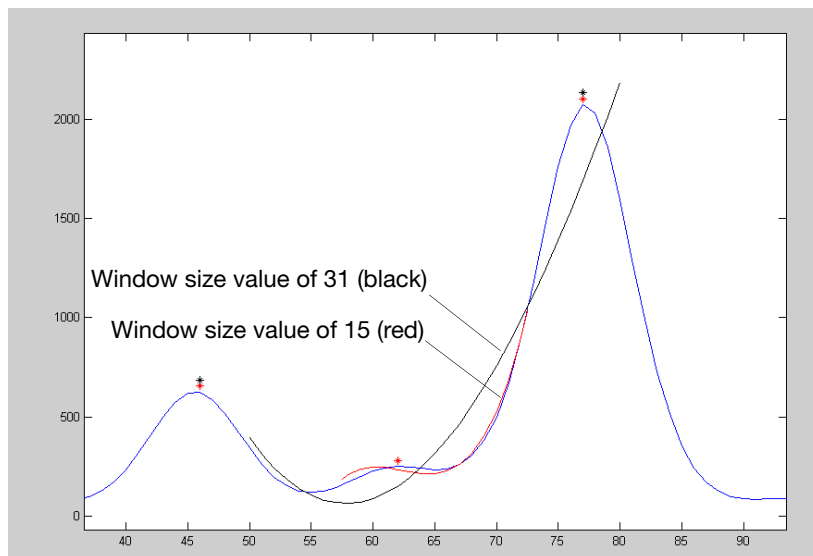


Figure B-2 Electropherogram showing the same peaks as in [Figures B-1](#) after increasing the window size value while keeping the polynomial degree the same

Optimizing Peak Detection Sensitivity: Example 1

**Initial
Electrophero-
gram**

The figure below shows two resolved alleles of known fragment lengths (that differ by one nucleotide) detected as a single peak. The analysis was performed using a polynomial degree of 3 and a peak window size of 19 data points.

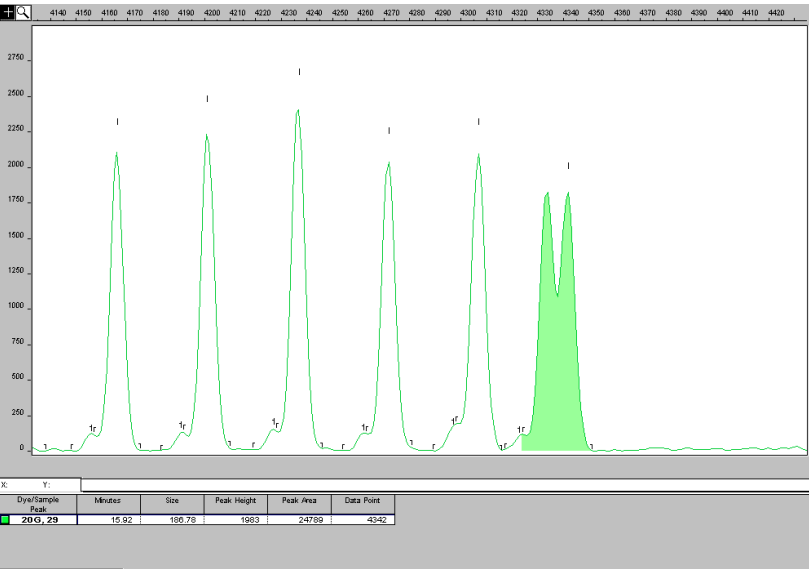


Figure B-3 Electropherogram showing two resolved alleles detected as a single peak

Note: For information on the tick marks displayed in the electropherogram see [“Examining Peak Definitions”](#) on page B-18.

Effects of Decreasing the Window Size Value

The figure below shows that both alleles are detected after re-analyzing with the polynomial degree set to 3 while decreasing the window size value to 15 (from 19) data points.

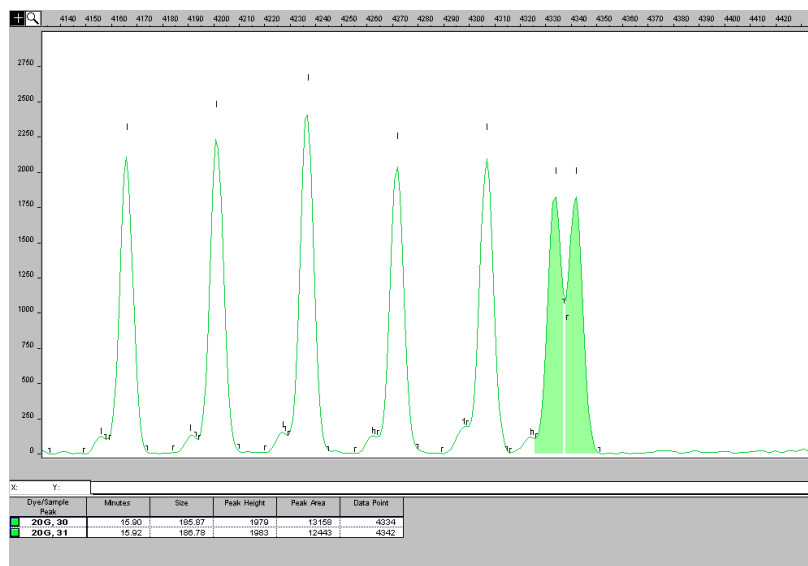


Figure B-4 Electropherogram showing the alleles detected as two peaks after decreasing the window size value

Optimizing Peak Detection Sensitivity: Example 2

Initial
Electrophero-
gram

The figure below shows an analysis performed using a polynomial degree of 3 and a peak window size of 19 data points.

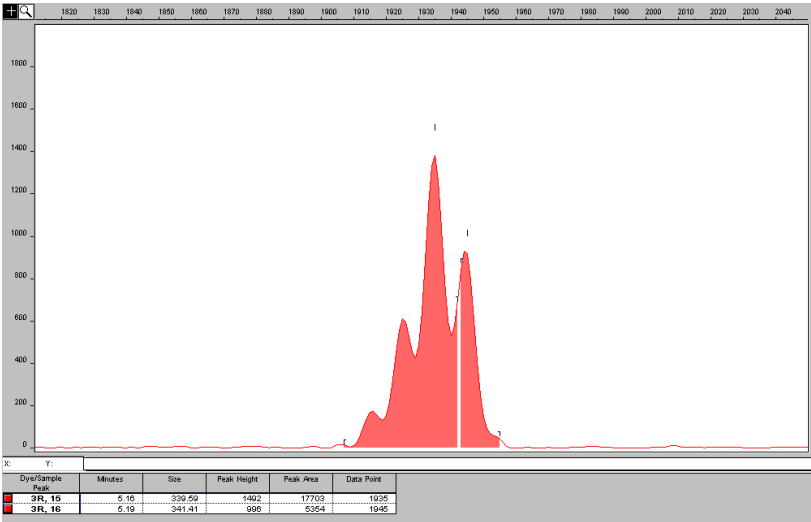


Figure B-5 Electropherogram showing four resolved peaks detected as two peaks

Effects of Reducing the Window Size Value and Increasing the Polynomial Degree Value

The figure below shows the data presented in the figure above (see [page B-22](#)) re-analyzed with a window size value of 10 and polynomial degree value of 5.

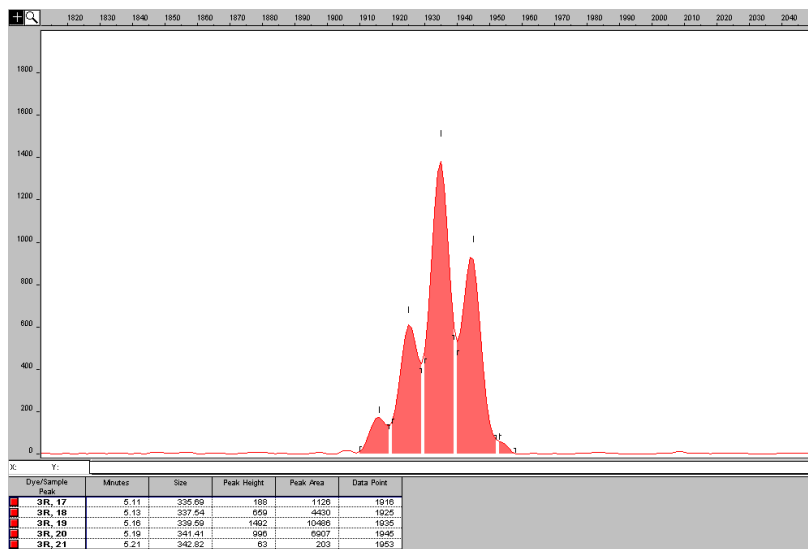


Figure B-6 Electropherogram showing all four peaks detected after reducing the window size value and increasing the polynomial degree value

Optimizing Peak Detection Sensitivity: Example 3

Effects of Extreme Settings

The figure below shows the result of an analysis using a peak window size value set to 10 and a polynomial degree set to 9. This extreme setting for peak detection led to several peaks being split and detected as two separate peaks.

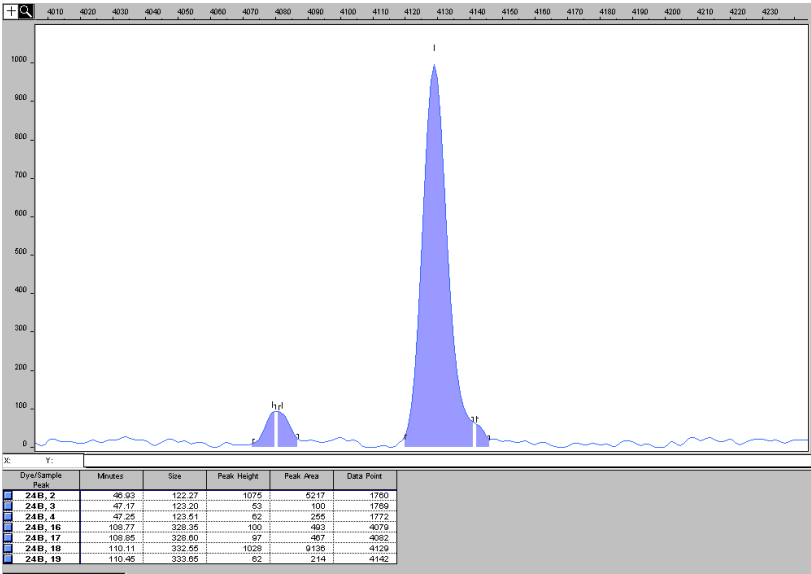


Figure B-7 Electropherogram showing the result of an analysis using extreme settings for peak detection

Peak Detection: Slope Threshold for Peak Start and Slope Threshold for Peak End Parameters

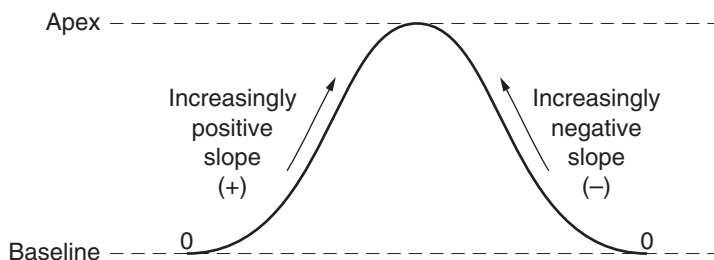
About These Parameters

Use the Slope Threshold for Peak Start and Slope Threshold for Peak End parameters to adjust the start and end points of a peak.

This parameter can be used to better position the start and end points of an asymmetrical peak, or a poorly resolved shouldering peak, to more accurately reflect the peak position and area.

How These Parameters Work

In general, from left to right, the slope of a peak increases from the baseline up to the apex. From the apex down to the baseline, the slope becomes decreasingly negative until it returns to zero at the baseline.



If either of the slope values you have entered exceeds the slope of the peak being detected, the software overrides your value and reverts to zero.

Guidelines for Using These Parameters

As a guideline, use a value of zero for typical or symmetrical peaks. Select values other than zero to better reflect the beginning and end points of asymmetrical peaks.

A value of zero will not affect the sizing accuracy or precision for an asymmetrical peak.

Using These Parameters

Use the table below to move the start or end point of a peak.

IF you want to move the...	THEN change the...
start point of a peak closer to its apex 	Slope Threshold for Peak Start value from zero to a positive number
end point of a peak closer to its apex 	Slope Threshold for Peak End value to an increasingly negative number

Note: The size of a detected peak is the calculated apex between the start and end points of a peak and will not change based on your settings.

Slope Threshold Examples

**Initial
Electrophero-
gram**

The initial analysis with a value of 0 for both the Slope Threshold for Peak Start and the Slope Threshold for Peak End value produced an asymmetrical peak with a noticeable tail on the right side.

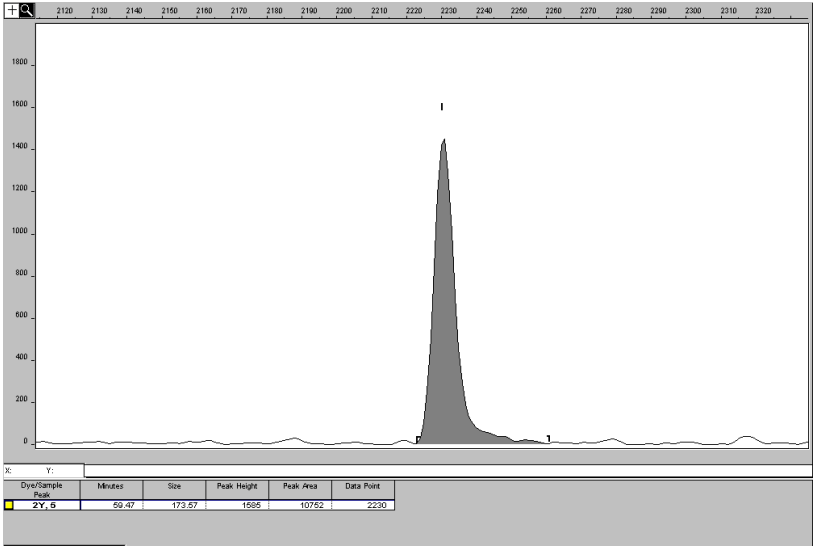


Figure B-8 Electropherogram showing an asymmetrical peak

**Electrophero-
gram After
Adjustments**

After reanalyzing with a value of -35.0 for the Slope Threshold for Peak End, the end point that defines the peak moves closer to its apex, thereby removing the tailing feature. Note that the only change to tabular data was the area (peak size and height are unchanged).

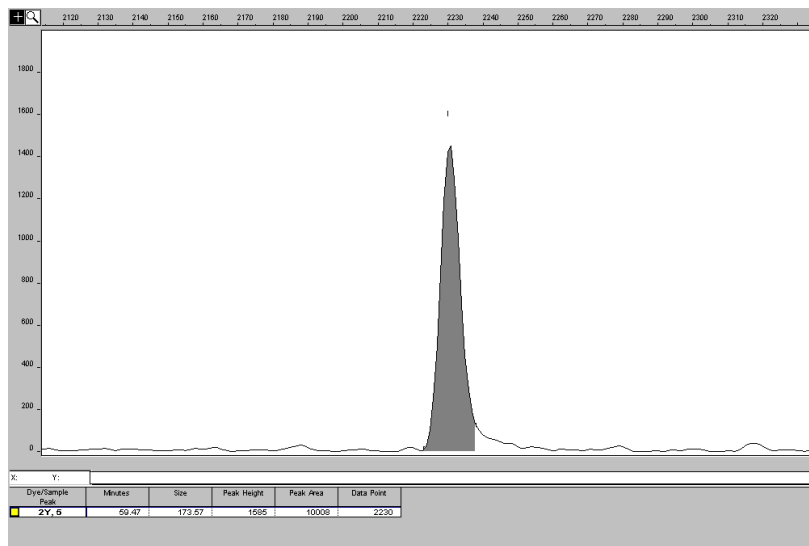


Figure B-9 Electropherogram showing the effect of changing the slope threshold for peak end

Size Calling Methods for Classic and Advanced Modes

Overview There are four size calling methods that you can use with the GeneMapper *ID* software v3.1. They are as follows:

- Least Square
- Cubic Spline Interpolation
- Local Southern
- Global Southern

Least Square Method Both Least Squares methods (2nd Order and 3rd Order) use regression analysis to build a best-fit size calling curve. This curve compensates for any fragments that may run anomalously. As a result, this method normally results in the least amount of deviation for all the fragments, including the size standards and the samples.

Depending on whether you choose the 2nd or 3rd Order Least Squares Method in the Analysis Parameters dialog box, the resulting size curve is either a quadratic or a cubic function. The software uses the known standard fragments and the associated data points to produce a sizing curve based on Multiple Linear Regression.

Advantages

In [Figures B-10](#) and [Figure B-11 on page B-30](#), you can see that in nearly all instances the mobility of an individual DNA fragment is coincident with the best curve fit of the entire data set. Stated differently, the mobility of most DNA fragments is strictly length-dependent. This method automatically compensates for fragments that run anomalously.

GeneMapper *ID* software v3.1 calculates a best-fit least squares curve for all samples, regardless of the size calling method you choose. The curve is black in the Standard Sizing Curve window.

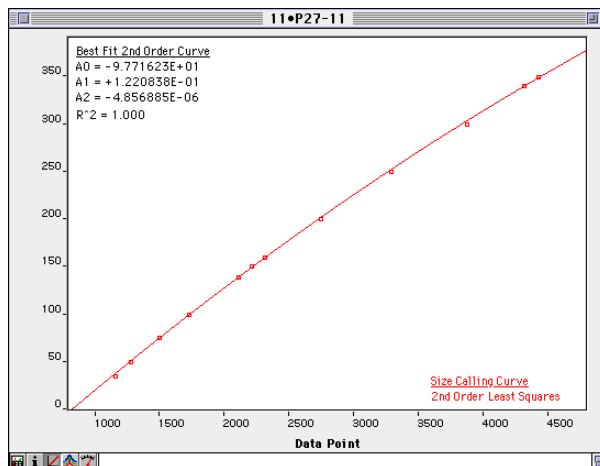


Figure B-10 2nd Order Least Squares size calling curve

Note: This graph was generated using GeneScan® 3.7.1 software. These results are similar to results obtained when you use GeneMapper *ID* software v3.1.

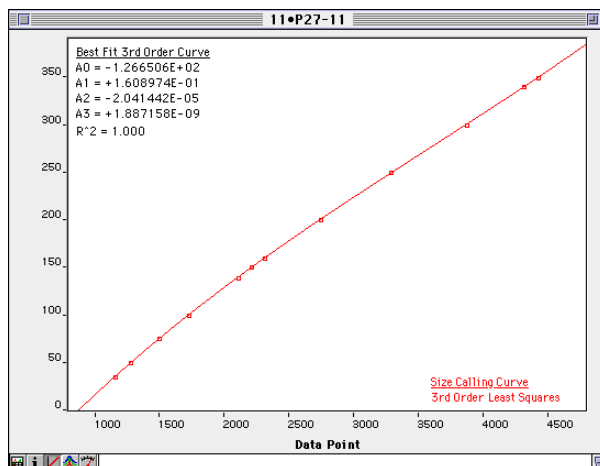
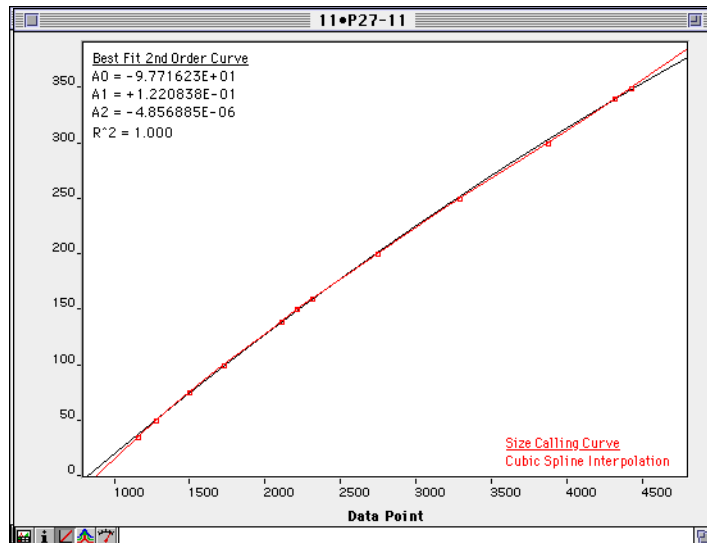


Figure B-11 3rd Order Least Squares size calling curve

Note: This graph was generated using GeneScan 3.7.1 software. These results are similar to results obtained when you use GeneMapper *ID* software v3.1.

Cubic Spline Interpolation Method

By definition, the Cubic Spline method forces the sizing curve through all the known points of the selected size standard. Although this enforcement produces exact results for the values of the standards themselves, it does not compensate for standard fragments that may run anomalously.



Note: This graph was generated using GeneScan 3.7.1 software. These results are similar to results obtained when you use GeneMapper *ID* software v3.1.

Possible Local Sizing Inaccuracy

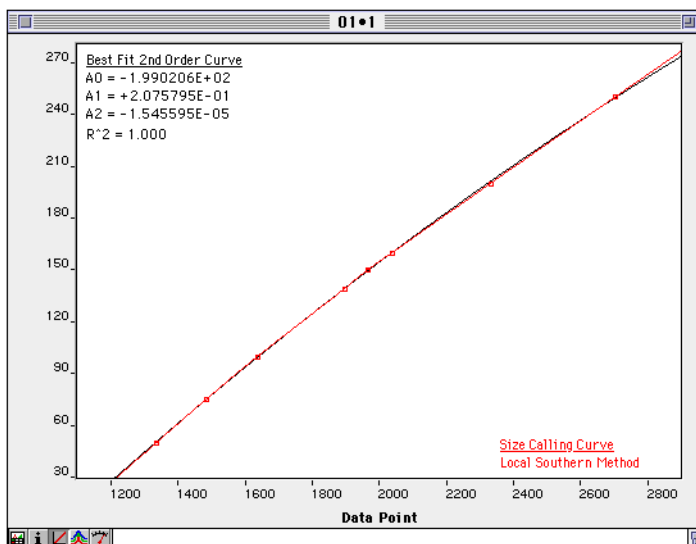
Mobility of any DNA fragment can be affected by its sequence, and by secondary and tertiary structure formation. If any internal size standard fragment has anomalous mobility, the Cubic Spline method may exhibit local sizing inaccuracy.

For example: Assume that a standard fragment is close in molecular length to an unknown sample fragment. Assume further that the standard fragment runs anomalously. The Cubic Spline method assigns the official value to this standard fragment, even though it may be slightly incorrect. The size of the unknown fragment is then likely to be calculated incorrectly as well.

Note: This method does not determine the amount of sizing accuracy error.

Local Southern Method

The Local Southern method determines the sizes of fragments by using the reciprocal relationship between fragment length and mobility, as described by E. M. Southern (1979).



Note: This graph was generated using GeneScan 3.7.1 software. These results are similar to results obtained when you use GeneMapper *ID* software v3.1.

The Equation

$$L = [c/(m-m_0)] + L_0$$

The equation attempts to describe the reciprocal relationship between the mobility, m , and the length, L_0 , of the standard fragments.

How This Method Works

This method, which is similar to the Cubic Spline method, uses the four fragments closest in size to the unknown fragment to determine a best fit line value. Only the region of the size ladder near the fragment of unknown length is analyzed.

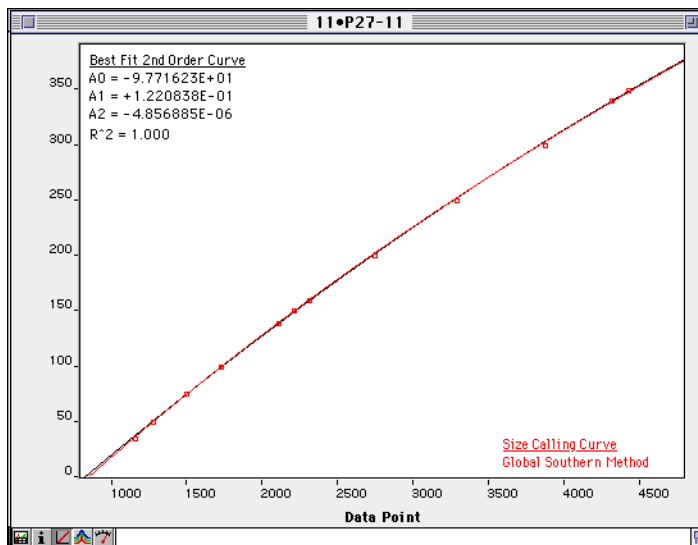
Note: Size estimates may be off if any of the standard fragments run anomalously.

The following table lists how the Local Southern method works:

1. The fitting constants of the curve are calculated for each group of three neighboring points on the standard. A separate curve is created for each set of three points.
2. A curve is then created by using three standard points (two points below and one point above the fragment) and a fragment size is determined.
3. Another curve is created by looking at an additional set of three points (one point below and two points above the fragment) and another value is assigned.
4. The two size values are averaged to determine the unknown fragment length.

Global Southern Method

This method is similar to the Least Squares method in that it compensates for standard fragments that may run anomalously. The method creates a best-fit line through all the available points, and then uses values found on that line to calculate the fragment values.



Note: This graph was generated using GeneScan 3.7.1 software. These results are similar to results obtained when you use GeneMapper *ID* software v3.1.

The Equations

The following table describes how the equations work:

Equation	Description
$L = [c/(m-m_0)] + L_0$	Attempts to describe the reciprocal relationship between the mobility, m , and the length, L_0 , of the standard fragments.
$\sum (L_i - (c/(m_i-m_0) + L_0))^2$	The fitting constants L_0 , m_0 , and c are calculated by a least squares fit to minimize the left side quantity.

