GETTING STARTED GUIDE

applied biosystems
life technologies

GeneMapper®*ID-X* Software

Version 1.5 – Basic Features

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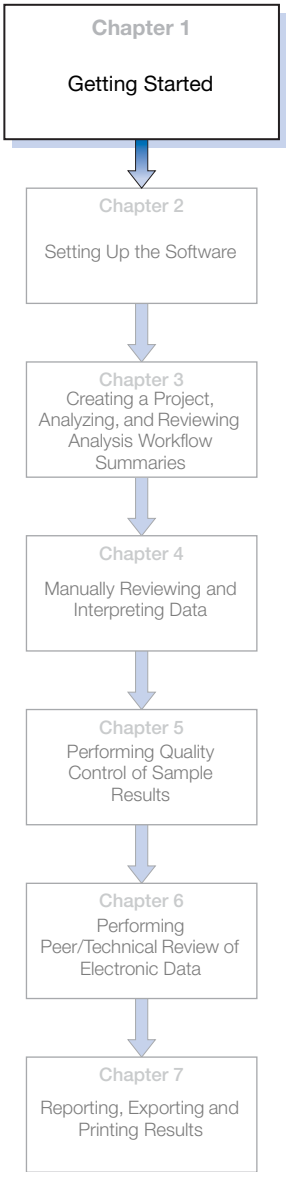
About this guide

Revision history

Revision	Date	Description
B	May 2015	Remove draft watermark.
A	February 2015	New document for v1.5 software. Replaces v1.0 Getting Started Guide Pub. no. 4375574. Added ACC information.

1

Getting Started



This chapter covers:

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Overview of the GeneMapper® *ID-X* Software

GeneMapper® *ID-X* Software is an automated genotyping software solution for all human identification (HID) data analysis needs, including forensic casework, databasing, and paternity testing. The workflow for performing AmpFℓSTR® kit data analysis with GeneMapper *ID-X* Software is shown below.

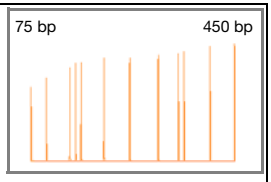
Process samples with AmpFℓSTR® kits

Run samples on instrument:

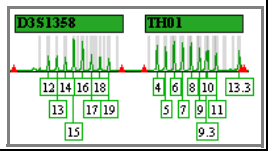
- 310 Genetic Analyzer
- 3100 Genetic Analyzer
- 3130/3130*xl* Genetic Analyzer
- 3500/3500xL Genetic Analyzer

Analyze samples in GeneMapper® *ID-X* Software:

- Size based on:
  - Size standard run with chemistry
  - Size standard specified in software



- Genotype based on:
  - Allelic ladders run with chemistry
  - Panels and bins specified in software



Note: GeneMapper® *ID-X* Software has undergone a verification process. However, human identification laboratories that choose to use GeneMapper *ID-X* Software for data analysis should perform their own appropriate validation studies.

Features of the GeneMapper® *ID-X* Software

The software provides a quality value system and a set of streamlined data review tools and features for both expert system and traditional manual review workflows. Included in the system are the following features:

Analysis Requirement Check

The analysis requirement check identifies unmet requirements before analysis starts. For example, the software checks if there is a sample with Sample Type = Allelic Ladder listed in the Samples table. The system can be set up to stop analysis and display an alert if this sample type is not found.

Allelic Ladder Quality Assessment

The allelic ladder quality assessment:

- Evaluates allelic ladders (based on system-defined allelic ladder quality requirements) before proceeding to sample analysis
- Flags run folders without at least one passing allelic ladder. You can review allelic ladders before proceeding with analysis.
- Automatically excludes low-quality ladders from analysis and continues analysis with passing ladders. You can optionally override the software assessment and use low-quality ladders to generate bin offsets.

Analysis Summary

For efficient data evaluation, the analysis summary provides:

- An easy-to-view summary of analysis results
- An overview of allelic ladder, control, and sample quality
- A separation of passing samples from samples that do not meet one or more quality thresholds
- Interactive links to specific categories of samples (passing/check/low quality, allelic ladder/control/sample)

Comprehensive Quality Value System

Note: For more information on the Quality Value system, see “The GeneMapper® ID-X Software Quality Value System” on page 7.