How This Method Works

All points in the standard are weighted equally and the curve is not constrained to go through any specific point. The software can analyze a large range of fragment sizes with this method.

DNA fragments that are...	Are sized using...
not bracketed within the size standard curve	a second order least squares curve extrapolation.
bracketed within the size standard curve	the method that was chosen.

For best results, use a standard that brackets all the fragments of interest.

Project Window Software Interface



This appendix gives detailed descriptions of the features of the GeneMapper™ *ID* Software Version 3.1 application, including details of each major application window.

This appendix covers:

Overview of the Project Window	C-2
Project Window Menus	C-10
Project Window File Menu Dialog Boxes	C-27
Printing Tables and Electropherograms	C-34
Add Samples to Project Dialog	C-37
Project Window Edit Menu Dialog Boxes	C-45

Overview of the Project Window

Menus and Dialog Boxes

The Project window has a number of menus and dialog boxes associated with it. The drop-down menus available on each window (for example, File, Edit, View) provide a set of commands specific to the window. This section will describe the menu commands and dialog boxes associated with a given window.

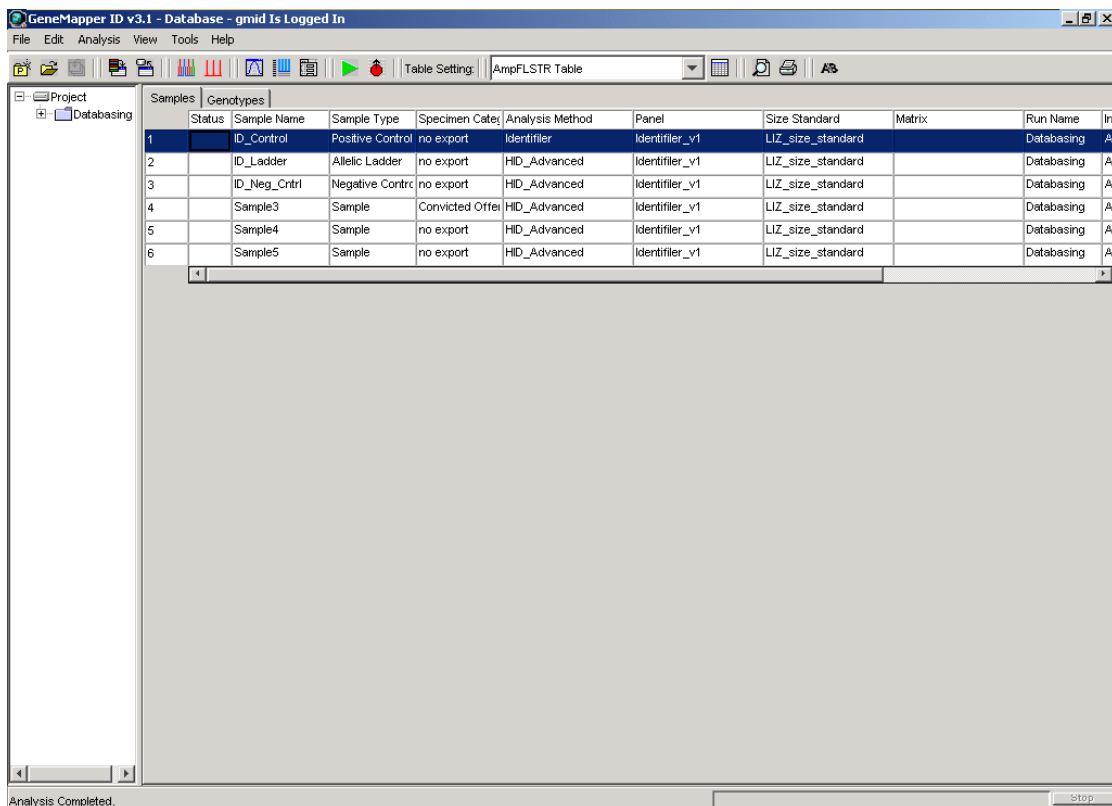
Samples View

The following information is provided in this section for the Samples view.

Topic	See Page
Appearance of Samples View	C-3
Samples View Columns	C-4
Samples View Features	C-6

Appearance of Samples View

The Samples view of the Project window displays the samples you want to analyze or have already analyzed. This view looks much like the following example when sample data has been imported.



Note: You can hide and show columns in the table using Table Setting.

Samples View Columns

The columns of the Samples view are explained in the table below.

	Status	Sample Name	Sample Type	Specimen Category	Analysis Method	Panel	Size Standard	Matrix	Run Name	In
1		ID_Control	Positive Control	no export	Identifier	Identifier_v1	LIZ_size_standard		Databasing	A
2		ID_Ladder	Allelic Ladder	no export	HID_Advanced	Identifier_v1	LIZ_size_standard		Databasing	A
3		ID_Neg_Cntrl	Negative Control	no export	HID_Advanced	Identifier_v1	LIZ_size_standard		Databasing	A
4		Sample3	Sample	Convicted Offender	HID_Advanced	Identifier_v1	LIZ_size_standard		Databasing	A
5		Sample4	Sample	no export	HID_Advanced	Identifier_v1	LIZ_size_standard		Databasing	A
6		Sample5	Sample	no export	HID_Advanced	Identifier_v1	LIZ_size_standard		Databasing	A

Samples view columns and descriptions:

Column	Description
Sample or row number	From application; used to select a single entire row.

Samples view columns and descriptions: *(continued)*

Column	Description
Status	A  symbol indicates that the sample needs to be analyzed or reanalyzed. Note: These symbols indicate status of each sample, and they control which samples will be analyzed during the next analysis if the Analyze icon is selected. There are other analysis options which ignore the status field. - The Status field next to each sample contains the  symbol prior to analysis. This field is empty after analysis, if analysis was successful. - Resetting parameters such as the Panel, Size Standard, Sample Type, and the Analysis Method resets the symbol for a given sample, enabling reanalysis.
Sample File	From sample sheet; not editable.
Sample Name	From sample sheet; editable, free text.
Comments	Free text. Editable.
Sample Type	Valid types are Sample, Control, Allelic Ladder, and Primer Focus, Negative Control Editable via Sample Type drop-down list box
Analysis Method	The list is editable via the Analysis Method Editor in the Analysis Methods tab in the GeneMapper Manager window.
Panel	Panel window contents come from Panel Manager. Editable via Panel window.
Size Standard	The list is editable via the Size Standard Editor in the Size Standards tab in the GeneMapper Manager window.
Matrix	The list is editable via the Matrix Editor in the Matrices tab in the GeneMapper Manager. For the ABI PRISM® 377 DNA Sequencer and the ABI PRISM® 310 Genetic Analyzer instruments, the matrix file is created in the Matrices Tab in the GeneMapper Manager window.
Run Name	From sample sheet; not editable.

Samples view columns and descriptions: (continued)

Column	Description
Instrument Type	From sample sheet; not editable.
Instrument ID	From sample sheet; not editable.
Run Date and Time	From sample sheet; not editable.
REF	Reference data;  indicates that the selected sample is defined as reference data in the Panel Manager.
SQI	Sizing quality invalidated; checkmark indicates that sizing quality value is 1.0.
User Defined 1-3	Also from the vertical bar enabled in the sample sheet fields. See Chapter 2, “Getting Started with GeneMapper ID Software.” Editable free text.

Note: Refer to Appendix A for PQV information.

Samples View Features

Feature	Description
Select Row(s)	Click the Row Header box; drag to select a continuous range.
Select Column(s)	Click the Column Header box; drag to select a continuous range.
Resize columns	Click and drag between columns to change column width.
Quick sort column	Shift-click the column header to sort ascending; shift-click again to sort descending.
Deselect Row	Ctrl-click the Row Header box

Genotypes View

The Genotypes view, shown below, displays the results of allele calling analysis. Each row or “record” in the table is a single marker for a sample. For example, if a panel specified for a sample contains four markers, the Genotypes table contains four records for that sample (each in a separate row).

Sample Name	Run Name	Panel	Marker	Dye	Allele 1	Allele 2	ADO	AE	OS	BIN	PHR	LPH	SPU	AN	BD	CC	OVL	GQ
Sample5	Databasing	Identifier_v1	AMEL	R	X	Y										NA		
Sample3	Databasing	Identifier_v1	CSF1PO	B	10	12										NA		
Sample4	Databasing	Identifier_v1	CSF1PO	B	10	13										NA		
Sample3	Databasing	Identifier_v1	D13S317	G	9	13										NA		
Sample4	Databasing	Identifier_v1	D13S317	G	12	13										NA		
Sample3	Databasing	Identifier_v1	D16S539	G	9	11										NA		
Sample4	Databasing	Identifier_v1	D16S539	G	12	13										NA		
Sample5	Databasing	Identifier_v1	D18S51	Y	15	18										NA		
Sample3	Databasing	Identifier_v1	D19S433	Y	12.2	15										NA		
Sample4	Databasing	Identifier_v1	D19S433	Y	12	14										NA		
Sample5	Databasing	Identifier_v1	D19S433	Y	13	14										NA		
Sample3	Databasing	Identifier_v1	D21S11	B	30	31										NA		
Sample4	Databasing	Identifier_v1	D21S11	B	28	29										NA		
Sample5	Databasing	Identifier_v1	D21S11	B	29	30										NA		
Sample3	Databasing	Identifier_v1	D3S1358	G	15	18										NA		
Sample4	Databasing	Identifier_v1	D3S1358	G	14	17										NA		
Sample5	Databasing	Identifier_v1	D3S1358	G	15	17										NA		
Sample3	Databasing	Identifier_v1	D5S818	R	12	13										NA		
Sample5	Databasing	Identifier_v1	D5S818	R	11	13										NA		
Sample3	Databasing	Identifier_v1	D7S820	B	9	10										NA		
Sample4	Databasing	Identifier_v1	D7S820	B	8	11										NA		
Sample3	Databasing	Identifier_v1	D8S1179	B	12	14										NA		
Sample4	Databasing	Identifier_v1	D8S1179	B	10	16										NA		
Sample5	Databasing	Identifier_v1	D8S1179	B	12	13										NA		
Sample3	Databasing	Identifier_v1	FGA	R	24	26										NA		
Sample4	Databasing	Identifier_v1	FGA	R	21	26										NA		
Sample5	Databasing	Identifier_v1	FGA	R	24	26										NA		
Sample3	Databasing	Identifier_v1	TH01	G	7	8										NA		

Genotype View Columns

The table in the Genotypes view can display some of the columns in the Samples view (see table on [page C-4](#)), plus these unique columns.

Genotypes view columns and descriptions:

Column	Description
Marker	Not editable; name of the marker associated with the record; created in the Panel Manager.

Genotypes view columns and descriptions: (continued)

Column	Description
Dye	From Panel Manager; not editable; indicates which dye is associated with the marker (B=blue, G=green, Y=yellow, R=red, or O=orange). Note: Not used for SNP Genotyping (SNaPshot® kit analysis).
Allele 1, 2, etc.	Not directly editable in the table cell; allele calls are editable only in the plot windows. The call for Allele 1. Allele calls and names are defined in the Panel Manager as bin names. The Allele call text box displays a list of valid calls plus one of the following: • OL (Off-ladder) for Human Identification (HID) applications • ? (for unknown) for microsatellite and SNP genotyping (SNaPshot kit) applications • blank for no allele calls. Note: When alleles shown in the Genotypes table are edited in the Plot window, the allele call in the table also changes and confidence value indicators in the table turn to grey triangles.
Size 1, 2, etc.	Peak size for Allele. Not editable.
Height 1, 2, etc.	Peak height for Allele. Not editable.
Peak Area 1 & 2	Peak area for Allele. Not editable.
Mutation	Indicates allele falling within a mutant bin.
AE Comment	Displays last user-edited comment.

Genotypes view columns and descriptions: (continued)

Column	Description
ADO (Allele Display Overflow)	- The box is labeled with an  when the number of alleles the marker calls exceeds the number to display previously set by you. - The user specifies how many alleles to display in the Table Settings parameters. (The default is two.) - There are six columns for each allele to indicate name, size, height, area, mutation, and comment.
AE (Allele Edit)	- The box displays unlabeled immediately after analysis. - The box is labeled with an  when the marker allele calls have been edited by you. - The marker allele calls can be edited in the Plot window. The user can enter comments in the Allele Edit column while editing allele calls.
User Defined 1-3	Editable; free text. Also from vertical bar enabled in sample sheet fields. See Chapter 2, "Getting Started with GeneMapper ID Software."

Note: Refer to Appendix A for PQV column information.

Project Window Menus

Introduction The tables in this section describe the following Project Window menus.

Topic	See Page
File Menu	C-10
Edit Menu	C-12
Analysis Menu	C-13
View Menu	C-15
Tools Menu	C-17
Help Menu	C-26

Command Activation and Special Terms

The commands in the Project Window menu can be activated in two ways:

- Clicking the command in the menu, or
- Using the special key combinations shown to the right of the name in the menu.

File Menu The commands are described in the table below.

<u>F</u> ile	<u>E</u> dit	<u>A</u> nalysis	<u>V</u> iew	<u>T</u> ools
<u>N</u> ew Project...				Ctrl+N
<u>O</u> pen Project...				Ctrl+O
<u>S</u> ave Project				Ctrl+S
Save Project As...				
Add S <u>a</u> mples to Project...				Ctrl+I
<u>E</u> xport Table...				Ctrl+E
<u>P</u> rint...				Ctrl+P
<u>L</u> og Out				
<u>E</u> xit				Alt+F4

Item	Description	Enabling
<u>N</u> ew Project (Ctrl+N)	Clears sample files, results, and project name from the Project window, displaying a blank Project. If previous project has pending changes, the following alert message is displayed: <i>Do you want to save changes? [Yes] [No] [Cancel]</i>	Always enabled except when a blank Project is displayed.
<u>O</u> pen Project (Ctrl+O)	Displays the Open Project dialog box. If previous Project has pending changes, the following alert message is displayed: <i>you want to save changes? [Yes] [No] [Cancel]</i>	Always enabled.
<u>S</u> ave Project (Ctrl+S)	Saves Project to the file named at the start of analysis. Named projects are saved to the GeneMapper <i>ID</i> database without a dialog box.	Enabled when the Project table has pending changes.
<u>S</u> ave Project As...	Displays the Save dialog box.	Always enabled.
A <u>d</u> d <u>S</u> amples To Project (Ctrl+I)	Opens the Add Samples To Project dialog box.	Always enabled.
<u>E</u> xport Table (Ctrl+E)	Displays the Export table dialog box. Exports table as tab or comma delimited text file.	Enabled when the Project table contains data.
<u>P</u> rint (Ctrl+P)	Displays the Print dialog box. The standard Print Setup dialog is opened from the Print dialog.	Enabled when the project table contains data.

Item	Description	Enabling
<u>L</u> og Out	Closes the Project window and displays the Login window. Displays a Save alert message if the Project has pending changes.	Always enabled.
E <u>x</u> it (Alt+F4)	Exits the GeneMapper <i>ID</i> application; displays Save alert message if Project has pending changes.	Always enabled.

Edit Menu The commands in the Edit menu are used to manage the contents of the Project window by performing standard actions like undo, delete, select, and by enabling access to settings for Preferences.

E dit	A nalysis	V iew	T ools
Delete from Project			
Select A <u>l</u> l		Ctrl+A	
Fill D <u>o</u> wn		Ctrl+D	
F <u>i</u> nd...		Ctrl+F	
S <u>o</u> rt...		Ctrl+G	

Item	Description	Enabling
D <u>e</u> lete From Project	Deletes the selected sample(s) from the Project and the database and displays the following alert message: <i>Deleting the selected sample(s) will delete both samples and results from project. This action cannot be undone.</i> [OK] [Cancel] Note: Each use of the command removes a single run or sample. If you want to remove all samples and the associated folder, use the Select All command before using the Delete from Project command. This action does not delete the actual sample file or sample data.	Enabled when a Project sample is selected.

Item	Description	Enabling
Select <u>A</u> ll (Ctrl+A)	Selects all samples in the active view.	Always enabled.
Fill <u>D</u> own (Ctrl+D)	Fills the selected column with the contents of the first cell. This command is applicable only to Analysis Method, Panel, Size Standard, and Matrix columns.	Enabled when a column is selected or multiple cells are selected.
<u>F</u> ind (Ctrl+F)	Displays the Find dialog box.	Enabled when the table contains data.
<u>S</u> ort (Ctrl+G)	Displays the Sort dialog box.	Enabled when the table contains data.

Analysis Menu The commands in this menu control analysis and display of the data in a Project and access to a number of other windows that support the Project window, such as the Analysis Manager and Panel Manager. The following table is provided as a reference for the Analysis menu.

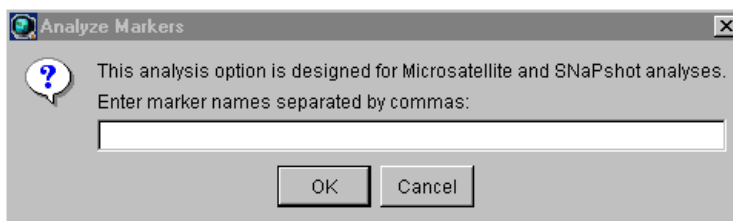
Analysis	<u>V</u> iew <u>T</u> ools <u>H</u> elp
<u>D</u> isplay Plots	Ctrl+L
Size Match Editor...	
<u>A</u> nalyze	Ctrl+R
Analyze <u>S</u> electd Samples	
<u>A</u> nalyze All	Ctrl+Shift+R
Analyze <u>M</u> arkers...	
Analysis Meth <u>o</u> d Editor...	
Size Standard Editor...	
<u>B</u> ring Errors to Top	Ctrl+B
Bring Non-concordant <u>S</u> amples to Top	

Item	Description	Enabling
<u>D</u> isplay Plot (Ctrl+L)	Opens the Plot window.	Enabled when one or more samples or records are selected.
Size Match Editor	Opens the Size Match Editor window.	Enabled only when sized samples are present in the Project.

Item	Description	Enabling
Analyze (Ctrl+R)	Analyses all samples eligible for analysis. The symbol is shown in the Status column.	Enabled when Samples tab is visible and at least one sample in the table requires analysis.
Analyze <u>S</u> elect Samples	Analyzes selected samples only.	Enabled when Samples tab is visible.
Analyze All (Ctrl+Shift+R)	Analyzes all samples whether or not the Status column reports that the sample is eligible for analysis, including those previously analyzed. The following alert message is displayed: <i>Analyze all samples? This may change your existing results. This action cannot be undone.</i> [OK] [Cancel]	Enabled when Samples tab is visible.
Analyze <u>M</u> arkers	Analyzes only the marker(s) you select within each sample. (" Analyze Markers Dialog Box " on page C-15.)	Enabled when Samples tab is visible.
Analysis Meth <u>o</u> d Editor	Allows you to change the settings for analysis.	Enabled when a sample with an analysis method is selected.
Size Standard Edit <u>o</u> r	Allows you to edit base pair and dye settings.	Enabled when one or more samples or records are selected.
Low Quality To Top (Ctrl+B)	Sorts all Low Quality (SQ or GQ column) samples or results records to the top of the table.	Enabled when analyzed samples are present in the table.
Non-concordan <u>t</u> Samples to Top	Brings non-concordant samples to the top of the analysis page.	—

Analyze Markers Dialog Box

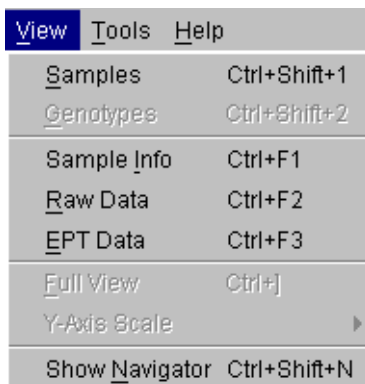
The Analyze Markers dialog box enables you to select which marker, or set of markers separated by commas, will be analyzed within each sample.



This option is only enabled when you are in the Project window Samples tab and samples are present. Does not work with HID samples.

View Menu

The View menu is used to hide/show the Project window navigation pane and switch between the two Project window tabs. The following table is provided as a reference for the View menu.

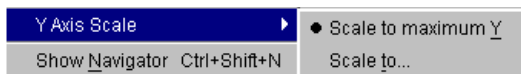


Item	Description	Enabling
<u>S</u> amples (Ctrl+Shift+1)	Switches to the Samples tab.	Always enabled.
<u>G</u> enotypes (Ctrl+Shift+2)	Switches to the Genotypes tab.	Enabled when results are present.

Item	Description	Enabling
Sample Info (Ctrl+F1)	Switches to the Info tab for the selected sample in the project.	Always enabled.
Raw Data (Ctrl+F2)	Switches to the Raw Data tab for the selected sample in the project.	Always enabled.
EPT Data (Ctrl+F3)	Switches to the EPT tab for the selected sample in the project.	Always enabled.
Full View (Ctrl+J)	Shows the full view of the Raw Data or EPT graph.	Enabled only when viewing the Raw Data or EPT Data of a sample.
Y-Axis Scale	Changes Y-axis scale. (See “Y Axis Scale” on page C-16.)	Enabled only when viewing the Raw Data or EPT Data of a sample.
Show Navigator (Ctrl+ Shift+N)	Switches the navigator pane (shows/hides the pane).	Always enabled.

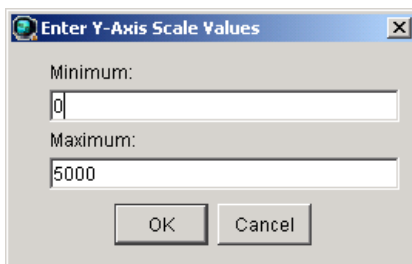
Y Axis Scale

The Y Axis Scale option is only enabled when viewing the raw data or EPT data of a sample file.

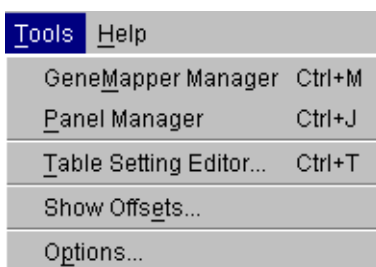


There are two options:

- **Scale to maximum Y** - scales to the maximum Y value for the selected sample.
- **Scale to** - opens a dialog box in which a user can define the value of the Y axis.



Tools Menu The Tools menu provides access to the GeneMapper Manager, Panel Manager, and Table Setting Editor applications.



Item	Description	Enabling
GeneMapper Manager (Ctrl+M)	Opens the GeneMapper <i>ID</i> dialog box. (See Chapter 5, “Using GeneMapper Manager.” .)	Always enabled.
Panel Manager (Ctrl+J)	Opens the Panel Manager dialog box. (See Chapter 4, “Using Panel Manager.”)	Always enabled.
Table Setting Editor (Ctrl+T)	Opens the Table Setting Editor dialog box. (See “Table Settings Tab” on page 5-27.)	Only enabled when samples are present. Only allows editing of a currently selected Table Profile.

Item	Description	Enabling
Show Offsets	Displays bin offsets for selected samples.	Always enabled.
Options	Opens the Options dialog box. (See “Options Dialog Box” on page C-18.)	Always enabled.

Options Dialog Box

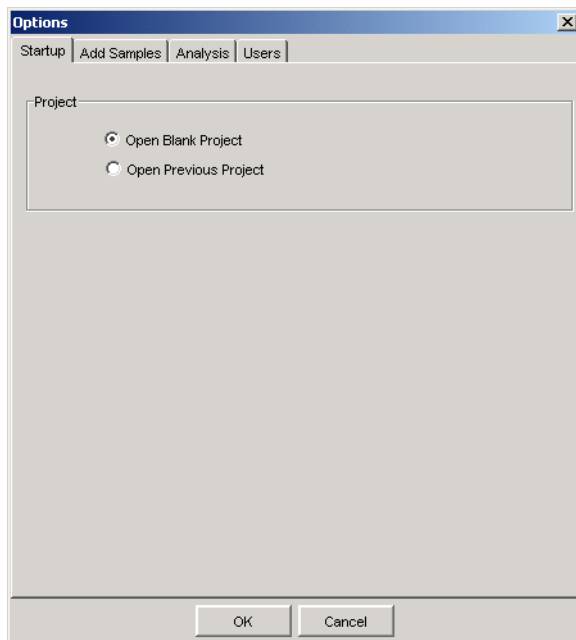
The Options dialog box contains several tabs for setting preferences associated with various software features. The following tabs are described below:

Topic	See Page
Startup Tab	C-19
Add Samples Tab	C-20
Analysis Tab	C-22
Users Tab	C-24

Note: Each registered user has his or her own set of preferences. When you set these options, it only affects the currently logged in user.

Startup Tab

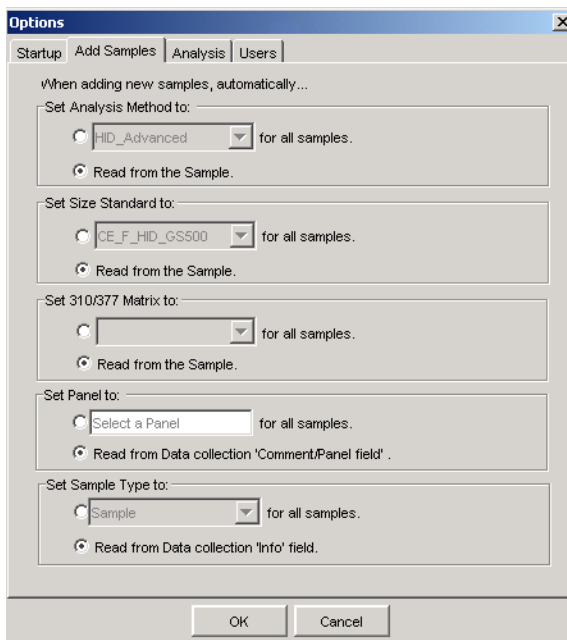
The Startup tab contains preferences associated with the startup of the software.



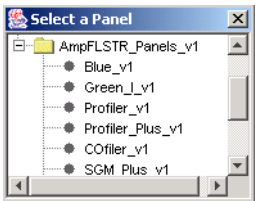
Element	Description
Open Blank Project option button	Sets the software so that upon launch, a blank Project window opens.
Open Previous Project option button	Sets the software so that upon launch, the last Project viewed will open.

Add Samples Tab

The Add Samples tab contains preferences associated with the adding of samples.



Element	Description
Set Analysis Method to drop-down menu and Read from the Sample option button	When you add samples, the Analysis Method property is set to the value in the text box, which contains the list of analysis methods from the GeneMapper <i>ID</i> database.
Set Size Standard to drop-down menu and Read from the Sample option button	When you add samples, set the size standard property to the value in the text box, which contains the list of size standards from the GeneMapper <i>ID</i> database.
Set 310/377 Matrix to drop down menu Read from the Sample option button	Set the matrix file to the dye you are using or read from the sample file.

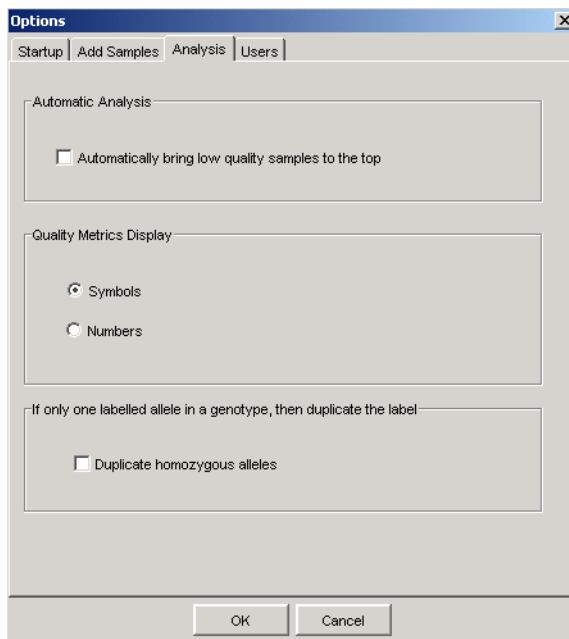
Element	Description
Set Panel to • Select a Panel for all samples option button and window • Read from the Data collection 'Comment/Panel field' option button	When you add samples, the panel for all samples is set to the value in the text box. When the option button is selected, the following choices may be made: • Leave the text alone, which selects the existing name, or • Click the field to present the Select a Panel window (to select an existing panel) The Select a Panel window contains the list of panels from the GeneMapper <i>ID</i> database.  When you add samples, the panel for all samples is set to the value in the Sample Sheet “comment” column.
Set Sample Type to • Sample type for all samples option button and drop-down menu • Read from Data collection 'Info' field option button	When you add samples, the Sample Type for all samples is set to the value chosen from the Sample Type drop-down menu, contains choices for Sample, Control, Allelic Ladder, and Primer Focus. When you add samples, the GeneMapper <i>ID</i> software does a “best guess” match, comparing the contents of the field specified in the Sample type box with the list of sample types (Sample, Control, Allelic Ladder, Negative Control, and Primer Focus). • If a match is found, then the Sample Type property for the sample is set to that sample type. • If a match is not found, then the Sample Type property for the sample is set to “Sample.”

Note: The Panel and Sample Type properties accounts for close spellings and abbreviations as shown in the following table.

Case	Result
A sample file field contains either Control, Ctrl, Crl, Cntrl, or a similar variation.	The sample type for the sample is set to Control.

Analysis Tab

The Analysis tab contains preferences associated with the analysis of data.

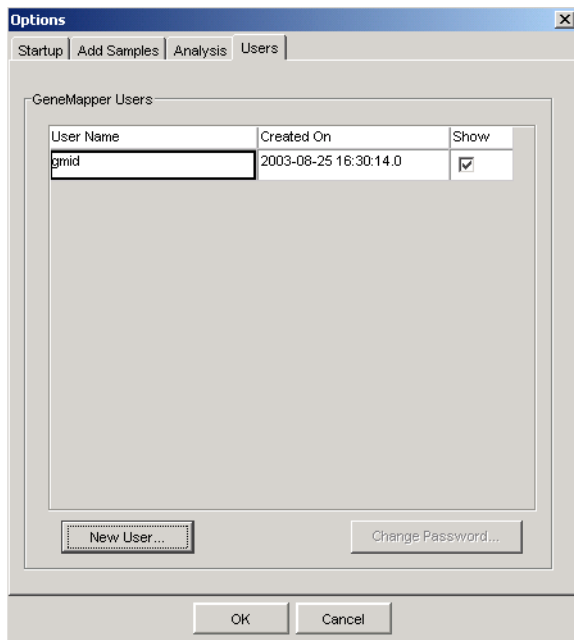


Element	Description
Automatic Analysis group box: Automatically brings errors to the top of the table check box	When samples are analyzed, the software sorts the table automatically to bring samples with errors to the top of the table. Same functionality as the Bring Errors To Top icon in the Project window.

Element	Description
Quality Metrics Display group box: Symbols button Numbers button	Display quality values in project tables as symbols (green square, yellow triangle, red octagon), or numerical values (0.0, ..., 1.0). Note: If Symbols (default) is chosen, then all quality values are shown as symbols. If Numbers is chosen, then only SQ and GQ columns are shown as numbers; the other result columns are symbols.
If only one allele is labeled in a genotype, then duplicate the label group box: Duplicate homozygous alleles checkbox	If this option is checked, single alleles will be displayed in the Genotypes table and in the Plot window as two labels to indicate presumed homozygotes.

Users Tab

The Users tab contains preferences associated with managing registered users.

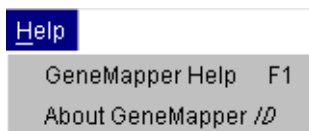


Note: Once created, user names cannot be deleted.

Element	Description
User Name field	Displays the list of registered users. • User Name column = Your name • Created On column = Date user was created • Show column = Shows your name in User Name drop-down menu of the login window, if checked. Clearing a Show column check box for a user blocks login for that user (can be done by anyone running GeneMapper ID software). Shift-click the column label headers to sort the list by that column.

Element	Description
New User button	Opens the New User dialog box.  To add a user name for GeneMapper <i>ID</i> software, click New User to open a dialog box and then: - Type a user name into the dialog box. - Enter a password using only alphabetic characters. - Confirm the password. - Click OK to assign the user name and close the dialog. When you select this user name on launching GeneMapper <i>ID</i> software, the name is used to identify the preferences set by you. Note: User names must be unique. If the new user name is not unique, an alert message will be displayed. Note: Users added here are for this GeneMapper <i>ID</i> software installation and is visible to all other users. The names are converted to lower case and passwords are not case sensitive.
Change Password	Opens the Change Password dialog box, and allows you to change the password for the selected user. 

Help Menu The Help menu provides access to online help and to the About GeneMapper *ID* software window.



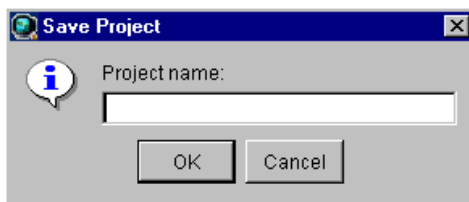
Item	Description	Enabling
GeneMapper Help (F1)	Opens the GeneMapper <i>ID</i> User Manual PDF file using Acrobat Reader. Does not open the tutorial guides.	Always enabled.
<u>A</u> bout GeneMapper <i>ID</i>	Opens the About GeneMapper <i>ID</i> window and displays the software version.	Always enabled.

Project Window File Menu Dialog Boxes

Introduction The following dialog boxes are accessed from the Project window File menu. This section describes each of the dialog boxes.

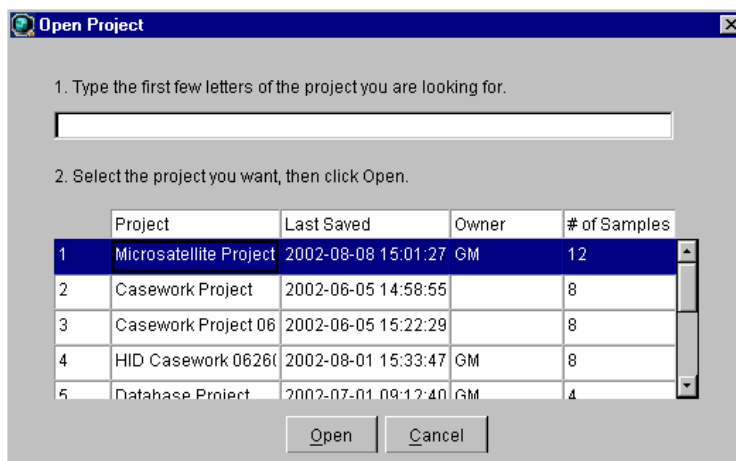
Note: This section contains information on all application dialog boxes except for the “Add Samples to Project” dialog box, which is covered under its own section (See [“Add Samples to Project Dialog” on page C-37](#)).

Save Dialog Box The Save dialog box, accessed from the New Project window, is presented when a project already exists in the Project window, and provides you with the option of saving the project.



Open Project Dialog Box

The Open Project dialog box displays the list of projects in the database. You use the first field to find a project quickly, then click the Open button to open the selected project (or double-click the project name).



Note: Once you have opened a Project window, you may resize it horizontally and vertically to increase the width and depth of the Project table.

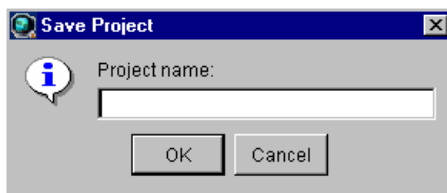
The following table describes the Open Project dialog box window elements.

Element	Description
Search field	Typing in this field selects the first item in the project list whose name matches the characters.

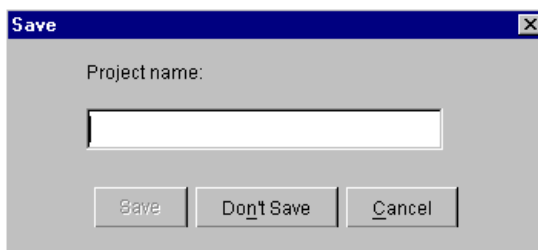
Element	Description
Project table	Lists all the projects in the database. • Project = Name of the project. • Last Saved = Date and time stamp of when the project was last modified. • Owner = Name of the user who created the project. • # of Samples = the number of samples in the project. Note: Columns may not be moved or hidden. You may resize the width of the columns by dragging between the column headers. Only one row in the table may be selected at a time. You may navigate up and down in the table using arrow keys. Shift-clicking a column header sorts the data by that column.
Open	Opens the selected project. If previous project has pending changes, the following alert message is displayed before the Open Project dialog box opens: <i>Do you want to save changes? [Yes] [No] [Cancel]</i>
Cancel	Closes the Project Manager dialog box.

Save Project and Save Dialog Boxes

The Save Project and Save boxes enable you to save projects to the database. The first dialog box is presented for the Save command if you want to save a project before it is analyzed. Using Save after analysis does not require a dialog box because you are required to name a Project before analysis.



The Save dialog box opens when you select the Save As command.



The elements of these dialog boxes are described in the table below.

Element	Description
Project name	Text field for the name of the project; accepts alphanumeric characters. The database requires standard Windows interface restrictions on length and symbol characters.
OK	Saves the project to the database. Saved projects are tagged with the Project Name, User Name, and Time/Date Stamp. If the name you entered already exists, an alert message is displayed. Cancel closes the alert dialog and displays the Save Project dialog box again.
Cancel	Closes the dialog box and does not save the project.
Save	Saves the project under the name entered in the dialog box.
Don't Save	Aborts the Save action.

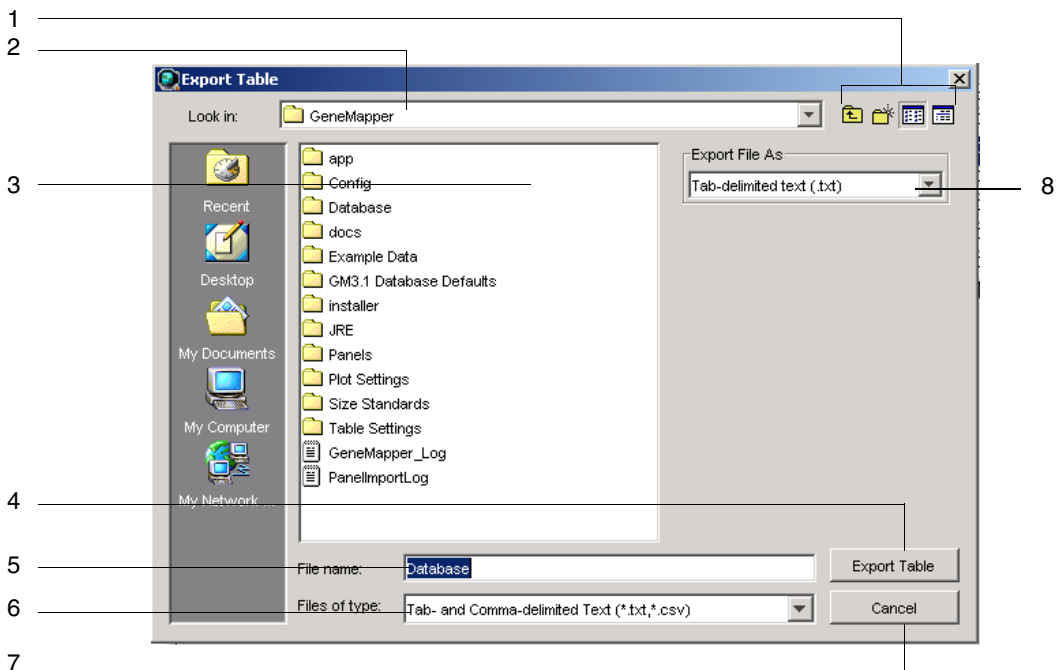
Export Table Dialog Box

The Export Table dialog box allows you to choose where to save the exported file.

The file types supported include:

- Tab-delimited text (.txt)
- Comma-separated values (.csv)

Note: Export operates on the currently-displayed tab. For example, if the Samples tab is displayed, Export would export the tab-delimited data shown in the Samples table. Exported data includes column headers.

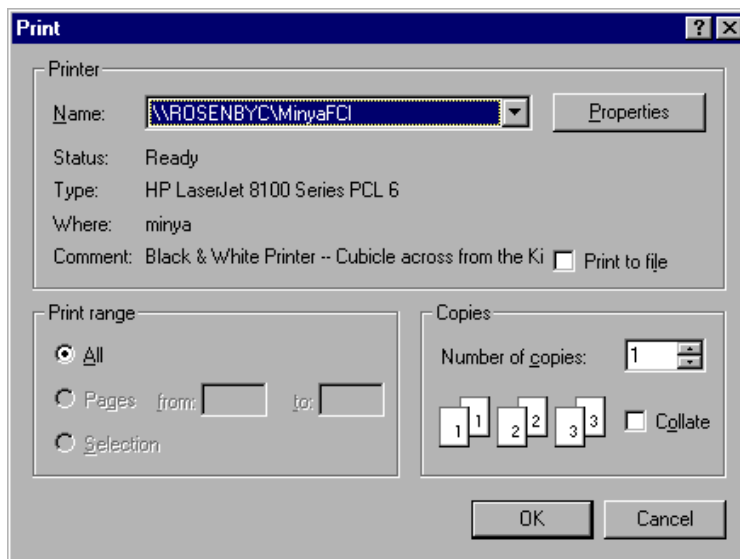


Export table callouts

Item	Name	Description
1	Toolbar	Click these icons to:
		 Move display up one level in main pane.
		 Move display to Home level in main pane. This is usually Profiles\<user>.
		 Create a new folder at the present directory level.
		 Present a list of the contents of the selected folder
		 Present details of the selected folder.
2	Drop-down directory menu	Select drive letter and/or folder.
3	Folder/file display pane	The contents of the folder selected in the toolbar are displayed here.
4	Export Table button	Exports the selected table.
5	File name field	Enter a file name to use for the exported table.
6	Show files of type field	This is a display filter for files (folders are always shown). Use to select the file extensions for display: • All files • .txt files • .csv files • .txt&.csv
7	Cancel button	Closes the Export Table window without exporting a table.
8	Export file as field	Select the format of the file you want to export.

Note: Exported text tables show PQVs as numbers even if they are displayed as symbols.

Print Dialog Box The Print dialog box, shown below, is a print file dialog box that controls printing. The Samples table, the Genotypes table, Panel Manager views, and plot windows may be printed.



Item	Description
Properties button	Presents the dialog box used to set up the printer.
Name field	Shows the currently selected printer. Note: The drop-down menu to the right allows selection of other available printers.
Status	Indicates status of selected printer.
Type	Shows type of printer.
Where	Shows the path name of the selected printer.
Comment	Check box enables printing the project to a file.
Page range	All is the only option available when working with tables.
Number of Copies	Select the number of copies to print.
Collate options	Check box enables collating of copies.

Printing Tables and Electropherograms

Introduction All tables and electropherograms can be printed as what-you-see-is-what-you-get (WYSIWYG), except for these conditions:

- Only the number of columns shown in the table printout below are printed out on a single sheet of paper in the Portrait mode. Additional columns to the right are printed out on additional sheets of paper. Change to the Landscape mode to minimize the number of sheets of paper used.

Note: To reduce the number of sheets of paper printed for a table, hide columns you do not want to print using the Table Settings Editor.

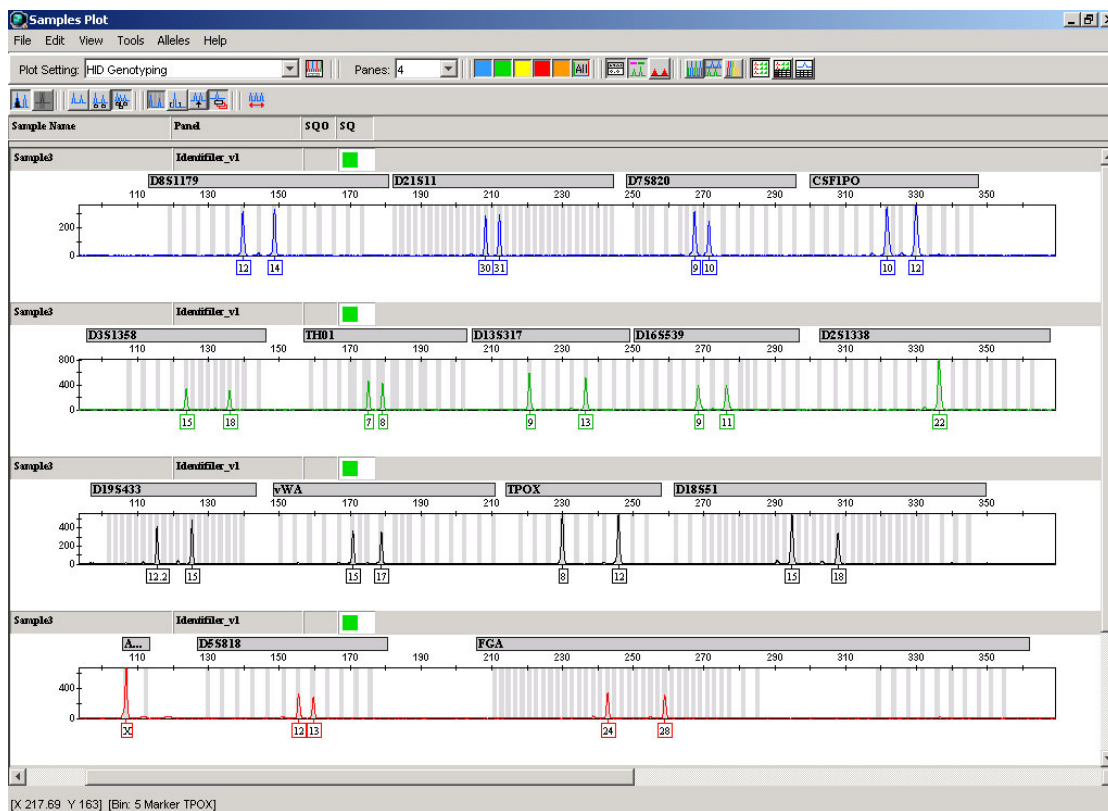
- Printed plots are sized vertically to fill a single sheet of paper:
 - With four plots, the plots are sized close to the same size as the default size on your monitor.
 - Six plot panes are a practical limit for a single sheet.

Example Table Printout

Sample Name	Run Name	Panel	Marker	Dye	Allele 1	Allele 2
ID_Neg_Cntrl	Databasing	Identifier_v1	D8S1179	B		
ID_Neg_Cntrl	Databasing	Identifier_v1	D21S11	B		
ID_Neg_Cntrl	Databasing	Identifier_v1	D7S820	B		
ID_Neg_Cntrl	Databasing	Identifier_v1	CSF1PO	B		
ID_Neg_Cntrl	Databasing	Identifier_v1	D3S1358	G		
ID_Neg_Cntrl	Databasing	Identifier_v1	TH01	G		
ID_Neg_Cntrl	Databasing	Identifier_v1	D13S317	G		
ID_Neg_Cntrl	Databasing	Identifier_v1	D16S539	G		
ID_Neg_Cntrl	Databasing	Identifier_v1	D2S1338	G		
ID_Neg_Cntrl	Databasing	Identifier_v1	D19S433	Y		
ID_Neg_Cntrl	Databasing	Identifier_v1	VWA	Y		
ID_Neg_Cntrl	Databasing	Identifier_v1	TPOX	Y		
ID_Neg_Cntrl	Databasing	Identifier_v1	D18S51	Y		
ID_Neg_Cntrl	Databasing	Identifier_v1	AMEL	R		
ID_Neg_Cntrl	Databasing	Identifier_v1	D5S818	R		
ID_Neg_Cntrl	Databasing	Identifier_v1	FGA	R		
Sample3	Databasing	Identifier_v1	D8S1179	B	12	14
Sample3	Databasing	Identifier_v1	D21S11	B	30	31
Sample3	Databasing	Identifier_v1	D7S820	B	9	10
Sample3	Databasing	Identifier_v1	CSF1PO	B	10	12
Sample3	Databasing	Identifier_v1	D3S1358	G	15	18
Sample3	Databasing	Identifier_v1	TH01	G	7	8
Sample3	Databasing	Identifier_v1	D13S317	G	9	13
Sample3	Databasing	Identifier_v1	D16S539	G	9	11
Sample3	Databasing	Identifier_v1	D2S1338	G	22	
Sample3	Databasing	Identifier_v1	D19S433	Y	12.2	15
Sample3	Databasing	Identifier_v1	VWA	Y	15	17
Sample3	Databasing	Identifier_v1	TPOX	Y	8	12
Sample3	Databasing	Identifier_v1	D18S51	Y	15	18
Sample3	Databasing	Identifier_v1	AMEL	R	X	
Sample3	Databasing	Identifier_v1	D5S818	R	12	13
Sample3	Databasing	Identifier_v1	FGA	R	24	28
Sample4	Databasing	Identifier_v1	D8S1179	B	10	16
Sample4	Databasing	Identifier_v1	D21S11	B	28	29
Sample4	Databasing	Identifier_v1	D7S820	B	8	11
Sample4	Databasing	Identifier_v1	CSF1PO	B	10	13
Sample4	Databasing	Identifier_v1	D3S1358	G	14	17
Sample4	Databasing	Identifier_v1	TH01	G	9	9.3
Sample4	Databasing	Identifier_v1	D13S317	G	12	13
Sample4	Databasing	Identifier_v1	D16S539	G	12	13
Sample4	Databasing	Identifier_v1	D2S1338	G	25	
Sample4	Databasing	Identifier_v1	D19S433	Y	12	14
Sample4	Databasing	Identifier_v1	VWA	Y	16	18
Sample4	Databasing	Identifier_v1	TPOX	Y	8	
Sample4	Databasing	Identifier_v1	D18S51	Y	18	
Sample4	Databasing	Identifier_v1	AMEL	R	X	
Sample4	Databasing	Identifier_v1	D5S818	R	12	
Sample4	Databasing	Identifier_v1	FGA	R	21	26
Sample4	Databasing	Identifier_v1	D8S1179	B	12	13
Sample5	Databasing	Identifier_v1	D21S11	B	29	30

Note: Additional pages are created to accommodate the number of rows in the table. Each additional page that continues the table to the right would have the same row numbers as shown in the left column.

Example Plot Printout



Note: The number of plots per page is set by the Panes dialog box in the Plots window. The example above was scaled down vertically and would be printed on the whole height of the page on a printer.

Add Samples to Project Dialog

Introduction This section provides general information about:

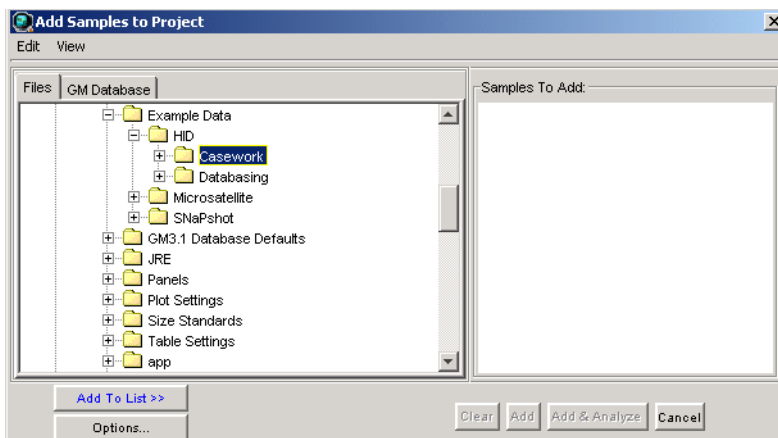
- the Add Samples to Project dialog
- the Edit and View menus available in the window
- using the three tabs in the window.

This following topics are covered in this section.

Topic	See Page
General Information and Procedure	C-37
Edit Menu	C-38
View Menu	C-40
Files Tab	C-41
GM Database Tab	C-42

General Information and Procedure

The Add Samples to Project window provides a navigation pane to enable you to add Samples to the project. You can also access the dialog box by clicking the Add Sample icon on the Project window toolbar.



To add samples to the Project:

1. Click  (**Add Samples To Project**) on the Project window toolbar.

Add Samples to Project window opens to the last tab viewed by you (default is Files tab).

Sample files can reside on the local hard drive(s), mapped network drive(s), and removable media drives.

2. In **Files** tab, navigate to folder containing sample files. Select the folder, then click the **Add to List** button.

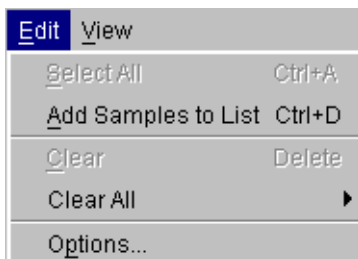
Folder is added to the **Samples To Add** field. Individual samples within a folder can be selected and added. Selecting the folder adds all samples.

3. Click the **Add** button.

Add Samples to Project window closes and the project window becomes visible. Samples are added to the bottom of the Project window table.

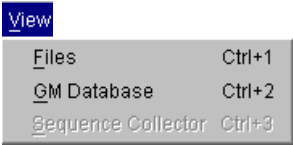
Edit Menu

The commands in the Edit menu of the Add Samples window are used to manage the contents of the window. The commands are described in the table below.



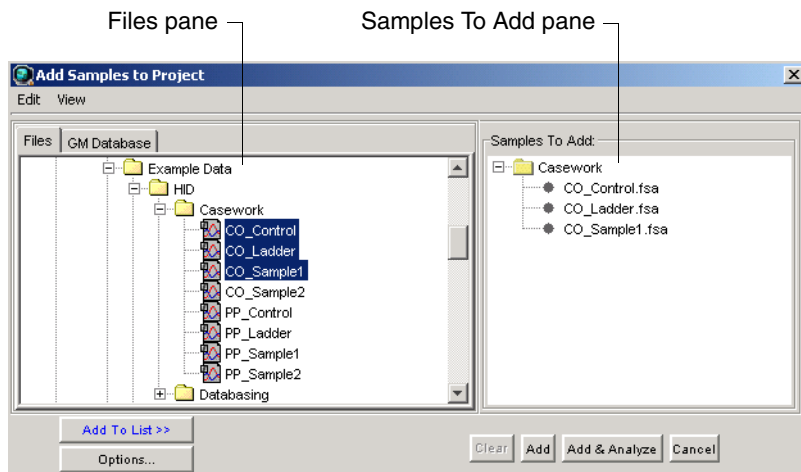
Item	Description	Enabling
Select All (Ctrl+A)	Selects all the items of a given type.	Enabled when a single valid selection has been made (e.g., sample file).
Add Samples to List (Ctrl+D)	Adds the selected item(s) from the Files, GeneMapper <i>ID</i> database.	Enabled when a sample file or folder with files in the Files, GeneMapper <i>ID</i> database is selected.
Clear (Delete)	Clears (deletes) the selected item(s) in the Samples to Add list.	Enabled when a valid selection is made.
Clear All	Hierarchical menu. Clears all sample files and folders from the specified field: • <u>S</u>amples to add • <u>C</u>riteria Note: The Criteria subcommand functions in the GeneMapper <i>ID</i> Database tab but not in the Files tab and is used to clear all search criteria from the Criteria Selector List (see the figure on page C-42). If undo is not possible, the following alert message is displayed: <i>Clear all items? You cannot undo this action. [Yes] [No] [Cancel]</i>	Enabled when the field is present and an item exists in that field.
Options	Opens the Options dialog box in the Add Samples tab.	Always enabled.

View Menu The View menu of the Add Samples window is used to switch between the three sources for samples and refresh the file directory. The commands are described in the table below.



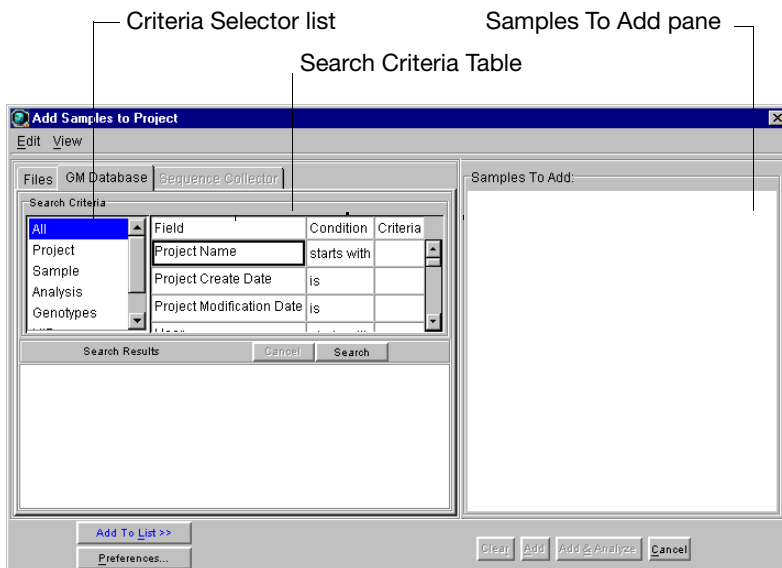
Item	Description	Enabling
Files (Ctrl+1)	Opens the Files tab.	Always enabled.
GM Database (Ctrl+2)	Opens the GeneMapper <i>ID</i> Database tab.	Always enabled.

Files Tab The Files tab enables you to choose sample files from local or mapped networked storage devices.



Element	Description
Files pane	Contains the directory structure configured for the computer running GeneMapper <i>ID</i> software. The navigation pane provides a tree viewer that works like the MS Windows Explorer interface. You can use Shift-click for continuous multiple selection and Ctrl-click for discontinuous multiple selections within a folder.
Samples To Add pane	Contains the folders and files that the system will add. This field also uses the tree viewer interface. You can use Shift-click and Ctrl-click as described above.
Add To List button	Adds the selected items from the files pane to the Sample to Add field.
Options button	Opens the Options window.
Clear button	Clears the selected files from the Samples To Add pane.
Add button	Adds the Samples To Add list into the Project window.
Cancel button	Closes the Add Samples to Project dialog box.

GM Database Tab The GM (GeneMapper *ID*) Database tab enables you to choose sample files that have previously been added into a GeneMapper *ID* project. This database uses a search interface to extract samples that match your criteria.



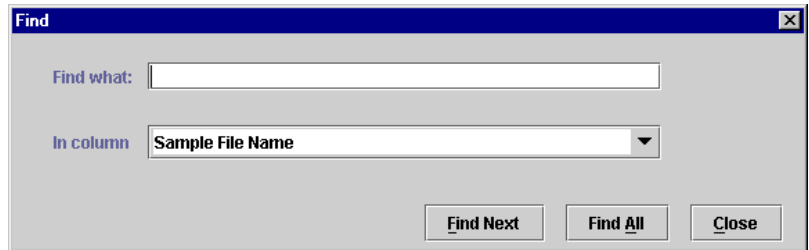
This feature allows mixing and matching sample files. For example, you can merge sample files from a number of small projects into a single large project.

Criteria and Rules	Description
Search criteria	This list and table contains the search criteria from which to extract samples from the GeneMapper <i>ID</i> database. Note: Locations of the list and table are pointed out in the figure on the previous page. • Criteria selector list = list of properties (columns in the project window) grouped by topic. • Field column = Text field that contains the properties (columns) in the selected group criterion. • Condition column = Combo box that contains conditions that the system will use to search for samples. Search rules include: – Is – Contains – Starts With – Date criteria - Presents a calendar widget to specify the date • Criteria column = Field for specifying the text string associated with the search rules which are case sensitive.
Rules for Search criteria	Criteria accumulate as they are entered in different rows and to different groups of criteria. Multiple items in the search criteria list use the natural language “and” for searching. For example, the natural language of the Criteria field is: “Display samples whose Panel property contains “XXX” and display samples whose Collection Instrument property contains “YYY”.”

Element	Description
Search Criteria Cancel button	Enabled when a search is in progress; search stops when clicked. Note: You may not stop the search during data transfer from the GeneMapper <i>ID</i> database.
Search button	Searches the database using the criteria list and displays the search results in the Search Results field.
Search Results field	Displays the results of the database search (sample files).
Add To List button	Adds the selected items from the Results field to the Samples to Add field.
Samples To Add field	Contains the folders and files that the system will add. This field uses the tree viewer interface.
Options button	Opens the Options window.
Clear button	Clears the selected files from the Samples To Add field.
Add button	Adds the Samples to Add list into the Project window.
Cancel button	Closes the Add Samples to Project dialog box.

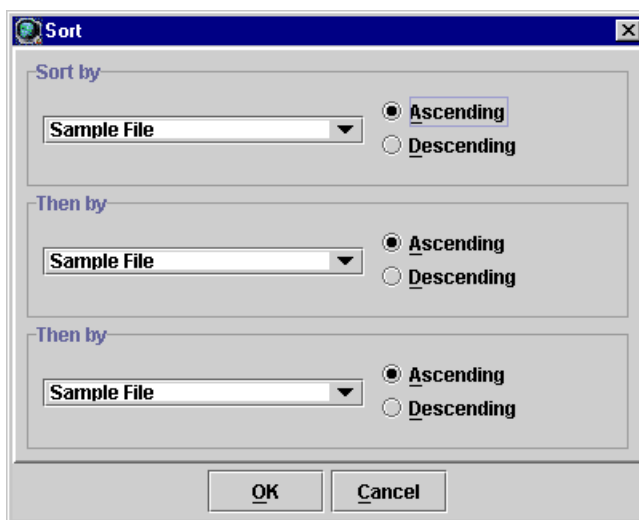
Project Window Edit Menu Dialog Boxes

Find Dialog Box The elements of the Find dialog box is described below.



Element	Description
Find what	Field containing the search string.
In column	List box containing the names of columns in the table.
Find Next	Finds the next instance of the search string in the specified column and selects it.
Find All	Finds and selects all rows in the table that contain the search string in the specified column.
Close	Closes the Find dialog box. Table selections remain intact.

Sort Dialog Box The Sort dialog box, shown below, allows sorting of sample files and results by ascending or descending order in up to three steps.



The Sort dialog box has three entry fields for entering sort criteria (the “Then by” fields allow successive sorting by two other criteria).

Element	Description
Sort by (Then by)	Sort box containing the column which will be sorted.
Ascending and Descending buttons	Option buttons for choosing if the sort is ascending or descending in alphabetical/numerical order.
OK button	Closes the Sort dialog box and performs the sort.
Cancel button	Closes the Sort dialog box. Does not perform the sort.

GeneScan Size Standards

D

This appendix covers:

Introduction	D-2
GeneScan 120 Size Standard.	D-3
GeneScan 350 Size Standard.	D-4
GeneScan 400HD Size Standard.	D-5
GeneScan 500 and GeneScan 500(-250) Size Standards	D-7

Introduction

The GeneMapper™ ID Software Version 3.1 comes with several ready-to-use GeneScan™ size standard definition files that you can choose from to analyze fragments run on ABI PRISM® genetic analyzers.

The following table lists the ready-to-use size standards stored in the GeneMapper ID database.

If using size standard...	Select this...
GeneScan™ 120 LIZ®	GS120LIZ
GeneScan™ 400HD	GS400HD
GeneScan™ 500	GS500
GeneScan™ 500(-250)	GS500(-250)
GeneScan™ 500 LIZ®	GS500LIZ
GeneScan™ 500(-250) LIZ®	GS500(-250)LIZ

Note: The GeneScan™ 350 size standard is not automatically stored in the database; however, it is provided in the **GeneMapper > Size Standards** folder for import.

GeneScan 120 Size Standard

About This Size Standard

The GeneScan™ 120 Size Standard is useful for sizing fragments between 15 and 120 base pairs. It is used when analyzing SNaPshot® kit samples.

How It Is Prepared

All aspects of the preparation of the GeneScan-120 LIZ size standard are proprietary. Each fragment contains a single LIZ fluorophore.

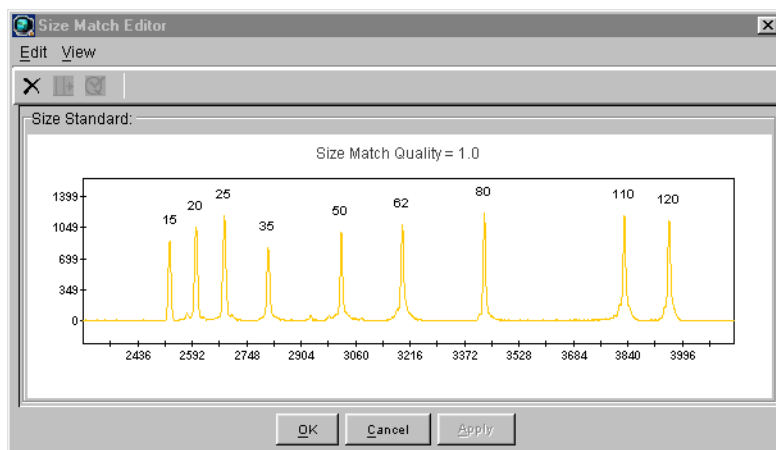
GeneScan 120 Molecular Lengths

The following table lists the GeneScan-120 LIZ denatured molecular fragment lengths (nucleotides) for the nine fragments.

Fragment Length	Fragment Length
15	62
20	80
25	110
35	120
50	

Electropherogram of GeneScan 120

The following screen shows an electropherogram of GeneScan-120 LIZ run under denaturing conditions.



GeneScan 350 Size Standard

About This Size Standard

The GeneScan™ 350 Size Standard is useful for sizing fragments between 35 and 350 base pairs. The native fragments are uniformly spaced to provide accurate size calling.

How It Is Prepared

The GeneScan 350 Size Standard is prepared by *Pst*I digestion of plasmid DNA, followed by ligation of a ROX™ dye-labeled 22-mer oligodeoxynucleotide to the cut ends. A subsequent enzymatic digestion with *Bst*UI yields DNA fragments containing a single ROX dye (See “[GeneScan 350 Molecular Lengths](#)” below).

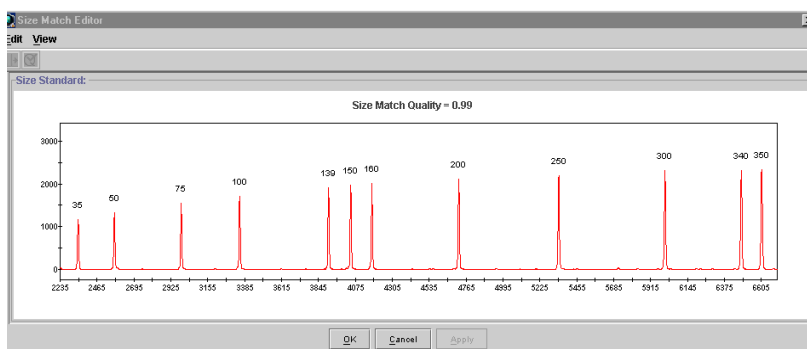
GeneScan 350 Molecular Lengths

The following table lists the GeneScan 350 Denatured Fragment Molecular Lengths (Nucleotides) for the 12 fragments.

Fragment Length	Fragment Length	Fragment Length
35	139	250
50	150	300
75	160	340
100	200	350

Electropherogram of GeneScan 350

The following screen shows an electropherogram of GeneScan 350 run under denaturing conditions.



GeneScan 400HD Size Standard

About This Size Standard The GeneScan™ 400HD (High Density) Size Standard is used to determine fragment lengths between 50 and 400 base pairs.

Special Uses The high density of marker bands in this standard makes it particularly useful for microsatellite analysis. All fragments have been checked for migration that is true to size under a wide variety of run conditions on all ABI PRISM® instruments.

How It Is Prepared All aspects of the preparation of the GeneScan 400HD Size Standard are proprietary. Each fragment contains a single ROX fluorophore.

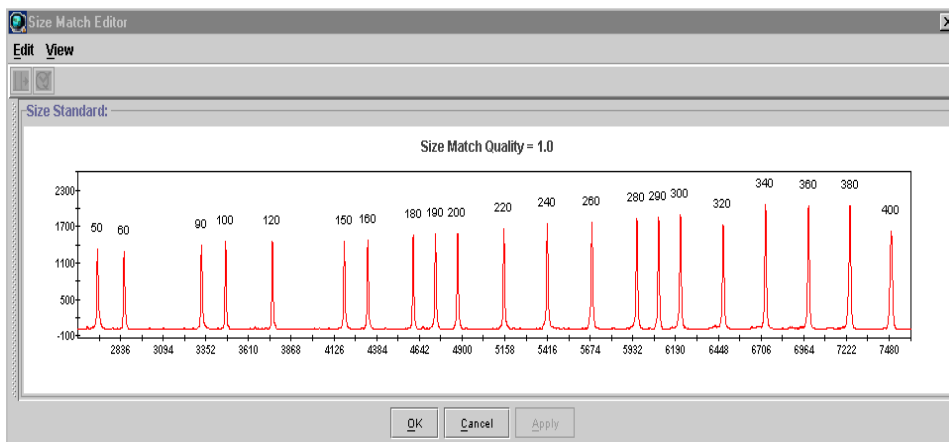
Fragment Lengths The following table lists the lengths of the 21 fragments that make up the GeneScan 400HD Size Standard.

Fragment Length	Fragment Length	Fragment Length
50	180	290
60	190	300
90	200	320
100	220	340
120	240	360
150	260	380
160	280	400

Denaturing Fragments Although the GeneScan 400HD Size Standard is made of double-stranded DNA fragments, only one of the strands is labeled. Consequently, even if the two strands migrate at different rates under denaturing conditions you will not need to worry about peak splitting.

Electropherogram of GeneScan 400HD

The following screen shows an electropherogram of GeneScan 400HD.



GeneScan 500 and GeneScan 500(-250) Size Standards

About These Size Standards

The GeneScan™ 500, the GeneScan™ 500 LIZ®, the GeneScan™ 500(-250), and the GeneScan™ 500(-250) LIZ® Size Standards are useful for sizing fragments between 35 and 500 base pairs. The native fragments are uniformly spaced to provide accurate base calling.

The GeneScan 500(-250) size standard contains all of the fragments in the GeneScan GS 500 Size Standard except for the 250 bp fragment. Since it is identical in other respects, it is not discussed separately in this section.

How It Is Prepared

The GeneScan 500 all Size Standard is prepared by *Pst*I digestion of plasmid DNA, followed by ligation of a LIZ or ROX dye-labeled 22-mer oligodeoxynucleotide to the cut ends. A subsequent enzymatic digestion with *Bst*UI yields DNA fragments containing a single ROX or LIZ dye (see [“GeneScan 500 Molecular Lengths”](#) below).

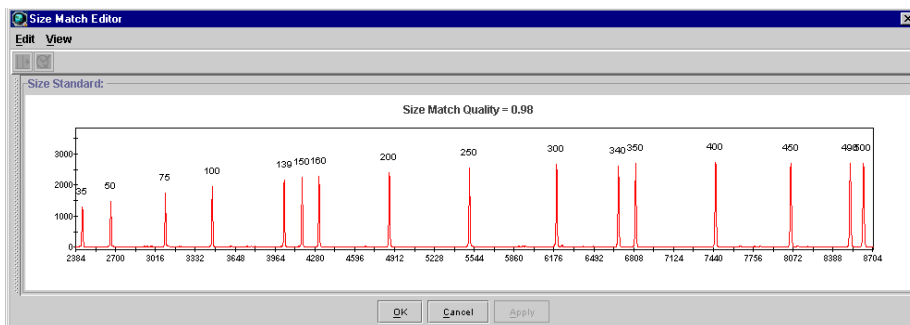
GeneScan 500 Molecular Lengths

The following table lists the GeneScan 500 denatured fragment molecular lengths (nucleotides) for the 16 fragments.

Fragment Length	Fragment Length	Fragment Length
35	160	400
50	200	450
75	250	490
100	300	500
139	340	
150	350	

Electropherogram of GeneScan 500

The screen below shows an electropherogram of GeneScan 500 size standard run under denaturing conditions.



Converting Macintosh Sample Files

E

This appendix describes how to use sample file conversion programs to prepare Macintosh® computer-generated fragment analysis sample files for transfer to a Microsoft® Windows®-based format and vice versa.

This appendix covers:

Converting Macintosh Sample Files E-2

Converting Macintosh Sample Files

About Converting Sample Files

Applied Biosystems created two conversion programs that prepare sample files for transfer from a Macintosh computer to computers running Microsoft Windows NT operating systems, and vice versa. These sample file conversion programs run only on a Macintosh computer.

The sample file conversion programs do not perform the file transfer from computer to computer. They set attributes of the files so that they can be used on the destination computer. For example, when transferring a fragment analysis sample file from a Macintosh computer to a computer running the Windows operating system, a file extension is required and the conversion program adds *.fsa* to the sample file name. For more detailed information on how these conversion programs function, refer to the SimpleText file entitled “About Conversion Programs” located in the same folder as the sample file conversion programs.

Installing Conversion Programs

To install the sample file conversion programs on a Macintosh computer:

1. Insert the GeneMapper *ID* software CD-ROM into your Macintosh computer’s CD-ROM drive.
An icon displays for the CD-ROM on the right-hand side of the screen.
2. Double-click the CD-ROM icon.
A CD-ROM window displays containing files and folders.
3. Double-click the *CONVFOLD* folder.
Inside this folder are two files, *CONVPROG.HQX* and *README.TXT*, which contain the installation instructions.
4. Copy the *CONVPROG.HQX* file to your local hard drive by click-drag the file over to the local hard drive icon, and releasing.
5. To decompress the *CONVPROG.HQX* file, drag and drop the file on the Stuffit Expander™ icon .

Note: You can download a free version of Stuffit Expander from <http://www.stuffit.com/expander>.

Note: Decompressing the *CONVPROG.HQX* file creates a *Conversion Programs* folder on the local hard drive. This folder contains the conversion programs and the SimpleText file “About Conversion Programs.” This file is a seven-page document that describes in detail how to use the conversion programs, why they are necessary, solutions to common problems, and possible alternative programs.

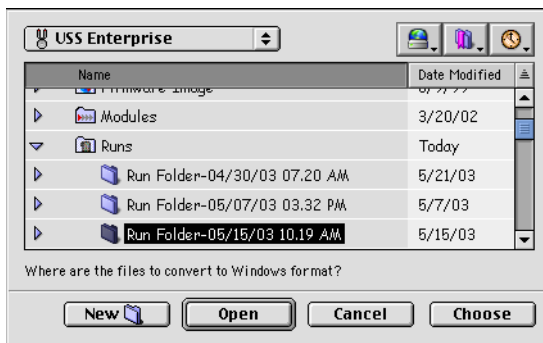
Converting Macintosh Sample Files to Microsoft Windows Files

To convert Macintosh computer sample files for use on a computer running Microsoft Windows operating system:

1. Double-click the  icon to start the program.

Sample File Mac to Win

The following dialog box opens.



Note: On Macintosh computers running operating system 8.0 or less, this dialog box has a different appearance. For more information, refer to the SimpleText file “About Conversion Programs” mentioned above in [“Installing Conversion Programs.”](#)

2. Using the triangle-shaped icons to the left of the folder names, navigate to the folder that contains the fragment analysis sample files you want to convert.
3. Select the folder.
4. Click **Choose**.

If there are no problems, the program performs the task and quits automatically. When you open the folder, the sample files have the file extension *.fsa*.

Note: To convert sample files created on a computer running the Microsoft Windows operating system for use on a Macintosh computer, follow steps 1-3 above; in Step 1, double-click the **Sample File Win to Mac** icon.

Transferring Data and Backing Up Data

F

This appendix covers:

Transferring Data	F-2
Backing Up Data and Cleaning the Database	F-7

Transferring Data

If you want to view/re-analyze a project created on a different copy of GeneMapper/computer, you have two options:

- Login to the GeneMapper database where the project is saved from a networked computer with GeneMapper installed. Refer to Chapter 2 (New Host Option) for more details on logging into a host computer.
- Export the project from GeneMapper Manager (Projects Tab) window on the computer it was created on, and import it into the GeneMapper Manager on the computer you wish to view it on.

IMPORTANT! If you want to open a project that was created on a different computer by exporting and importing, there are several things you must keep in mind and several components linked to the project that need to be in place in order to have full access to the information contained within the project. Therefore, follow the instructions below to ensure proper transfer of data.

Logging into Another GeneMapper Database from a Networked Computer

This will allow you have full access to the project. You will be able to re-analyze the sample files, apply new analysis settings, view all plots, raw data, EPT data, genotypes table and allele calls. However, you will not be able to save this project in your database. If you want this project saved in your database, you must export it and import it into your database manually.

Transferring Data from One Copy of GeneMapper to Another (Exporting/ Importing)

To view sample and genotype plots, and sample and genotype table, including all PQVs and allele calls:

1. Make sure the GeneMapper database you are importing the project into contains the same exact panels, bins, analysis method and size standard originally used to create the project. If this is not the case, export these items from the original copy of GeneMapper and import them into the GeneMapper copy on which you would like to view the data.

Note: This step must be done prior to importing the project.

2. Export the project from the original GeneMapper database and import it into another GeneMapper database of choice. These operations are done within the GeneMapper Manager window.

3. Open the imported project from the File menu in the main project window. You now have access to your data and genotypes. You can also make allele edits. Please note that you will not have access to the raw or EPT data, nor will you be able to reanalyze the data. Therefore, if you make a marker or bin modification, you can save these changes, but you will not be able to reanalyze using those changes. Refer to the next procedure to gain access the raw and EPT data, and for reanalysis capabilities.

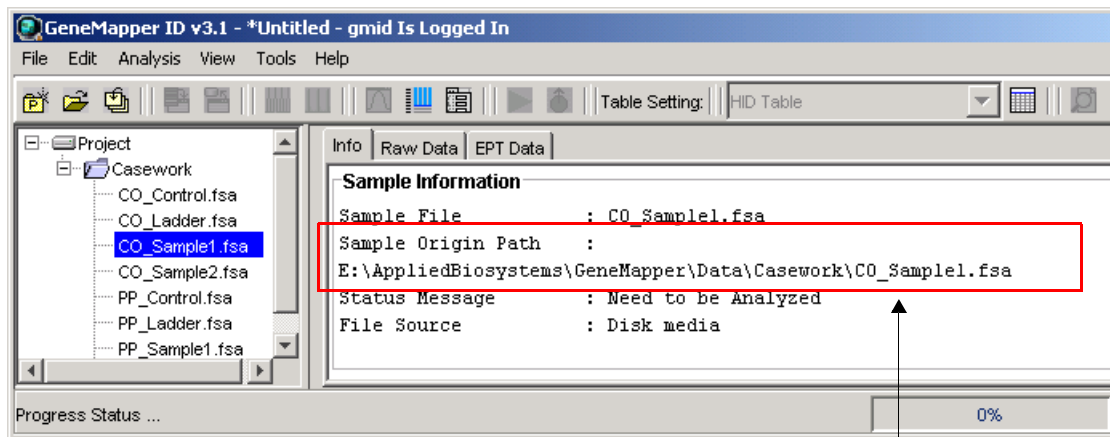
To view raw data, EPT data, and to reanalyze imported projects (in addition to the above):

Note: When you first analyze a set of samples and create a project, a link is formed between the sample file's original location and the information contained within the GeneMapper project. If you move the data from its original location, this link is broken and the software will no longer be able to access the raw data to perform reanalysis. If you want to reanalyze a project with a different copy of GeneMapper, gain access to the raw and EPT data, or if you have moved the data from its original location, you must re-establish the link. Not only will re-establishing the link allow you to reanalyze the project, but it will also allow you to view the raw and EPT data. Follow the steps below to re-establish the link.

1. Make sure the GeneMapper database you are importing the project into contains the same exact panels, bins, analysis method and size standard originally used to create the project. If this is not the case, export these items from the original copy of GeneMapper and import them into the GeneMapper copy you would like to view the data on.

Note: This step must be done prior to importing the project

2. Export the project from the original GeneMapper database and import it into another GeneMapper database of choice. These operations are done within the GeneMapper Manager window.
3. Open the project from the File menu.
4. View the Sample Info to determine the path between the project and the original sample file.

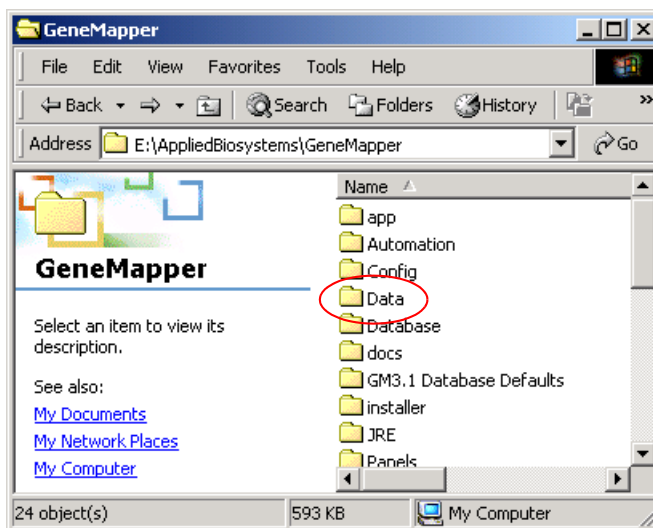


Pathway to the samples used in this project

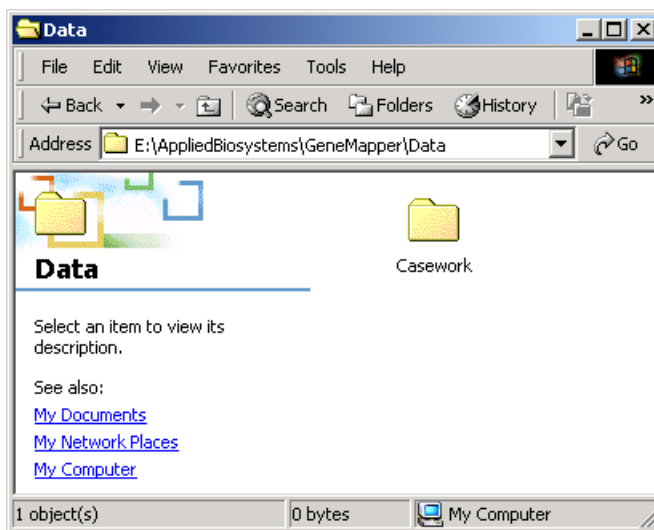
5. Create or re-create this path. You must use the exact same file names in order to successfully establish the link.

In the example above, you need to create the exact same folders (Data and Casework) within
E:\AppliedBiosystems\GeneMapper.

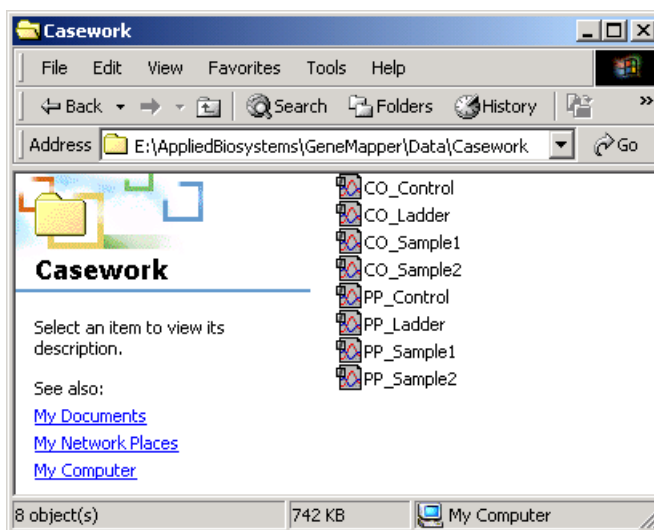
- a. In the GeneMapper folder, create a new folder titled **Data**.



- b. In the Data folder, create a new folder titled **Casework**.



- c. Transfer the original sample files (.fsa files) generated by Data Collection software into the appropriate folder, in this case it is the Casework folder.



To view raw data, EPT data, and re-analyze imported projects without establishing the link:

1. Make sure the GeneMapper database you are importing the project into contains the same exact panels, bins, analysis method and size standard originally used to create the project. If this is not the case, export these items from the original copy of GeneMapper and import them into the GeneMapper copy you would like to view the data on.
2. Import the sample files (.fsa files) into a blank project.
3. Select the appropriate analysis settings and analyze the project.

Backing Up Data and Cleaning the Database

Backing Up Data The GeneMapper database stores all projects (not including sample files), size standard definitions, analysis methods, plot and table settings, panels, bins, and matrices. Therefore, it is recommended that you back these components up in case of a computer failure or crash. In order to do this, you must export each component individually through the GeneMapper Manager and the Panel Manager. By exporting the components, they are not deleted from the database. In order to delete the items, you must select them within the GeneMapper Manager or Panel Manager and select delete.

Cleaning the GeneMapper Database The GeneMapper software may potentially perform slower as the GeneMapper database fills ups. Therefore, it is recommended that you back up all of your projects and delete them from the database periodically. Keep in mind that when you export a project, the sample files are *not* linked. Therefore, if you need to re-import the project at a later date, you will need to follow the instructions above in the transferring data section.

Sample Sheets and Plate Records

G

The appendix covers:

Information from the Data Collection Sample Sheet
or Plate Record G-2

Information from the Data Collection Sample Sheet or Plate Record

Introduction The GeneMapper *ID* software uses the sample sheet output from the ABI PRISM Data Collection instrument to identify the lane number and contents of each sample during analysis. This section describes the sample sheet information used by the GeneMapper *ID* software so that you can correctly set up the sample sheet.

For detailed information on how to complete a sample sheet correctly, see the user guide for the appropriate ABI PRISM instrument.

Transfer of Information Data is transferred from a sample sheet into GeneMapper *ID* software as described in the following table.

Sample Sheet	GeneMapper <i>ID</i> Software
Sample file name and Sample name Note: These names are the same between a sample sheet and GeneMapper <i>ID</i> software.	Sample file name and Sample name
Sample Info or Color Info	Sample type (Control, Allelic Ladder, Sample, Negative Control)
Color comment	Comment (if selected in the Samples View table)
Comment after Panel name. Use the pipebar () to separate text into user defined columns.	User Defined Columns 1-3

Using Sample Sheet Information The sample sheet information is essential for associating the nature of sample fragments with individual dye/lanes and tables in the GeneMapper *ID* software.

For example, the following figures show how the GeneMapper *ID* software incorporates information entered in the Sample Name, Color Info, and the Color Comment fields of a sample sheet into GeneMapper *ID* software Project tables. It is your responsibility to ensure that the entries on the sample sheet for your particular ABI PRISM instrument are correctly set up to provide GeneMapper *ID* software with the necessary information.

Sample Sheet

Plate Editor

File Edit

Plate Name: Profiler_Plus

Well	Sample Name	Dyes	Color Info	Color Comment	Project Name	Dy...	Run Module 1	Analysis Module
A1	Sample1	B	Sample1		3100_Project1	F	GeneScan36_POP4_Fmodule	GS500Analysis.gsp
		G	Sample1					
		Y	Sample1					
		R	Sample1	Size Standard				
		O	Sample1					
B1	Sample2	B	Sample2		3100_Project1	F	GeneScan36_POP4_Fmodule	GS500Analysis.gsp
		G	Sample2					
		Y	Sample2					
		R	Sample2	Size Standard				
		O	Sample2					
C1	Sample3	B	Sample3		3100_Project1	F	GeneScan36_POP4_Fmodule	GS500Analysis.gsp
		G	Sample3					
		Y	Sample3					
		R	Sample3	Size Standard				
		O	Sample3					
D1	Sample4	B	Sample4		3100_Project1	F	GeneScan36_POP4_Fmodule	GS500Analysis.gsp
		G	Sample4					
		Y	Sample4					
		R	Sample4	Size Standard				
		O	Sample4					
E1	Sample5	B	Sample5		3100_Project1	F	GeneScan36_POP4_Fmodule	GS500Analysis.gsp
		G	Sample5					
		Y	Sample5					
		R	Sample5	Size Standard				
		O	Sample5					

Comments:

OK

GeneMapper ID Software Project Table

GeneMapper ID v3.1 - "Untitled"

File Edit Analysis View Tools Help

Table Setting: HID Table

Status	Sample Name	Sample Type	Specimen Category	Analysis Method	Panel	Size Standard
1	sample1	Sample	no export	HID_Advanced	Profiler_Plus_v1	Profiler Plus st
2	sample2	Sample	no export	HID_Advanced	Profiler_Plus_v1	Profiler Plus st
3	sample3	Sample	no export	HID_Advanced	Profiler_Plus_v1	Profiler Plus st
4	sample4	Sample	no export	HID_Advanced	Profiler_Plus_v1	Profiler Plus st
5	sample5	Sample	no export	HID_Advanced	Profiler_Plus_v1	Profiler Plus st

User defined

Read from sample file

Find Command You can increase the utility of the Find (Ctrl+F) command in GeneMapper *ID* software by carefully planning the format of the information you put into the Sample Info or Color Info field of the sample sheet.

Example of How To Use the Find Command

One Method	
If...	Then...
you have 12 samples, numbered 1, 2, 3,..., 12, and you enter these numbers into the Sample Info field	when you search for all dye/lanes containing a 1 in the Sample Info field, not only will you select sample 1, you will also get samples 10, 11, and 12.
A Better Plan	
If...	Then...
you number the samples 01, 02, 03, and so on	a search for the text 01 would select only the desired dye/lanes.

Troubleshooting Sheet

H

This appendix covers:

Troubleshooting Sheets H-2

Troubleshooting Sheets

Due to the complex nature of troubleshooting, these following matrices are provided as a tool to collate the variables involved in the troubleshooting process with support groups, product groups and the software development team at Applied Biosystems. To address any issues that may arise when installing or running the software, please fill out the appropriate list of questions, and collect the necessary files before contacting Technical Support.

For Installation Issues

Collect the following files and complete the questions below:

- install.log (X:\AppliedBiosystems\GeneMapper)
- GeneMapper_Log.txt (X:\AppliedBiosystems\GeneMapper)

Does the computer and operating system meet the required computer specifications? List any potential discrepancies.
Did you receive any error messages or alerts? If so, what did they say (if possible, take screen captures)? At what point during the installation process did these errors/alerts occur?
How many installation attempts have you tried?
Are you logged in to the local computer?
Is the TCP/IP configured?
Do you have administrative privileges?
Is this a stand-alone computer, networked, or attached to an ABI P _S Instrument?

For Software Related Issues

Does the computer and operating system meet the required computer specifications?
List any potential discrepancies.

Problem description:

Did you receive any error messages or alerts? If so, what did they say (if possible, take screen captures)? At what point did these errors/alerts occur?

Is the problem reproducible?

Steps taken prior to encountering or to reproduce the problem:

- 1.
- 2.
- 3.
- 4.
- 5.

For Low Quality Analysis

Collect the following files and complete the questions below:

1. Sample files (.fsa files imported into GeneMapper ID)
2. Analysis Method (export of GeneMapper Manager)
3. Size Standard Definition, if different than default (export out of GeneMapper Manager)
4. Matrix File (310/377 Only, export out of GeneMapper Manager)
5. Panels and Binset if different than default (export out of Panel Manager)

Does the computer and operating system meet the required computer specifications?
List any potential discrepancies.

Problem description:

Did you receive any error messages or alerts? If so, what did they say (if possible, take screen captures)? At what point during analysis did these errors/alerts occur?

Is the problem reproducible?

Steps taken prior to encountering or to reproduce the problem:	
1.	
2.	
3.	
4.	
5.	
Size standard (default or custom)	
Peak detector mode (classic or advanced)	
Instrument model	
Dye Set (F or G5)	
Run module	

GeneMapper *ID* Software Verification



This appendix covers:

Summary of GeneMapper <i>ID</i> Software Verification.	I-2
AmpF ~~STR~~ STR Kits, Samples, Instrumentation, Hardware, and Software Used for Verification Testing	I-4
Results of Verification Testing	I-7
Conclusion.	I-39
References	I-40

Summary of GeneMapper ID Software Verification

GeneMapper™ ID Software v3.1 verification was performed to provide documentation to confirm the functionality of the software features. The test plan, which Applied Biosystems has defined as verification of the software, was designed to develop a set of criteria to evaluate the performance of this software for the human identification communities.

The verification was designed to evaluate four specific categories:

- Peak detection and genotyping
- Algorithm testing
- Workflow
- Data handling

These categories cover 24 test parameters that evaluate the software's robustness, performance, and features. The test parameters were performed using forensic-simulated samples amplified with the AmpFℓSTR® PCR Amplification kits and electrophoresed on defined ABI PRISM instrument platforms. These test parameters evaluate, for example, peak detection and the non-concordance of alleles; out-of-bin alleles, peak height ratios, low peak heights; the ability to process samples per color, per sample, and per marker; and import and export functionality.

GeneMapper ID Software v3.1 was specifically designed as a complete integrated software package for forensic, paternity, and databasing laboratories performing STR analysis. Currently, human identification laboratories worldwide use ABI PRISM® GeneScan® Software and ABI PRISM® Genotyper® Software as the standard for software analysis. GeneMapper ID software combines the functionality of these two software programs into one integrated software package. Among other things, this software analyzes the raw data collected from ABI PRISM instrument platforms and automatically identifies peaks, quantifies signal intensity, sizes each DNA fragment, and makes allele calls using defined panels and bin sets with its automated genotyping capabilities (Leibelt *et al.*, 2003).

Further, a comprehensive concordance study used forensic-simulated samples generated with seven different AmpFℓSTR kits. The data were processed on six instrument/computer platforms (ABI PRISM® 310 Genetic Analyzer for Apple® Macintosh® and Microsoft® Windows® NT operating system [OS], ABI PRISM® 377 DNA Sequencer for Macintosh and Windows NT OS, ABI PRISM® 3100

Genetic Analyzer, and ABI PRISM® 3100-*Avant* Genetic Analyzer). These data were analyzed with GeneScan Software v3.7.1 and Genotyper Software v3.7 (Windows NT OS), and GeneScan Software v3.1.2 and Genotyper Software v2.5.2 (Macintosh OS) (Boland *et al.* 2003) and these data have been used for the verification of GeneMapper ID Software v3.1.

Peak Detection Algorithms

GeneMapper ID software includes three peak detector algorithms, or modes, allowing different levels of user control over data analysis.

Classic Mode

The Classic mode provides the user with the same analysis parameters and produces results similar to those analyzed with GeneScan Software v3.1.2 (Macintosh OS). This algorithm aids the user in the adoption of GeneMapper ID software for laboratories that currently use the Macintosh OS and want to maintain current analysis parameter settings.

Advanced Mode

The Advanced mode provides the user with the same analysis parameters available in GeneScan Software v3.7.1 (Windows NT OS).

Basic Mode

The Basic mode is not recommended for human identification applications because it allows only for analysis using limited parameters consisting of a user-defined minimum peak height threshold.

AmpF ℓ STR Kits, Samples, Instrumentation, Hardware, and Software Used for Verification Testing

AmpF ℓ STR Kits Used The following AmpF ℓ STR PCR Amplification kits were used to amplify the samples used in the verification testing:

- 4-Dye Kits
 - COfiler[®]
 - Green[®] I
 - Profiler[®]
 - Profiler Plus[®]
 - SGM Plus[®]
- 5-Dye Kits
 - Identifiler[®]
 - SEfiler[™]

Sample Types Used [Table I-1](#) lists the DNA sample type, the total number of samples amplified, and the total number of alleles available for evaluation and comparison for the verification of GeneMapper *ID* software.

Table I-1 Samples used for software verification

DNA Sample Type	Number of Samples Amplified	Number of Alleles for Verification
Single source	16	7422
Mixture	4	3678
Degraded	3	2402
Inhibited	3	1318
Low quantity	3	2772
High quantity	3	2722

Thirty-two human genomic DNA samples were quantified using the QuantiBlot® Human DNA Quantitation kit and amplified with the AmpFSTR kits.

Note: For each sample type, all AmpFSTR kits used the sample quantity of DNA, except the Identifiler kit. The sample quantity of DNA for the Identifiler kit is denoted in parenthesis.

- **Single Source DNA Samples** – Eight female and eight male DNA samples were amplified with 2 ng total input DNA (1 ng for the Identifiler kit).
- **Mixture Samples** – Three mixtures of male and female DNA were amplified per kit. These samples were mixed in defined ratios of 1 to 9, 1 to 1, and 9 to 1, respectively.
- **High Quantity DNA Samples** – Three DNA samples were amplified with 8 ng total input DNA (4 ng for the Identifiler kit).
- **Low Quantity DNA Samples** – Three DNA samples were amplified with 250 pg total input DNA (125 pg for the Identifiler kit).
- **Degraded DNA Samples** – Three DNA samples were degraded with 2 units DNase I for 15 minutes with subsequent amplifications using 4 ng/μL degraded DNA (2 ng/μL for the Identifiler kit).
- **Inhibited DNA Samples** – Three 2 ng/μL total input DNA samples were amplified in the presence of 16 μM hematin (1 ng/μL for the Identifiler kit).

The samples were amplified using an Applied Biosystems GeneAmp® 9700 PCR system and the recommended cycling conditions. The amplified samples were injected or loaded on three or more ABI PRISM instrument platforms.

Instrument Platform and Computer Operating System

Table I-2 lists the instrument platforms and computer operating systems that were used to electrophorese the samples for verification testing.

Table I-2 Instrument and computer platforms

Instrument	Computer Operating System
310 Genetic Analyzer	Apple Macintosh® v9.1
	Microsoft® Windows® NT
377 DNA Sequencer	Macintosh v9.1
	Microsoft Windows NT
3100 Genetic Analyzer	Microsoft Windows NT
3100- <i>Avant</i> Genetic Analyzer	

Analysis Software

The software packages and computer configurations used to analyze the data are listed below:

Table I-3 Software and computer configuration used for analysis

Analysis Software and Version	Computer Configuration	Color Printer
GeneScan® Software v3.1.2	Macintosh® G3, 128 MB RAM, v9.1 operating system	HP LaserJet 4500N
Genotyper® Software v2.5.2		
GeneScan® Software v3.7.1	Dell Latitude C810 with a 1.1GHz processor, 512 MB RAM, Microsoft® Windows® 2000 operating system, Service Pack 3	
Genotyper® Software v3.7		
GeneMapper <i>ID</i> Software v3.1 (Classic and Advanced modes)		

Results of Verification Testing

The verification was designed to evaluate four specific categories:

- Peak detection and genotyping
- Algorithm testing
- Workflow
- Data handling

Peak Detection and Genotyping

Test Parameters The test parameters for the peak detection and genotyping category are described below:

1. Peak detection evaluation comparing the analysis results from GeneScan software (Macintosh OS) to GeneMapper *ID* software using the Classic algorithm, and GeneScan software (Windows NT OS) to GeneMapper *ID* software using the Advanced algorithm.
2. Genotyping evaluation comparing the analysis results from GeneScan software (Macintosh OS) to GeneMapper *ID* software using the Classic algorithm, and GeneScan software (Windows NT OS) to GeneMapper *ID* software using the Advanced algorithm.
3. Editing functionality test.
4. Locus concordance between duplicate amplifications of the same sample, and between AmpF ℓ STR kits.
5. Matrix and multi-component test using Classic and Advanced algorithms.
6. Size standard and size match editor using Classic and Advanced algorithms.

Peak Detection (Peak Height) Evaluation The first parameter evaluated is the comparison of peak height differences between GeneScan Software v3.1.2 (Macintosh OS) to GeneMapper *ID* Classic peak detection algorithm, and GeneScan Software v3.7.1 (Windows NT OS) to GeneMapper *ID* using the Advanced peak detection algorithm using a subset of the samples described in [Table I-1 on page I-4](#).

Table I-4 displays the results for the combined peak height differences in relative fluorescent units (RFU) for single source DNA samples and low quantity DNA samples for a total of 3483 alleles.

Table I-4 Peak detection evaluation

AmpF/STR PCR Amplification Kit	Instrument Platform	Mode Used	Number of Alleles	Range of Peak Height Difference (RFU)	Average Peak Height Difference (RFU)	Percent of Maximum Peak Height Difference
Profiler Plus and COfiler	310 Genetic Analyzer (Macintosh Operating System)	Classic	610	0-15	0.13	0.3
Identifiler			551	0-8	0.26	0.4
Profiler Plus and COfiler	310 Genetic Analyzer (NT Operating System)	Advanced	610	0-16	5.71	2.6
Identifiler			551	0-20	4.61	1.1
Profiler Plus and COfiler	3100 Genetic Analyzer		610	0-11	3.69	0.6
Identifiler			551	0-16	3.34	1.1

The GeneMapper *ID* Classic mode was designed to produce similar results to GeneScan Software v. 3.1.2 (Macintosh OS) so a laboratory could maintain its current analysis parameter settings. The average peak height difference measured is less than 1 RFU for the Classic mode, a very insignificant difference. Although a maximum difference of 15 RFU was observed in one allele, the maximum percent difference for this allele is only 0.3%. The maximum peak height difference measured in all samples evaluated is 20 RFU, observed in a single source DNA sample using the Advanced mode.

Peak Height Comparison Example Data

The same single source DNA sample was amplified with the AmpF Φ STR Identifiler PCR Amplification kit, processed on the 310 for Macintosh and NT OS, then analyzed with different versions of software to compare GeneScan for the Macintosh OS to the Classic mode in GeneMapper *ID* and to compare GeneScan for Windows NT OS to the Advanced mode in GeneMapper *ID* software. Note the similarities in peak heights measured in RFUs.

Table I-5 Peak height example data analyzed using different software

Software and Version	To view the data, see ...
GeneScan Software v3.1.2 (Macintosh OS) and Genotyper Software v2.5.2	Figure I-1 on page I-10
GeneMapper <i>ID</i> Software v3.1 in the Classic mode	Figure I-2 on page I-11
GeneScan Software v3.7.1 (Windows NT OS) and Genotyper v3.7	Figure I-3 on page I-12
GeneMapper <i>ID</i> Software v3.1 in the Advanced mode	Figure I-4 on page I-13

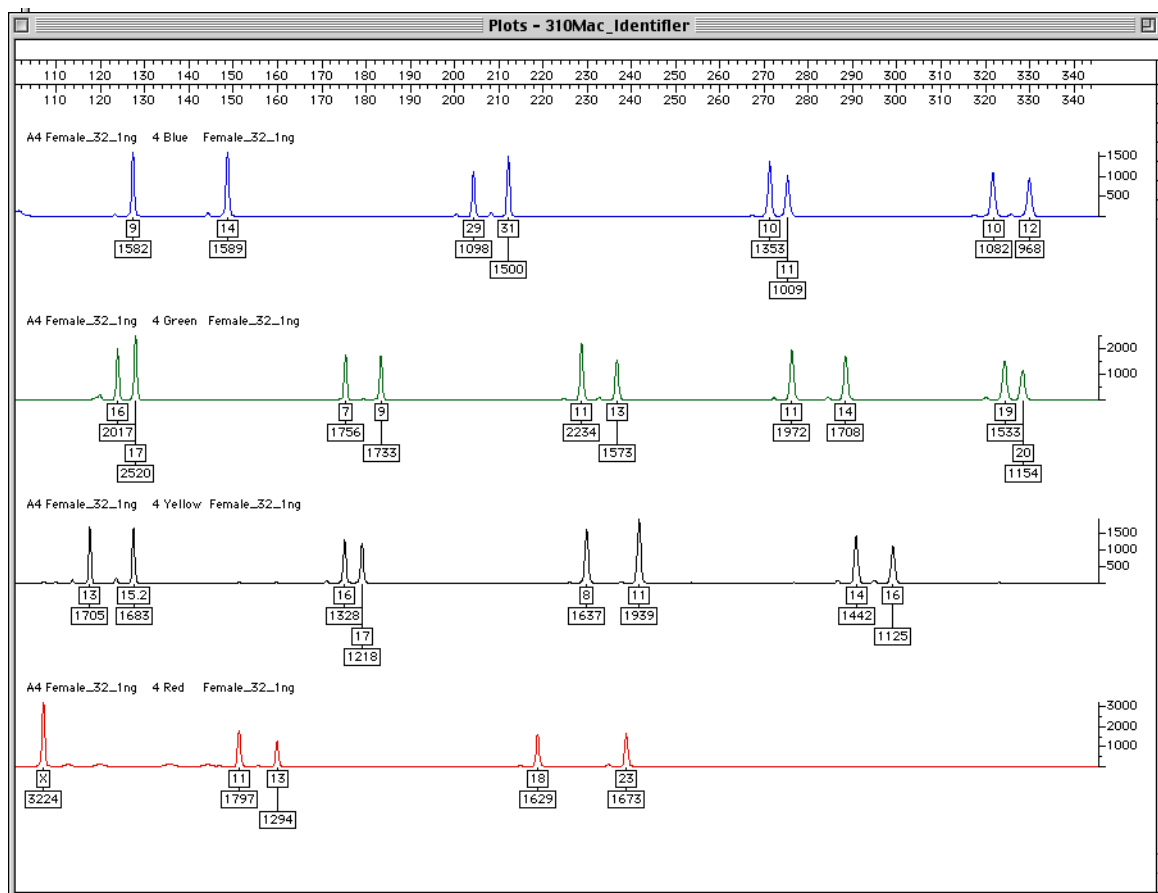


Figure I-1 Female single source DNA Sample amplified with the AmpF ϕ STR Identifier PCR Amplification kit, electrophoresed on a 310 Genetic Analyzer (Macintosh OS), then analyzed with GeneScan Software v3.1.2 and Genotyper v2.5.2.

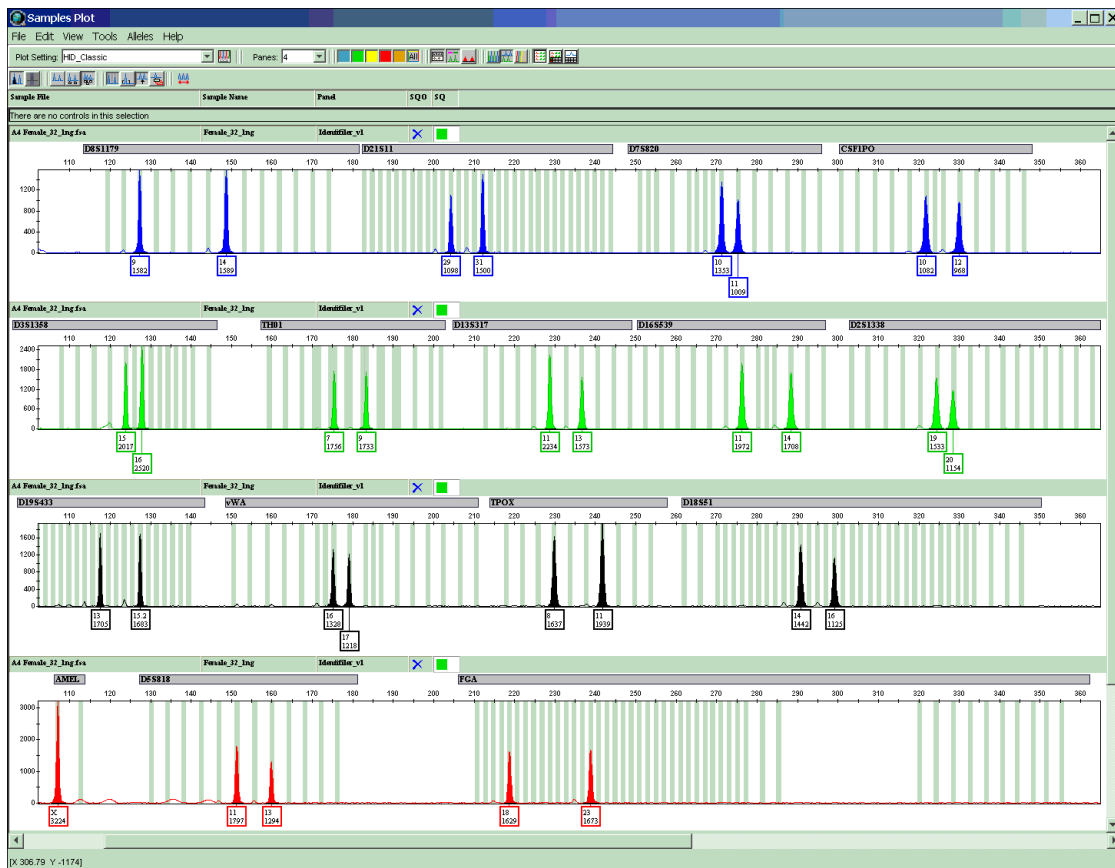


Figure I-2 Female single source DNA Sample amplified with the AmpF/STR Identifiler PCR Amplification kit, electrophoresed on an 310 Genetic Analyzer (Macintosh OS), then analyzed with GeneMapper ID software using the Classic algorithm.

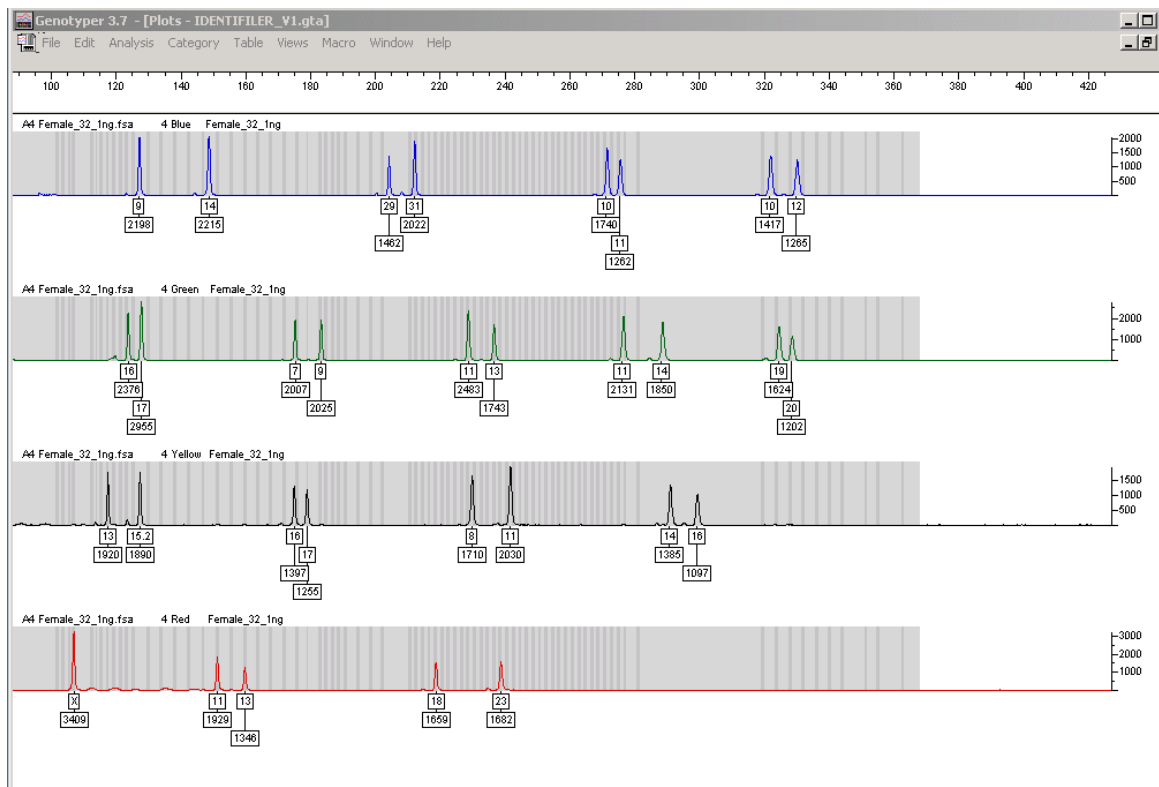


Figure I-3 Female single source DNA Sample amplified with the AmpF ϕ STR Identifiler PCR Amplification Kit, electrophoresed on an 310 Genetic Analyzer (Windows NT), then analyzed with GeneScan v3.7.1 software and Genotyper v3.7.

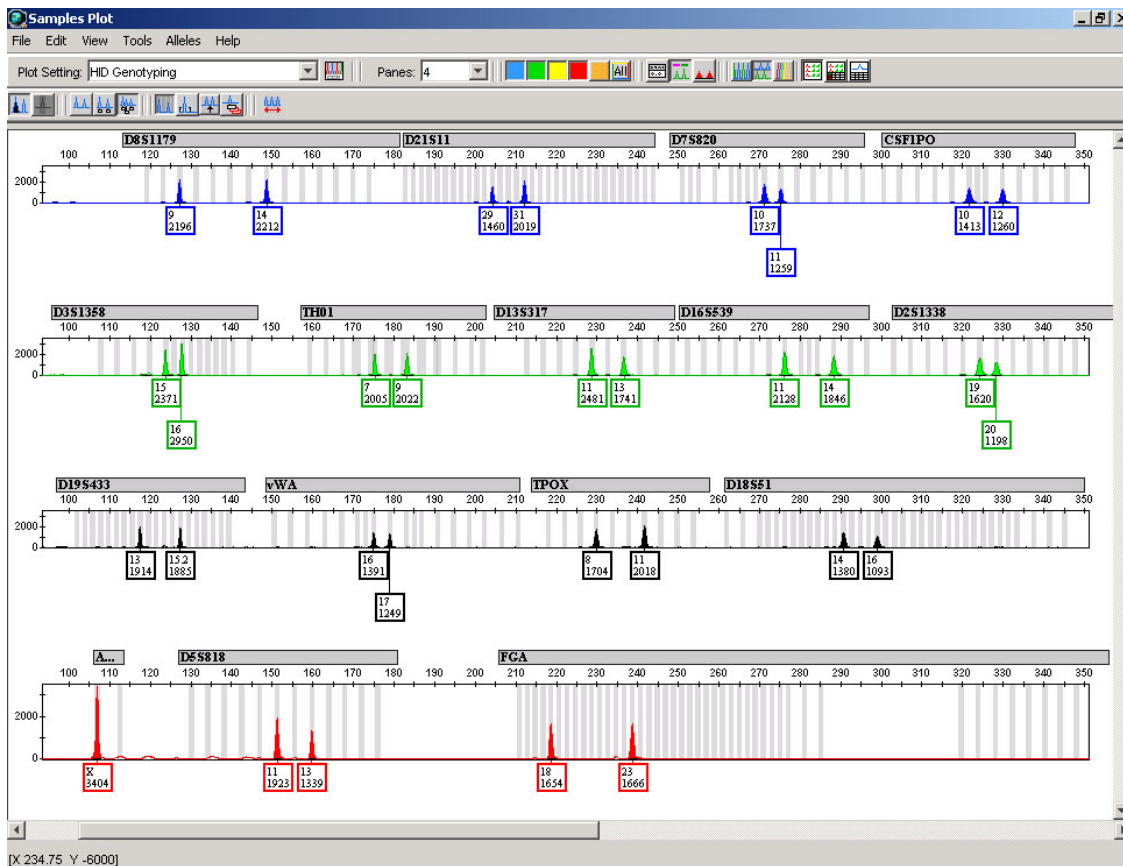


Figure I-4 Female single source DNA Sample amplified with the AmpF/STR Identifier PCR Amplification kit, electrophoresed on a 310 Genetic Analyzer for Windows NT OS, then analyzed with GeneMapper ID software using the Advanced algorithm.

Genotyping

The second parameter evaluated in the Peak Detection and Genotyping category is the genotyping evaluation comparing the results from data analysis with GeneScan Software v3.1.2 and Genotyper software v2.5.2 (Macintosh OS) to GeneMapper ID Software using the Classic algorithm, and GeneScan Software v3.7.1 and Genotyper software v3.7 (Windows NT OS) to GeneMapper ID Software using the Advanced algorithm. The analysis parameter settings for the software packages were defined according to Applied Biosystems recommendations (Applied Biosystems, 2002) with a

defined peak amplitude threshold (PAT) of 50 RFU. Five observations were documented when comparing the results from GeneScan and Genotyper software to the results of GeneMapper *ID* software after the trained analyst completed editing.

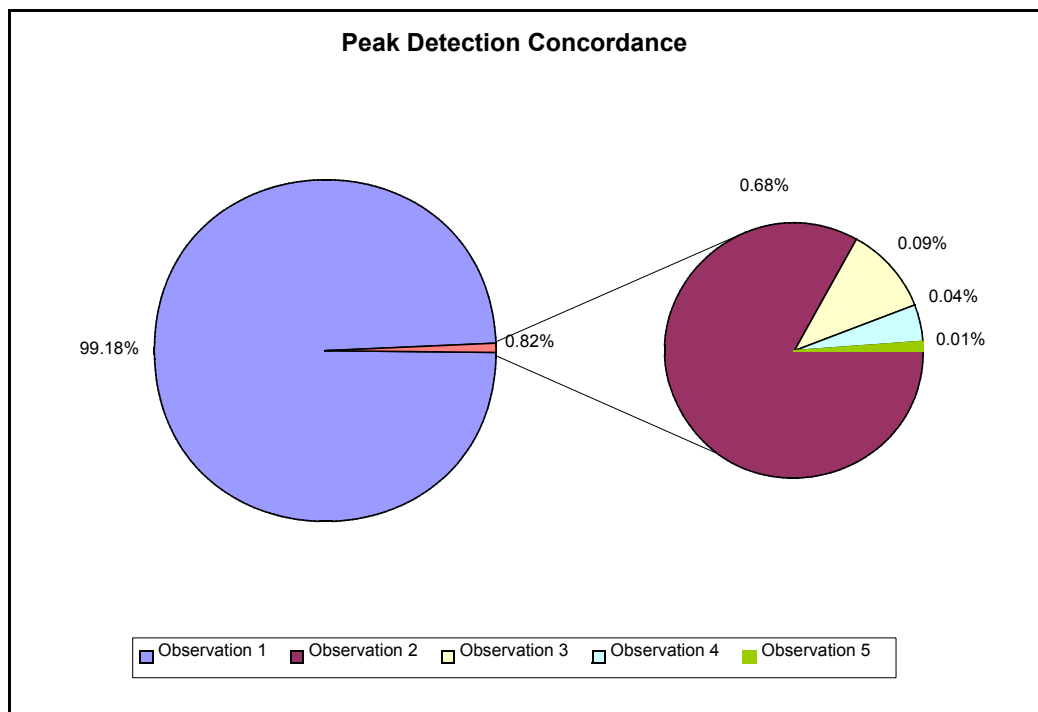


Figure I-5 Peak detection concordance observations

- **Observation 1**
99.18% (20,197/20,364 alleles) of the alleles compared are the same, whether analyzed with GeneScan and Genotyper software or GeneMapper *ID* Software v3.1.
Note: The remaining 0.82% of alleles are described in Observations 2 through 5.
- **Observation 2**
0.68% (139/20,364 alleles) of the alleles from low quantity, degraded, and inhibited DNA samples fell below the defined 50 RFU peak amplitude threshold setting in GeneMapper *ID* software Advanced mode. All of these alleles were analyzed to

be just below 50 RFU resulting in the peak not being called in GeneMapper *ID* software, whereas they were detected as 50 RFU or slightly greater in GeneScan software. As previously described, the GeneMapper *ID* software Advanced mode has an overall slightly lower peak height (2.6% or less) compared to GeneScan software v3.7.1.

- Observation 3

0.09% (18/20,364 alleles) of the alleles from one single source DNA sample at the D18S51 locus with the 12.2 allele are called in GeneMapper *ID* software, since the panels have been updated with known variants. The macros for Genotyper software have not been updated; Genotyper software calls these off-ladder alleles (or “OL Allele?”) and allows the end-user to edit the alleles accordingly. This example was chosen specifically for the off-ladder variant for verification purposes. This sample was used for both single source and mixture amplifications for all kits; therefore, it contributes to 18 observations of the 12.2 allele being called.

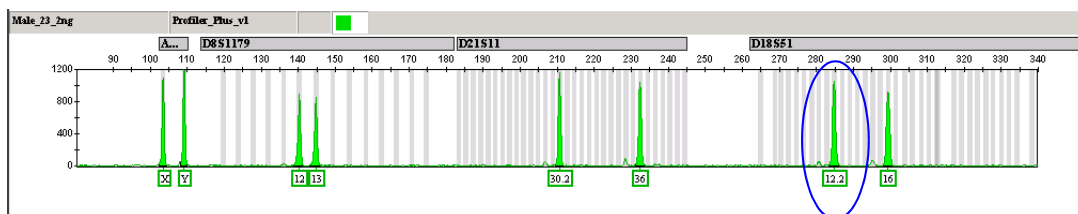


Figure I-6 GeneMapper *ID* software genotyped D18S51 locus with the 12.2 allele

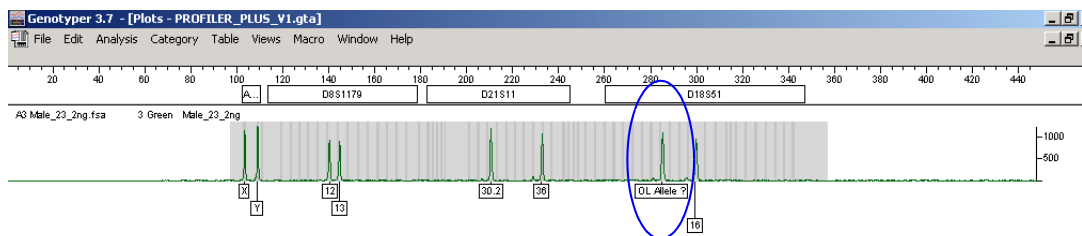


Figure I-7 Genotyper software genotyped D18S51 locus with the OL allele

- Observation 4

0.04% (8/20,364 alleles) of the alleles from the mixture data used in comparing GeneScan software v3.7.1 to GeneMapper *ID* software Advanced mode produced eight alleles where the minor contributor's alleles that are 4-bp smaller than the major contributor's alleles (*i.e.*, in the stutter position) are filtered, since the calculated ratio at these loci are just above (GeneScan software v3.7.1) or just below (GeneMapper *ID* software Advanced mode) the defined stutter ratio. Two examples are given below.

Platform and Ampf/str Kits	Software	Sample Type	Locus	Calculated Stutter Ratio		Macro /Panel Setting	
				Allele Peak Height (RFU)	Allele Peak Height (RFU)	% Stutter Calculated After Analysis	% Stutter Defined in Macro/Panel Setting
3100 Identifiler	Genotyper	1 to 9 mixture	D18S51	13/422	14/2472	17.07	17
	GeneMapper <i>ID</i>			<i>13/418^a</i>	14/2466	16.95	17
3100 Profiler Plus	Genotyper		TH01	6/72	7/1161	6.20	6
	GeneMapper <i>ID</i>			<i>6/67^a</i>	7/1157	5.79	6

a. The alleles and peak heights shown in italics were filtered out in GeneMapper *ID*.

- Observation 5

0.01% (2/20,364 alleles) of the alleles from offscale data analyzed with GeneScan software v3.7.1 and Genotyper software v3.7 (Windows NT OS) did not filter the actual stutter because of the limitations of the Genotyper software. Filtering compares Peak A to the next labeled peak, Peak B.

The label is removed from Peak A (the stutter peak) if Peak B (the true allele) meets two criteria:

- Peak B is higher than peak A by the specified percentage and
- Peak B is within the specified proximity size (in base pairs) range relative to peak A.

These two observations for verification did not meet these criteria due to an abundance of –A product, resulting in the detection of a third peak, Peak C. In these two observations, the filtering in Genotyper software compared Peak A to Peak C. The GeneMapper *ID* software appropriately filters the stutter and the shoulder peak using defined bin sets.

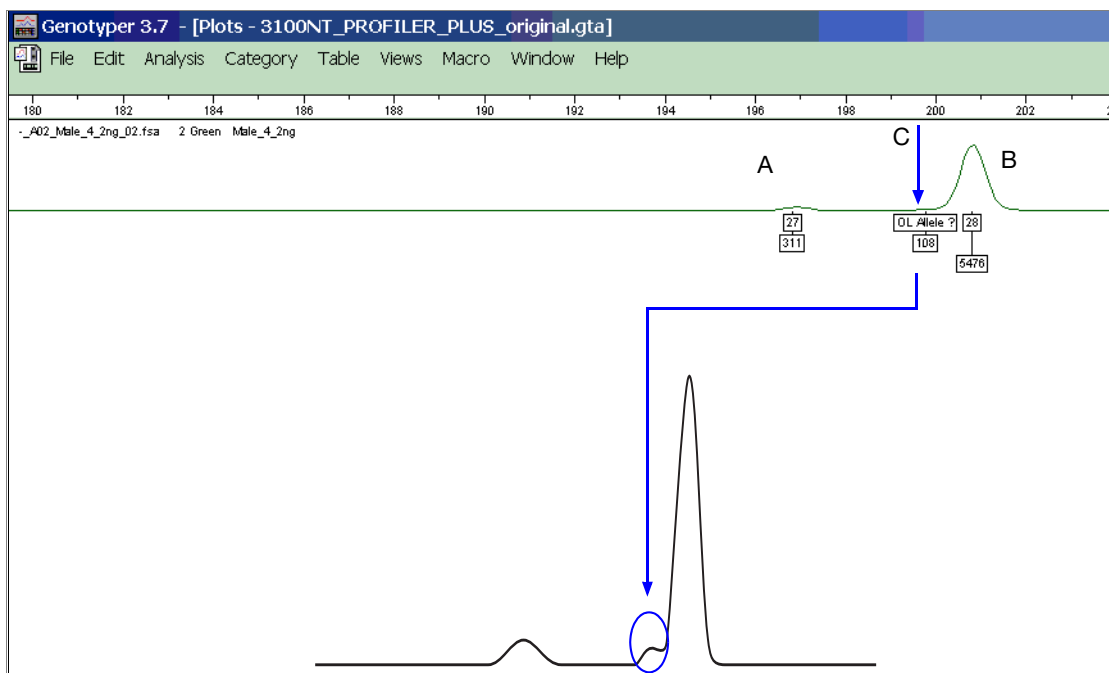


Figure I-8 Genotyper software v3.7

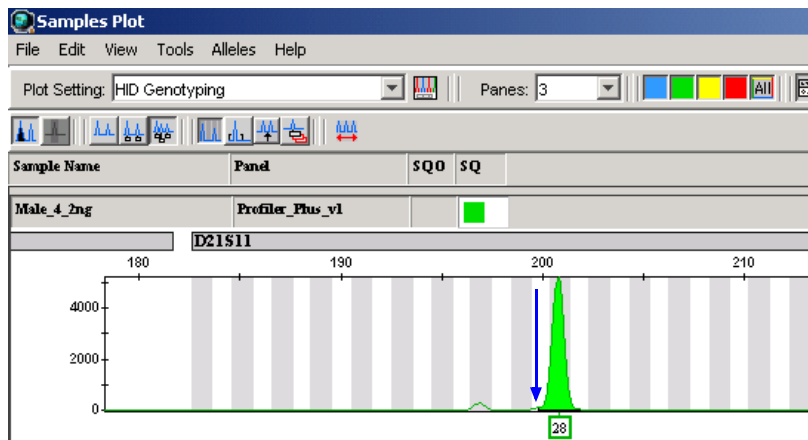


Figure I-9 GeneMapper™ ID Software v3.1 using the Advanced algorithm

Editing Functionality Test

The third parameter evaluated in the Peak Detection and Genotyping category is the editing functionality test. This parameter was evaluated with 500 alleles for the ability to change allele calls, view the history of the allele calls, and display the changed allele calls. These abilities functioned with a 100% success rate.

If multiple alleles have been edited, as shown in [Figures I-10](#), the history can be displayed for each edited peak by selecting the peak and displaying the history. If an allele is edited more than once, then only the last edit is displayed.

In [Figure I-10](#), one allele from each locus was “Removed” during the functionality test. Further, D8S1179 allele 12 was selected and its Allele History is displayed. In the Allele History table, GeneMapper ID software v3.1 “Created” this allele. In performing this functionality test for verification, the Allele History table states that this allele has been “Deleted.”

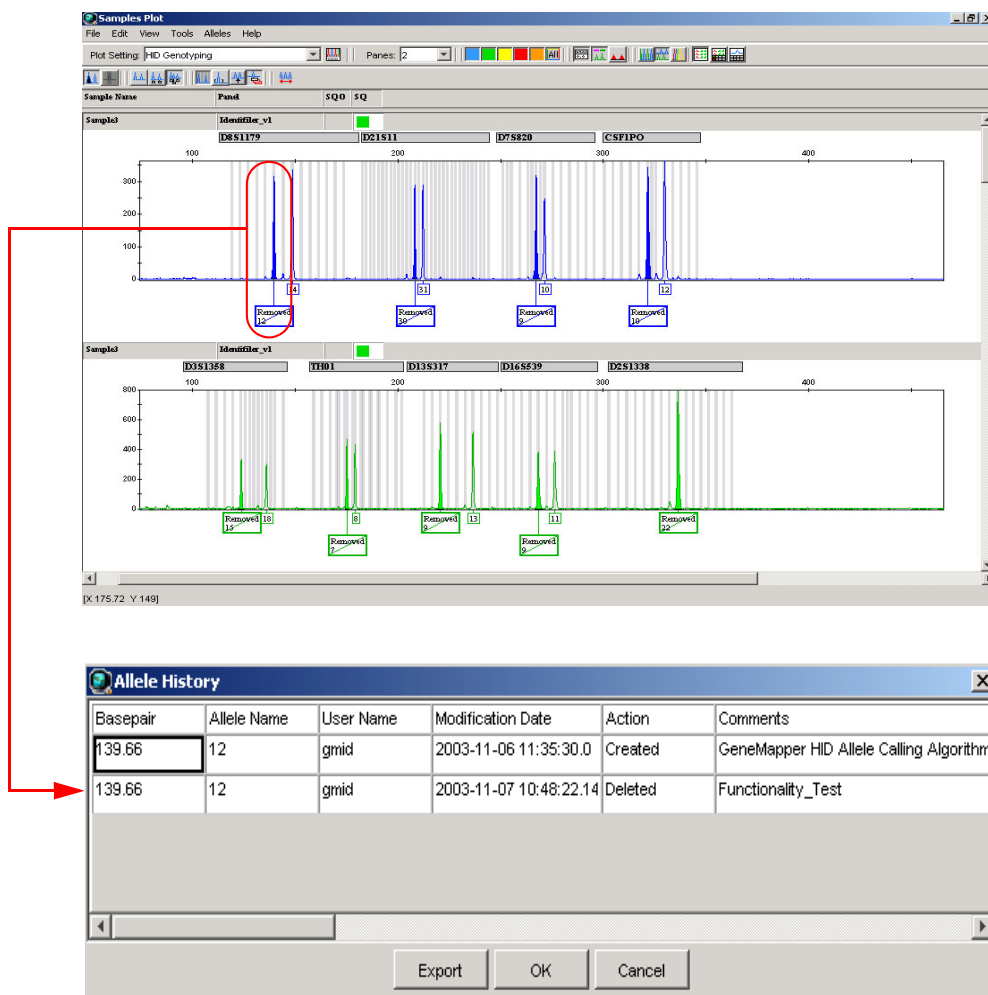


Figure I-10 Deleted Allele and the Allele History table

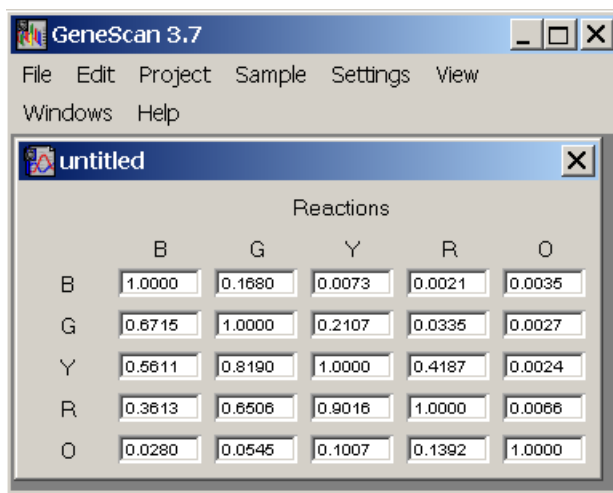
Locus Concordance Evaluation

The fourth parameter evaluated in the Peak Detection and Genotyping category is the locus concordance between duplicate amplifications of the same sample and locus concordance between the same sample using different AmpF Φ STR kits. The requirement is that samples must have the same name. Samples that have the same name are evaluated if the non-concordance feature is selected. 254

pairs were tested to verify the non-concordance feature in GeneMapper *ID* software using single source DNA samples, inhibited DNA samples, and degraded DNA samples. All pairs were accurately defined with GeneMapper *ID* non-concordance feature.

Matrix and Multi-component Test

The fifth parameter evaluated in the Peak Detection and Genotyping category is the matrix and multi-component test. The same DS-32 and DS-33 matrix sample files were electrophoresed on the 310 Genetic Analyzers (both Macintosh and Windows NT OS) and analyzed with both GeneScan software v3.7.1 (Windows NT OS) and GeneScan software v.3.1.2 (Macintosh OS). Then these same sample files were analyzed by the Classic and Advanced algorithms in the GeneMapper *ID* software. The mathematical matrices generated using the same scan data points, show identical values ([Figures I-11](#) and [I-12](#)).



The screenshot shows the GeneScan 3.7 software window with a menu bar (File, Edit, Project, Sample, Settings, View, Windows, Help) and a window titled 'untitled'. The main area displays a table of peak heights for five reactions (B, G, Y, R, O) across five lanes (B, G, Y, R, O). The values are as follows:

	B	G	Y	R	O
B	1.0000	0.1680	0.0073	0.0021	0.0035
G	0.6715	1.0000	0.2107	0.0335	0.0027
Y	0.5611	0.8190	1.0000	0.4187	0.0024
R	0.3613	0.6506	0.9016	1.0000	0.0066
O	0.0280	0.0545	0.1007	0.1392	1.0000

Figure I-11 Matrix created in the GeneScan Software v3.7

Matrix Editor

Matrix Description

Matrix Name: 310NT_5dyes

Description:

Matrix Settings

Select the Matrix Standard Sample File:

Number of Dyes: 5

B No File Selected for "B" Data Start At: 1000

G No File Selected for "G" Data Start At: 1000

Y No File Selected for "Y" Data Start At: 1000

R No File Selected for "R" Data Start At: 1000

O No File Selected for "O" Data Start At: 1000

Points: 100000

Create

Matrix Result

	B	G	Y	R	O
B	1.0000	0.1680	0.0073	0.0021	0.0035
G	0.6715	1.0000	0.2107	0.0335	0.0027
Y	0.5611	0.8190	1.0000	0.4187	0.0024
R	0.3613	0.6506	0.9016	1.0000	0.0066
O	0.0280	0.0545	0.1007	0.1392	1.0000

OK Cancel

Figure I-12 Matrix created in the GeneMapper ID software using the Advanced algorithm

Size Standard Test

The sixth parameter evaluated in the Peak Detection and Genotyping category is the size standard test using the size match editor in the Classic and Advanced modes. The size match editor calculates size standards differently depending on the algorithm used to generate the data.

- Classic algorithm

If the Classic algorithm is used to analyze data, a size quality value of 0.5 is associated with each sample ([Figure I-13 on page I-22](#)). The size standard peaks are identified by the manual assignment of size standard definitions to the associated data points within a run, similar to GeneScan software v.3.1.2 (Macintosh OS).

- Advanced algorithm

The Advanced algorithm, however, has a size quality value ranging from 0.0 to 1.0 associated with each sample (Figure I-14 on page I-23). Ratio matching is used to identify the size standard fragments, similar to GeneScan software v3.7.1 (Windows NT OS).

You can check and edit the sizing result. The sizing result is applied to the final genotype process component quality value (PQV). An analyst can check the size quality of each sample by highlighting the individual samples in the navigation pane. Further, after evaluating each size standard, an analyst may override the size quality value to 1.0 to confirm that each size standard was examined.

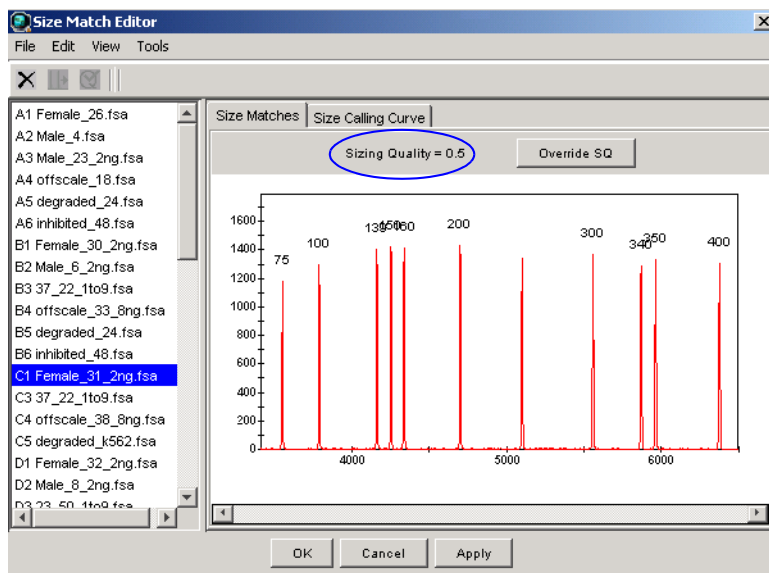


Figure I-13 Size Match Editor using the Classic algorithm displaying a sizing quality value of 0.5

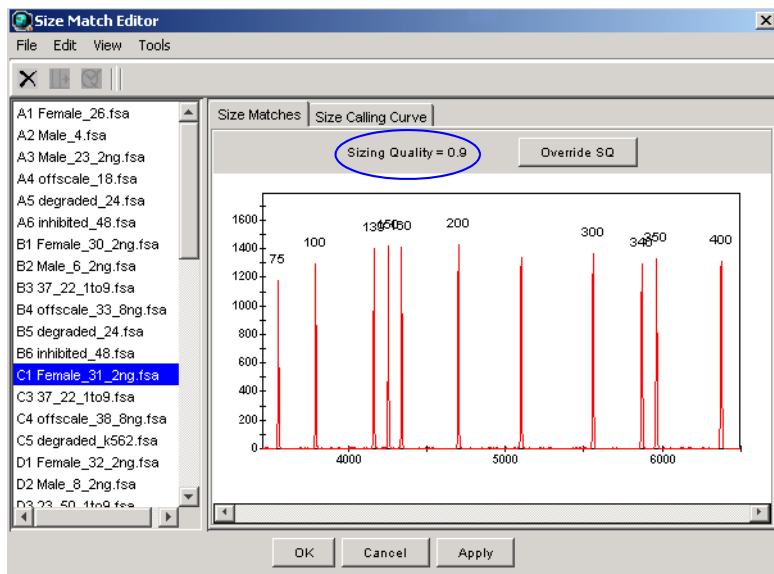


Figure I-14 Size Match Editor using the Advanced algorithm displaying a sizing quality value of 0.9

Algorithm Testing

The second category presented is the Algorithm testing.

Test Parameters The tested parameters are described below.

1. Global cut-off value test using the Classic and Advanced algorithms
2. Minus A cut-off value using the Classic and Advanced algorithm
3. Off-scale flag verification using Classic and Advanced algorithms
4. Off-scale display verification using Classic and Advanced algorithms
5. Out of bin allele flag verification using Classic and Advanced algorithms
6. Spectral pull-up flag verification using Classic and Advanced algorithms

7. Broad peak flag verification using Classic and Advanced algorithms
8. Peak height ratio flag verification using Classic and Advanced algorithms
9. Allele Number flag verification using Classic and Advanced algorithms
10. Low peak height flag verification using Classic and Advanced algorithms
11. Control concordance flag verification using Classic and Advanced algorithms
12. Overall genotype quality verification
13. Overall size quality verification

Global Cut-off Value Testing

The first parameter tested in the Algorithm testing category is the global cut-off value. Global cut-off value verification using Classic and Advanced algorithms were evaluated using the defined ABI PRISM platforms and AmpF/STR kits.

The cut-off value ignores peaks lower than the cut-off ratio (as defined) of the highest peak in the locus size range. Three different values were chosen to test 60 alleles for the global cut-off values of 0%, 10%, and 20%. The results show that the algorithms implemented in GeneMapper ID Software v3.1 properly filter peaks that meet the global cut-off specification.

In [Figure I-15](#), the higher the cut-off value, the less conservative the call (meaning less peaks are detected).

Note: GeneMapper ID software users should carefully consider the sample type when choosing the cut-off value.

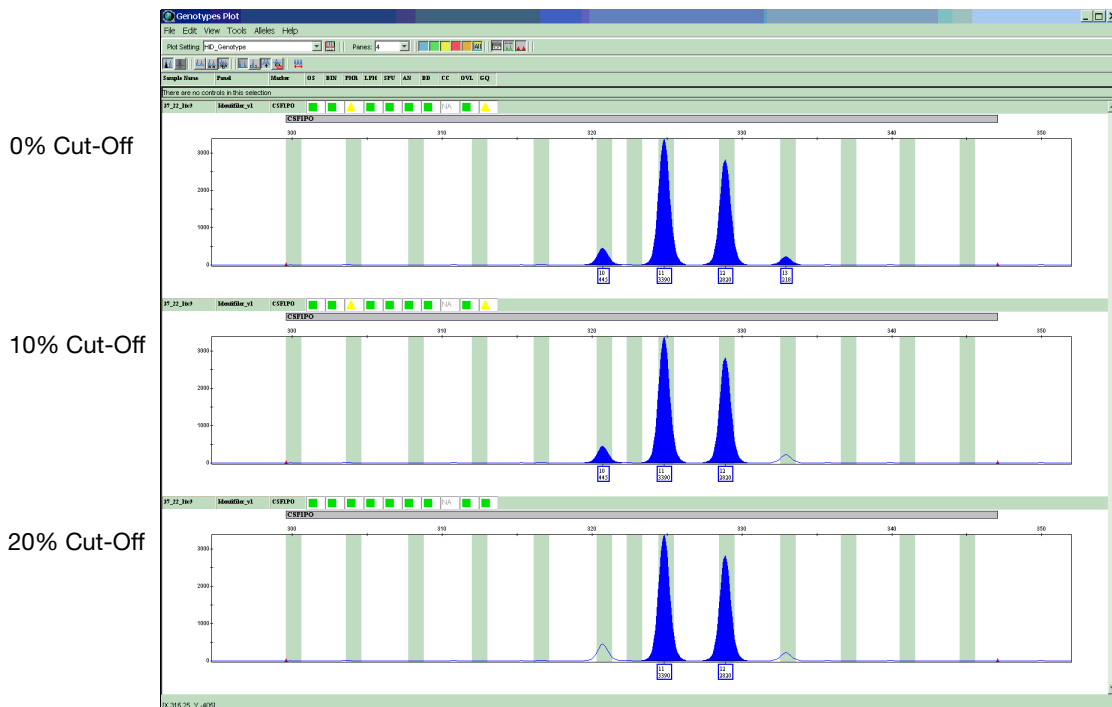


Figure I-15 Example of a 1 to 9 DNA mixture sample tested using three global cut-off values

Minus A Cut-off Value Testing

The second parameter tested in the Algorithm testing category is the Minus A cut-off value.

The Minus A peak is displayed as a shoulder peak to the parent peak (1 base pair less than the main allele). Three different values were chosen to test 2772 alleles in three high quantity DNA samples and 16 alleles in one single source sample for Minus A cut-off at ratios of 0%, 20% and 30%. The results ([Figure I-16 on page I-26](#)) show that the algorithms implemented in GeneMapper ID software properly filtered the percentages entered in the analysis method editor.

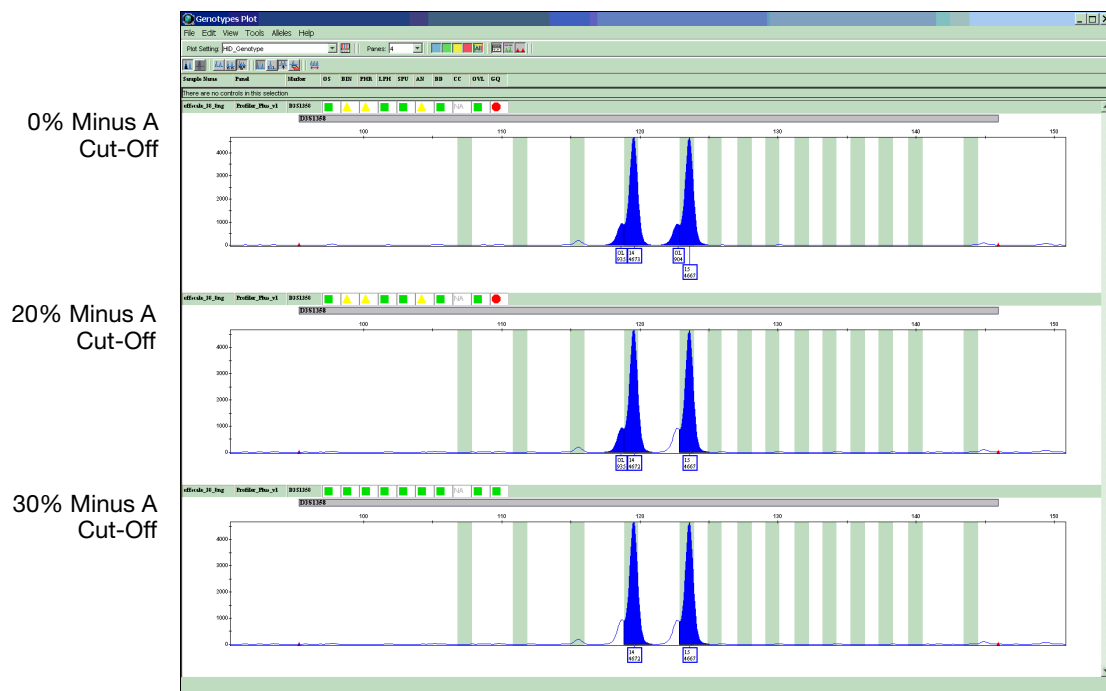


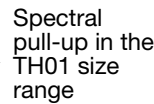
Figure I-16 Example of a high quantity (8ng) DNA sample tested using three Minus A cut-off values. Minus A is filtered with increasing cut-off values.

PQVs Parameters 3 to 7 in the Algorithm Testing category are the individual PQVs for the off-scale flag verification; the off-scale display verification; the out of bin flag verification; the spectral pull-up flag verification; and the broad peak flag verification using both the Classic and Advanced modes.

The weighted values for the individual PQVs were evaluated using the default threshold settings in GeneMapper *ID* Software v.3.1 to verify that the quality score given for each PQV is an accurate assessment of the sample.

A total of 2772 alleles comprising three (3) off-scale samples were evaluated. The PQV flags for the off-scale samples were presented accurately in the Classic mode. However, when these same off-scale samples were analyzed in the Advanced mode, it was observed that the PQV flags for the pull-up peaks that were caused by off-scale peaks (i.e., in the same position in a different color; see Figure I-17) are overridden and reflect the PQV flags for the true off-scale peak in that position.

It is recommended that an analyst carefully review all off-scale yellow PQV flags since it is known that the flags for the pull-up peaks in the same position as the true off-scale peak do not accurately reflect the pull-up peak in the Advanced mode. For example in Figure I-18 the spectral pull-up peak is in the same position in a different color of an off-scale allele; and the off-scale, spectral pull-up, and broad peak values do not accurately reflect the PQV flags that are displayed for that position. That is, the off-scale PQV flag is yellow (this spectral pull-up peak is not off-scale and should be flagged green), the spectral pull-up PQV flag is green (this is true spectral pull-up and should be flagged yellow), and the broad peak PQV flag is yellow (this peak is not broad and should be flagged green). As mentioned, these PQV flag colors for off-scale, spectral, and broad peak should be reversed for the pull-up peak but the PQV for the true off-scale peak is accurate. The out of bin PQV flag for all tested alleles accurately reflect the data.



Peak Height Ratio and Allele Number PQR Flag Verification

The eighth and ninth parameters tested in the Algorithm Testing category were the individual PQR for the peak height ratio (PHR) flag verification and allele number (AN) flag verification using both the Classic and Advanced modes. 3,678 alleles from mixture DNA samples and 16 alleles from a single source DNA sample for the peak height ratio flag and allele number flag were evaluated.

Results

100% of the alleles tested accurately reflect the PHR and AN PQR.

Example Data Showing PHR and AN PQR Flag Verification

	Sample Name	Run Name	Panel	Marker	Dye	Allele 1	Allele 2	ADO	AE	OS	BIN	PHR	LPH	SPU	AN	BD	CC	OVL	GQ
69	Sample4	Databasing	Identifier_v1	D3S1358	G	14	17												
70	Sample4	Databasing	Identifier_v1	TH01	G	9	9.3												
71	Sample4	Databasing	Identifier_v1	D13S317	G	12	13												
72	Sample4	Databasing	Identifier_v1	D16S539	G	12	13												
73	Sample4	Databasing	Identifier_v1	D2S1338	G	25													
74	Sample4	Databasing	Identifier_v1	D19S433	Y	12	14												
75	Sample4	Databasing	Identifier_v1	vWA	Y	16	18												
76	Sample4	Databasing	Identifier_v1	TPOX	Y	8													
77	Sample4	Databasing	Identifier_v1	D18S51	Y	18													
78	Sample4	Databasing	Identifier_v1	AMEL	R	X													
79	Sample4	Databasing	Identifier_v1	D5S818	R	12													
80	Sample4	Databasing	Identifier_v1	FGA	R	21	26												
81	Sample5	Databasing	Identifier_v1	D8S1179	B	12	13												
82	Sample5	Databasing	Identifier_v1	D21S11	B	29	30												
83	Sample5	Databasing	Identifier_v1	D7S820	B	11													
84	Sample5	Databasing	Identifier_v1	CSF1PO	B	10	11												
85	Sample5	Databasing	Identifier_v1	D3S1358	G	15	17												
86	Sample5	Databasing	Identifier_v1	TH01	G	6	9.3												
87	Sample5	Databasing	Identifier_v1	D13S317	G	11													
88	Sample5	Databasing	Identifier_v1	D16S539	G	11													
89	Sample5	Databasing	Identifier_v1	D2S1338	G	23													
90	Sample5	Databasing	Identifier_v1	D19S433	Y	13	14												
91	Sample5	Databasing	Identifier_v1	vWA	Y	17													
92	Sample5	Databasing	Identifier_v1	TPOX	Y	8	9												
93	Sample5	Databasing	Identifier_v1	D18S51	Y	15	18												
94	Sample5	Databasing	Identifier_v1	AMEL	R	X	Y												
95	Sample5	Databasing	Identifier_v1	D5S818	R	11	13												
96	Sample5	Databasing	Identifier_v1	FGA	R	24	26												

Figure I-19 Genotypes table of the CSF1PO locus of a 1 to 1 mixture displaying two yellow PQR flags (Peak Height Ratio and Allele Number) giving an overall genotype quality value of red

As illustrated in [Figures I-19](#), the CSF1PO locus is displaying a yellow flag for both PHR and AN. By default, the setting for PHR is 70% and the maximum number of alleles expected is 2. [Figure I-20](#) displays the electropherogram of the CSF1PO locus selected in [Figure I-19](#). In [Figure I-20](#), the PHR is below the default setting and the maximum number of expected alleles is greater than the default setting, 2. The software has appropriately flagged the CSF1PO locus for the PHR and AN PQV.

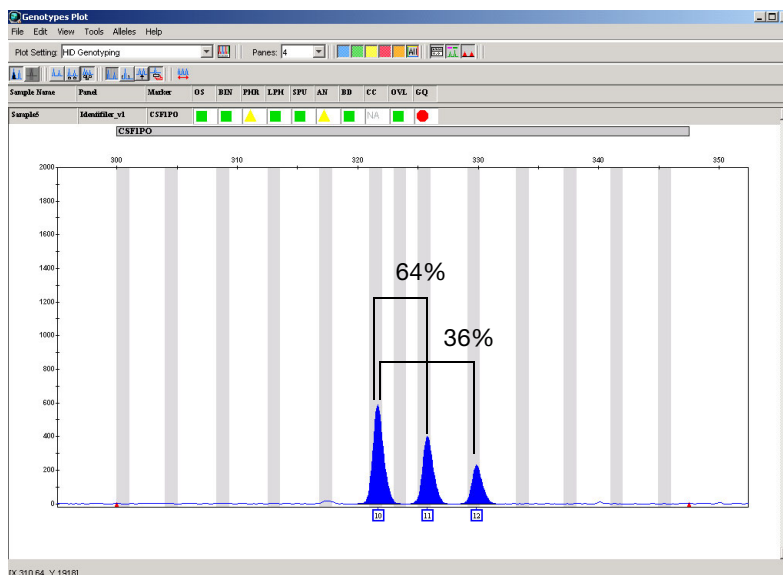


Figure I-20 The default peak height ratio value is set at 70% and the maximum allele number is set at 2 in GeneMapper ID software. Any peak below these values activates the PQV flags. The ratio between the first and second peak is 64% and the ratio between the first and third peak is 36%, resulting in a yellow peak height ratio PQV flag.

Low Peak Height PQV Flag

The tenth parameter tested in the Algorithm Testing category is the individual PQV for the low peak height (LPH) flag verification using both the Classic and Advanced modes. 2,772 alleles from low quantity DNA samples and 16 alleles from a single source DNA sample were evaluated.

Results

100% of the alleles tested accurately reflect the LPH PQV.

**Control
Concordance
PQV Flag**

The eleventh parameter tested in the Algorithm Testing category is the individual PQV for the control concordance (CC) verification using both the Classic and Advanced modes. 198 alleles for Control DNA 9947A, 96 alleles for Control DNA 007, and 30 negative control amplifications were evaluated.

Results

100% of the alleles tested accurately reflect the CC PQV.

Overall GQ PQV

The twelfth parameter tested in the Algorithm testing category is the final PQV for the overall genotype quality (GQ) verification using both the Classic and Advanced modes. 20,364 alleles were evaluated and accurately reflect the overall GQ PQV except when off-scale data is evaluated.

The genotypes table below shows two low quantity DNA samples amplified with the AmpF $\mathbb{L}$ STR Profiler Plus kit. For illustration purposes, two loci (D13S317 and D7S820) with their respective PQVs are calculated on page I-32.

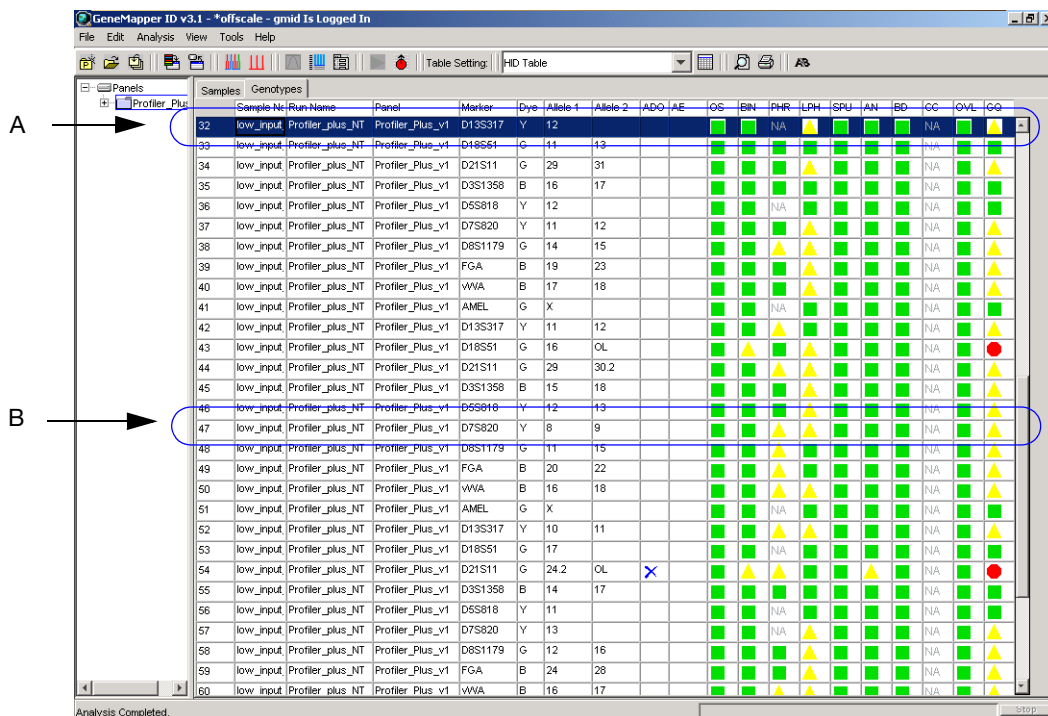


Figure I-21 Genotype table displaying PNVs

PNVs are calculated using an equation that takes the user defined PNVs to generate the genotype quality score. PNVs are weighted from 0-1, 0(weight) equals no effect on the final value and 1 (weight) equals full effect. The individual PNVs were evaluated using the default threshold values GeneMapper™ ID software. If a sample meets the individual PNV threshold defined in the software it is treated as a zero in the calculation. See examples below.

Overall Genotype Quality (GQ) PNV

Green = 0.75 to 1.0
 Yellow = 0.26 to 0.7
 Red = 0.0 to 0.25

The calculation for the Overall GQ PNV by the software is as follows:

$$GQ = 1 - [(1 - PNV_{OS}) * (1 - PNV_{BIN}) * (1 - PNV_{PHR}) * (1 - PNV_{LPH}) * (1 - PNV_{SPU}) * (1 - PNV_{AN}) * (1 - PNV_{BD}) * (1 - PNV_{CC}) * (1 - PNV_{OVVL})]$$

Example A

The LPH PQV has a weighted value of 0.3 in the default settings.

$$GQ = 1 - [(1 - 0_{OS}) * (1 - 0_{BIN}) * (1 - 0_{PHR}) * (1 - 0.3_{LPH}) * (1 - 0_{SPU}) * (1 - 0_{AN}) * (1 - 0_{BD}) * (1 - 0_{CC}) * (1 - 0_{OVL})]$$

$$1 - [1_{OS} * 1_{BIN} * 1_{PHR} * 0.3_{LPH} * 1_{SPU} * 1_{AN} * 1_{BD} * 1_{CC} * 1_{OVL}]$$

$$GQ = 1 - [0.3] = 0.7$$

An overall GQ PQV of 0.7 displays a yellow flag.

Example B

The PHR and LPH PQVs have weighted values of 0.3 in the default setting.

$$GQ = 1 - [(1 - 0_{OS}) * (1 - 0_{BIN}) * (1 - 0.3_{PHR}) * (1 - 0.3_{LPH}) * (1 - 0_{SPU}) * (1 - 0_{AN}) * (1 - 0_{BD}) * (1 - 0_{CC}) * (1 - 0_{OVL})]$$

$$1 - [1_{OS} * 1_{BIN} * 0.7_{PHR} * 0.7_{LPH} * 1_{SPU} * 1_{AN} * 1_{BD} * 1_{CC} * 1_{OVL}]$$

$$GQ = 1 - [0.49] = 0.51$$

An overall GQ PQV of 0.51 displays a yellow flag.

Overall Size Quality PQV Verification

The thirteenth parameter tested in the Algorithm Testing category is the final PQV for the overall size quality (SQ) verification using both the Classic and Advanced modes. 20,364 alleles for the overall size quality PQV were evaluated to test the overall sizing quality. The sizing quality for each sample is used in the calculation of the overall GQ PQV.

Results

The final size quality value for each sample displayed an accurate assessment of the sizing for each sample evaluated.

Example of Size Quality PQV Verification

The samples view below is highlighting the sample for the Control DNA 9947A and is displaying a red size quality value. As shown in [Figure I-23 on page I-35](#), the start scan collection point entered in the analysis method excluded the 75 base pair size fragment peak, resulting in the inaccurate labeling of the size standard peaks.

The screenshot shows the GeneMapper ID v3.1 software interface. The title bar reads "GeneMapper ID v3.1 - Database - gmid Is Logged In". The menu bar includes File, Edit, Analysis, View, Tools, and Help. The toolbar contains various icons for file operations and analysis. The "Table Setting" dropdown is set to "HD Table". On the left, a "Project" tree shows a "Databasing" folder with sub-items: ID_Control, ID_Ladder, ID_Neg_cn, ID_Sample, ID_Sample, and ID_Sample. The main window displays a table with the following data:

Samples	Category	Analysis Method	Panel	Size Standard	Matrix	Run Name	Instrument Type	SQO	SFNF	SNF	SQ	UD1
1	Identifier	Identifier_v1	sd			Databasing	AB3100					
2	HD_Advanced	Identifier_v1	sd			Databasing	AB3100					
3	HD_Advanced	Identifier_v1	sd			Databasing	AB3100					
4	Offe	HD_Advanced	Identifier_v1	sd		Databasing	AB3100					
5	HD_Advanced	Identifier_v1	sd			Databasing	AB3100					
6	HD_Advanced	Identifier_v1	sd			Databasing	AB3100					

The "SQ" column for the first row (Sample 1) contains a red circle, indicating a red size quality value. The status bar at the bottom left says "Analysis Completed." and the bottom right has a "Stop" button.

Figure I-22 The sample table displaying a red size quality value

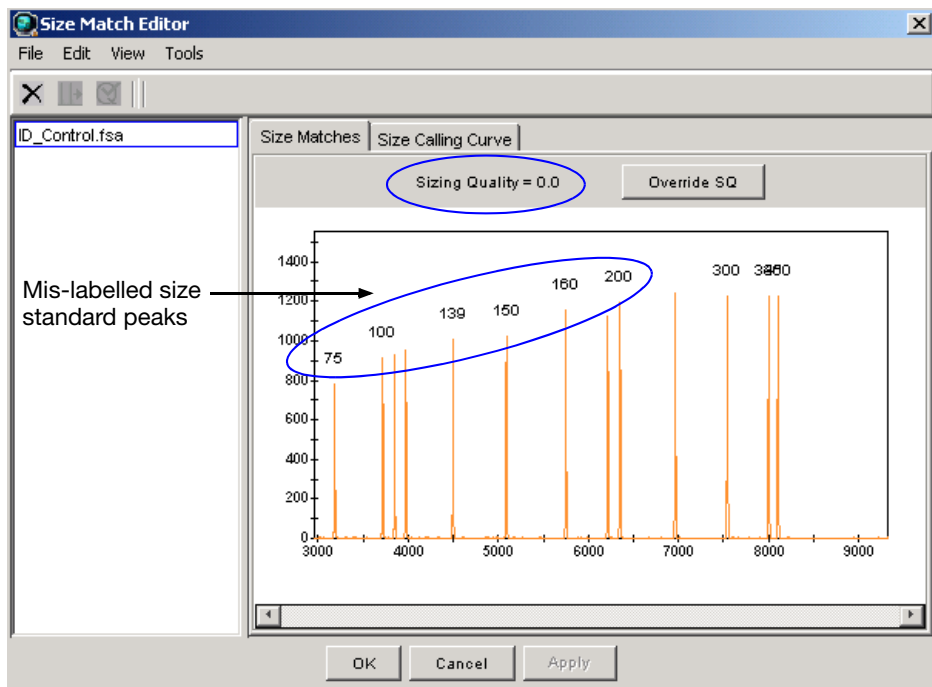


Figure I-23 Size Match Editor in the Advanced mode displaying the size standard for the control DNA 9947a amplified with the AmpF/STR Identifiler kit displaying a size quality value of 0.0

Workflow

The third category presented is Workflow testing.

Test Parameters

1. Sort and display samples per color, per sample and per locus functionality test
2. Display default and customized plot settings functionality test
3. Printing test functionality test

Process Samples Per Color, Per Sample, and Per Marker

The first parameter tested in the Workflow category is the ability to sort and display samples per color, per sample, and per locus. The genotype plot view enables a user to graphically sort and display data per sample, per color, and per locus and to assess the quality of the samples (i.e., the PQV).

Results:

The ability to display plots for the 20,364 alleles evaluated in these various views is 100% accessible and functional.

Display Default and Customized Plot Settings

The second parameter tested in the Workflow category is the ability to display default and customized plot settings. A user can create and edit plot settings. For example, a trained analyst wanting to interpret a mixture sample can create a plot setting to view electropherograms in only one color.

Six projects were evaluated for the functionality of the plot settings to display the data as defined in the plot settings editor. Five different predefined display settings were evaluated: AmpF Φ STR sizing, AmpF Φ STR genotyping, Overlay ROX, Overlay LIZ, and customized plot settings (displays only one color at a time).

Results

The ability to display data as defined in the plot settings editor is 100% functional with the six projects tested.

Printing

The third parameter tested in the Workflow category is the printing test.

In the GeneMapper *ID* software, various plot performance tests were conducted. These tests measure performance attributes such as time to display plots, time to scroll through plots and sample configurations. The result for the amount of time to display sample configurations (e.g., display different panes, samples and dyes) was less than ten seconds. In the GeneMapper *ID* software, the results for viewing the data in different plot configurations (e.g., five samples showing all dye colors in five panes) using three modes of zooming (no zoom, medium zoom and tight zoom) ranged from 0.15 seconds to 0.37 seconds. The printing test was only conducted on one printer; different printers may have different results.

Data Handling

The fourth category presented is data handling.

Test Parameters

The tested parameters are described below:

1. Import and export functionality test of projects, analysis methods, table settings, plot settings, matrices, and size standards
2. Verify CODIS export functionality using both export extension files CMF 1.0 and CMF 3.0.

Import/Export Functionality

The first parameter tested in the data handling category is the import and export functionality of projects, analysis methods, table settings, plot settings, matrices, and size standards.

Projects with their associated settings (i.e., analysis methods, table settings, plot settings, matrices, if applicable, and size standards) should be exported and archived regularly in order to maintain the database. Further, the associated settings must accompany the exported project in order to accurately assess the original analyses.

Note: When the project is exported, the sample files are not linked.

Results

The ability to export and import projects, analysis methods, table settings, plot settings, matrices, and size standards is 100% functional.

CODIS Export Functionality

The second parameter tested in the data handling category is the CODIS export functionality test.

One advantage of GeneMapper *ID* is the ability to add extension files that are recognized by the Combined DNA Index System (CODIS) software. These samples files can be exported and uploaded into the CODIS software.

Ten sample files (both single source and mixture samples using the AmpFISTR Identifiler, AmpFISTR Profiler Plus, and AmpFISTR COfiler PCR Amplification Kits) were successfully imported using both CODIS v. 5.6 and CODIS v. 5.7. The FGA locus in the Identifiler PCR kit has nine (9) extended alleles. (Applied Biosystems 2001). When performing the verification of GeneMapper *ID*, one (1) extended allele at the FGA locus was not recognized. The CODIS Change Control Board has been informed of this finding.

Conclusion

Applied Biosystems performed verification of GeneMapper™ *ID* v3.1 to assess the performance, robustness, and feature designs of four categories: 1) Peak detection and genotyping, 2) Algorithms, 3) Workflow, and 4) Data Handling. Within these categories, over 20,000 alleles and software features were verified for 24 specific test parameters. The data for this verification were evaluated and are summarized in this appendix.

The verification was performed using data generated with AmpF/STR kits using forensic-simulated and single source DNA samples and electrophoresed on ABI PRISM platforms. Our findings demonstrate that GeneMapper *ID* Software v3.1 with its default settings and panels and bin sets is valid for forensic, paternity and databasing analyses. This software accurately detects, genotypes, and performs quality checks on data analyzed with GeneMapper *ID* software. However, during the verification testing, the process component-based quality value (PQV) flags for spectral pull-up peaks associated with off-scale peaks can be inaccurate.

Further, our findings demonstrate that the genotypes between GeneScan and Genotyper software to GeneMapper *ID* software are concordant. Laboratories currently using GeneScan 3.1.2 for the Macintosh OS can maintain current analysis parameter settings when adopting GeneMapper *ID* software in the Classic mode. Applied Biosystems' verification testing demonstrates that human identification laboratories can successfully adopt GeneMapper *ID* Software v3.1.

References

Applied Biosystems. 2001. *ABI PRISM® AmpFLSTR® Identifiler™ PCR Amplification Kit User's Manual*.

Applied Biosystems. 2002. *ABI PRISM® GeneScan® Analysis Software for the Windows NT® Operating Systems: Overview of the Analysis Parameters and Size Caller*.

Boland, C.E., Leibelt, C.S., Daoudi, Y., and Roby, R.K. 2003. "Concordance Study of STR Results Using Multiple ABI PRISM® Genetic Analysis Instruments, AmpFLSTR® Kits, and ABI PRISM® Analysis Software." *Proceedings of the American Academy of Forensic Sciences*, Denver, Colorado, Publication Printers, Corp., pp. 24-25.

Leibelt, C.S., Boland, C.E., Brown, C.L., Hatch, T.L., Daoudi, Y., Lou, Y., and Roby, R.K. 2003. "Verification of GeneMapper™ ID Software for STR Analysis." *Proceedings of the American Academy of Forensic Sciences*, Denver, Colorado, Publication Printers, Corp., pp. 50-51.

Software Warranty

J

This appendix describes the software warranty provided by Applied Biosystems for the GeneMapper™ ID Software Version 3.1.

This appendix covers:

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Limited Product Warranty	J-2

Computer Configuration

Configuration Applied Biosystems supplies or recommends certain configurations of computer hardware, software, and peripherals for use with its instrumentation. Applied Biosystems reserves the right to decline support for or impose extra charges for supporting nonstandard computer configurations or components that have not been supplied or recommended by Applied Biosystems. Applied Biosystems also reserves the right to require that computer hardware and software be restored to the standard configuration prior to providing service or technical support. For systems that have built-in computers or processing units, installing unauthorized hardware or software may void the Warranty or Service Plan.

Limited Product Warranty

Limited Warranty Applied Biosystems warrants that for a period of ninety (90) days from the date the warranty period begins, its GeneMapper™ ID Software Version 3.1 will perform substantially in accordance with the functions and features described in its accompanying documentation when properly installed on the instrument system for which it is designated, and that for a period of ninety (90) days from the date the warranty period begins, the tapes, diskettes, or other media bearing the software product will be free of defects in materials and workmanship under normal use. If buyer believes that it has discovered a failure of the software to satisfy the foregoing warranty, and if buyer notifies Applied Biosystems of such failure in writing during the ninety (90) day warranty period, and if Applied Biosystems is able to reliably reproduce such failure, then Applied Biosystems, at its sole option, will either (i) provide any software corrections or “bug-fixes” of the identified failure, if and when they become commercially available, to buyer free of charge, or (ii) notify buyer that Applied Biosystems will accept a return of the software from the buyer and, upon such return and removal of the software from buyer’s systems, terminate the license to use the software and refund the buyer’s purchase price for the software. If there is a defect in the media covered by the above warranty and the media is returned to Applied Biosystems within the ninety (90) day warranty period,

Applied Biosystems will replace the defective media. Applied Biosystems does not warrant that the software will meet buyer's requirements or conform exactly to its documentation, or that operation of the software will be uninterrupted or error free.

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Effective Date**

Any applicable warranty period under these sections begins on the earlier of the date of installation or ninety (90) days from the date of shipment for software installed by Applied Biosystems personnel. For all software installed by the buyer or anyone other than Applied Biosystems, the applicable warranty period begins the date the software is delivered to the buyer.

Warranty Claims

Warranty claims must be made within the applicable warranty period.

**Warranty
Exceptions**

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Workflow for Storing Sample Files, Projects, and Second Reads



The following workflow applies only when the computers running GeneMapper *ID*™ software are connected to your network.

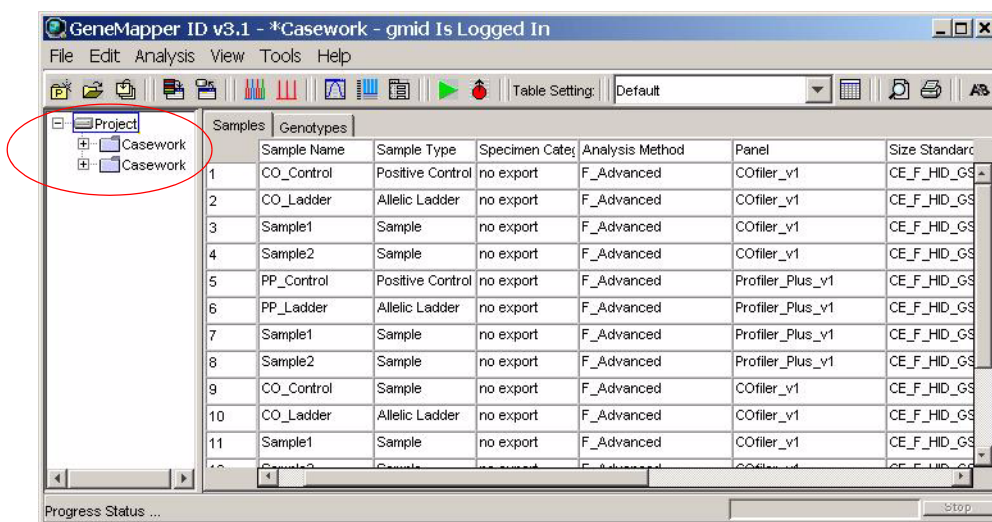
1. Verify that all copies of the GeneMapper *ID* software in the lab have exactly the same analysis method, size standard definition, panels, binset, and matrix (if applicable) imported into the GeneMapper Manager.
 - a. From the GeneMapper Manager, export the master copy of each of the files to a designated location on a server.
 - b. For each computer using GeneMapper *ID* software, use the GeneMapper Manager to import the files from the server.
 - c. Keep the master copy of the files on the server as a backup.
2. Store all sample files on a server that can be accessed by all computers with GeneMapper *ID* software. Do not move these sample files or download them to personal computers.

Note: The GeneMapper project requires an unaltered link to the sample files. If these sample files are moved, the link is broken and some functionality within the project, such as accessing the raw data, viewing certain plots, and re-analyzing samples is lost.

3. For the initial analysis of the data (by the first or lead analyst), import the sample files into a new project, perform analysis, edit the alleles, and save the project to a designated location on the server.
4. For the second analysis of the data:

Using Same Computer ...	Then Proceed to Step ...
Yes	5 and skip step 6
No	6

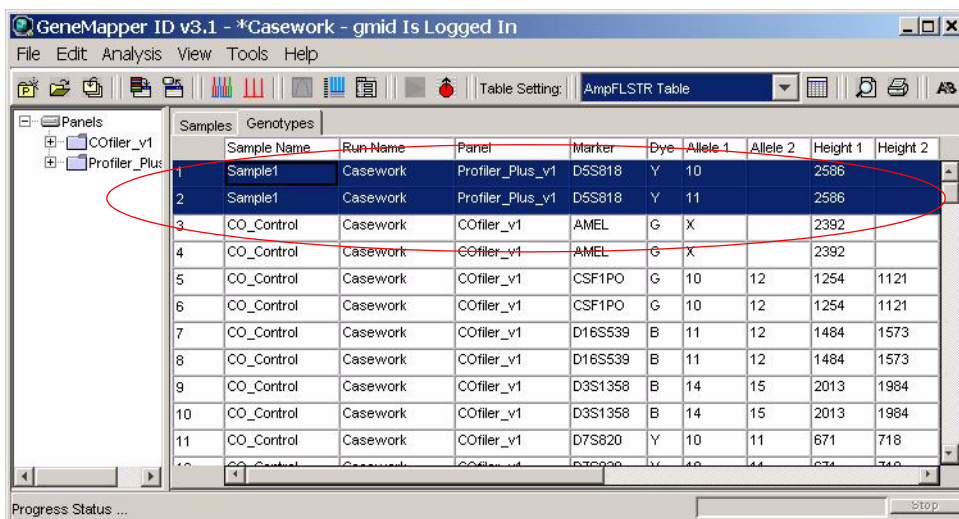
5. Select **File > Open Project**. Select the project to open, then click **Open**.
6. If you are using a different computer, the second analyst must:
 - a. Import the project created by the first (lead) analyst in the Project tab of GeneMapper Manager.
 - b. Select **File > Open Project**. Select the project to open, then click **Open**.
 - c. Import the same samples. Two folders display in the left navigation pane differentiating each set of analyses:



With the Project node highlighted in the left navigation pane, all sample files will be displayed. To display only the sample files associated with one analyst, highlight the first or second folder. The first folder is associated with the lead analyst and the second folder is associated with the second read. You can duplicate run folders on the server to give these folders different names to provide more obvious differentiation.

Note: To import the same sample files, the project must be saved, closed, then reopened. If you do try to import the same sample files without closing it first, an error message displays and the files are not imported.

7. Select the folder of samples just imported, then analyze, and edit the data, and save the project. Both sets of analysis are saved within the same project.
8. To compare results, Select the Project node in the left navigation pane of the main window. Select **Genotypes tab > Analysis Menu > Non-concordant sample to the top**. Any non-concordant calls then display at the top.



GeneMapper ID v3.1 - *Casework - gmid Is Logged In

File Edit Analysis View Tools Help

Table Setting: AmpFLSTR Table

Panels: COfiler_v1, Profiler_Plus

Sample	Genotypes	Sample Name	Run Name	Panel	Marker	Dye	Allele 1	Allele 2	Height 1	Height 2
1	Sample1	Casework	Profiler_Plus_v1	D5S818	Y	10			2586	
2	Sample1	Casework	Profiler_Plus_v1	D5S818	Y	11			2586	
3	CO_Control	Casework	COfiler_v1	AMEL	G	X			2392	
4	CO_Control	Casework	COfiler_v1	AMEL	G	X			2392	
5	CO_Control	Casework	COfiler_v1	CSF1PO	G	10	12		1254	1121
6	CO_Control	Casework	COfiler_v1	CSF1PO	G	10	12		1254	1121
7	CO_Control	Casework	COfiler_v1	D16S539	B	11	12		1484	1573
8	CO_Control	Casework	COfiler_v1	D16S539	B	11	12		1484	1573
9	CO_Control	Casework	COfiler_v1	D3S1358	B	14	15		2013	1984
10	CO_Control	Casework	COfiler_v1	D3S1358	B	14	15		2013	1984
11	CO_Control	Casework	COfiler_v1	D7S820	Y	10	11		671	718
12	CO_Control	Casework	COfiler_v1	D7S820	Y	10	11		671	718

Progress Status ... Stop

Glossary

Allele	A variant of a marker
Allele calling	Identification of alleles based on bin definitions; genotyping; GeneMapper <i>ID</i> software analysis
Bin	A fragment size or basepair range and dye color that define an allele
Bin set	A set of bin definitions for one source or set of experimental conditions, usually an instrument; bin sets are available inside a kit
Diploid, polyploid	Having 2 or more alleles, respectively, per gene or locus
Genotype	The set of allele calls for a marker or genetic locus; usually 2 alleles
GM ID	GeneMapper <i>ID</i> software
HID	Human identification and forensic applications
Kit	A set of panels; the grouping of panels in a kit is determined by the kit provider
Marker	A known microsatellite or SNP genotyping location
Microsatellite	Short tandem repeat marker (di-, tri-, tetra-nucleotide repeat)
Panel	A set of bin definitions for one or more markers; the grouping of markers in panels is determined by the kit provider
PQV	Process Quality Value - Quality values assigned to bins based on the success rate of the bins to locate allele peaks correctly
SNaPshot Kit Multiplex Analysis	Primer extension-based chemistry for SNP genotyping validation
SNP	Single-nucleotide polymorphism (used in this document to refer to SNaPshot® Kit System markers)

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