Manual Review Tools

For efficient data review in traditional manual review and expert systems workflows, the manual review tools provide:

- Process quality value (PQV) flags (, ,)
- Automatic spike labeling based on intelligent rules
- User-defined "artifact" peak labels
- Marker-specific quality value details with thresholds and observed values displayed to show deviations from thresholds and identify sources of anomalies
- Mark Sample for Deletion and Delete All Labels from Sample functions in the Samples plot to allow easy elimination of low-quality samples directly from the plot window
- Override Genotype Quality (GQ) and Composite Genotype Quality (CGQ) functions to "manually accept" genotypes at the marker and sample level
- Detailed label edit table display and visual indicators to indicate edits (gray PQVs) for electronic peer/technical review

Quality Control

The quality control features:

- Evaluate sample concordance and allele matching
- Support additional custom positive controls and allow automatic concordance checks of custom controls
- Compare samples in a project to one another to determine if they contain profiles similar to neighboring samples
- Compare samples in a project to laboratory reference and custom control profiles using a user-defined match percent threshold
- Export comparison results for further analysis and reporting

Chain-of-Custody Systems for Electronic Data

The chain-of-custody systems for electronic data can be custom-configured (or turned off) by the GeneMapper *ID-X* System administrator as needed. These systems provide:

- Security that controls user access to software functions and data, and allows custom configuration that meets the data-sharing needs of your laboratory and limits access to data when needed
- Auditing that tracks changes and provides audit history reports

- E-signature that requires user-authentication before changes are saved

For more information on these features, see the *GeneMapper® ID-X Software Administrator's Guide*.

Multi-User Database Environment

The multi-user database environment:

- Allows multiple users to share projects in a centralized database
- Facilitates efficient data sharing between analysts for second analysis and review
- Limits the need to import and export projects
- Allows central management of analysis settings

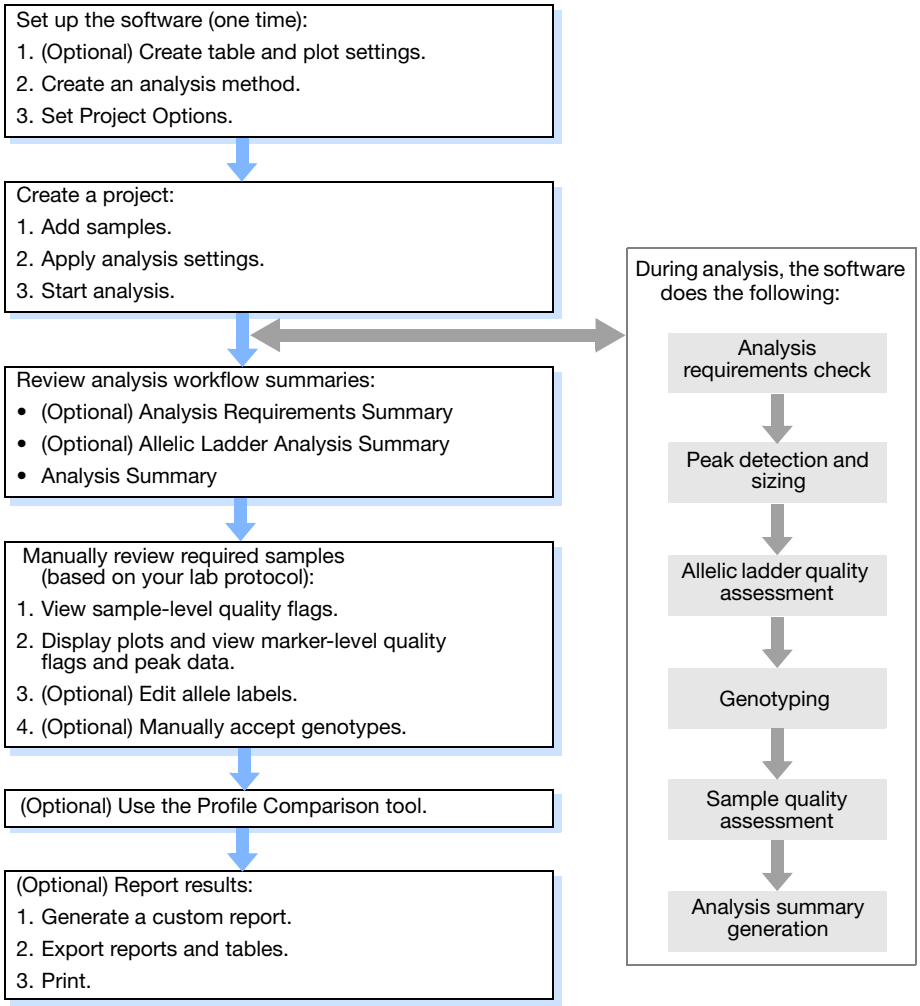
Report Manager

For customized table-formatted reports, the Report Manager:

- Allows you to change column names and column order
- Can export a traditional horizontal allele table for a selected group of samples for use as a genotype summary or import into LIMS or other downstream applications

GeneMapper® *ID-X* Software and Analysis Workflows

The following flowchart summarizes the steps for performing a typical data analysis workflow using the GeneMapper® *ID-X* Software. To the left are the steps the user performs when analyzing samples and interpreting results. To the right are the software operations that occur automatically during analysis.



The GeneMapper® *ID-X* Software Quality Value System

Overview

The GeneMapper® *ID-X* Software quality value system:

- Assesses the quality of allelic ladders before analysis and does not consider low-quality allelic ladders for genotyping.
- Assesses the quality of data at the sample and marker level using PQVs.
- Can be used in an optimized and validated expert system or traditional manual review workflow to quickly identify data quality issues and aid in interpretation of samples that do not meet all thresholds.
- Can be used in an optimized and validated expert system workflow to quickly segregate samples that require manual review from those that do not.

The PQV results of the quality assessment are displayed as color-coded flags: Pass ( ), Check, ( ), Low Quality ( ). The color of the flag depends on software-specified thresholds or user-defined thresholds set in the analysis method.

Optimizing and Validating an Expert System

Before using any software as an expert system, optimize and validate the thresholds for each AmpFℓSTR® kit and instrument platform combination by processing a variety of samples that challenge each of the different quality flags.

IMPORTANT! Different kit/instrument combinations may require different thresholds.

Quality Value System Checks and Assessments

The GeneMapper *ID-X* Software quality value system performs the following checks and assessments:

- **Analysis requirements checks** – Before analysis starts, identifies any conditions that may prevent analysis or cause unexpected results.
- **Sizing quality assessment** – Evaluates the quality of the size standard profile in each sample.
- **Allelic ladder quality assessment** – Evaluates allelic ladder quality. Also determines if an allelic ladder is used for creating bin offsets.
- **Marker-level quality assessment** – Evaluates labeled peaks within each marker. Contributes to the overall genotype quality assessment.
- **Sample-level quality assessment** – Evaluates the quality of the entire sample.
- **Genotype quality assessment** – Evaluates the quality of each marker in a sample. Contributes to the overall composite genotype quality assessment.

The following sections contain a brief description of each quality value system check and assessment along with a list of each sample-level and marker-level quality value.

Note: For more information on the quality value system, see the *GeneMapper® ID-X Software Reference Guide*.

Analysis Requirements Checks

The analysis requirements checks are performed and results displayed either in the Samples table before analysis starts or in the Analysis Requirements Summary after analysis starts.

Acronym	Full Name	Description and Flags
ARNM	Analysis Requirement Not Met	Indicates if all analysis requirements are met. These requirement checks are performed when analysis is started: • Sample File Not Found • Analysis Method Not Selected • Analysis Method Not Found in the Database • Panel Not Selected • Panel Not Found in the Database • Binset Not Selected • Binset in Analysis Method Does Not Match Binset Selected in the Panel Manager • Size Standard Not Selected • Size Standard Not Found in Database • Size Standard Dye Color is Not Present in the Sample Dye Set • Matrix Not Selected • Matrix Not Found or Contains Invalid Data • No Allelic Ladder Selected in Run Folder • GMID v3.x Analysis Method Selected • Basic or Classic Size Standard Selected • SNP Panel Selected

Sizing Quality Assessment

The quality value system evaluates the quality of the size standard profile within each sample (SQ) and allows you to flag size standards with poor peak resolution. Sizing quality assessment is displayed in the Samples table after analysis completes.

Acronym	Full Name	Description and Flags
SQ	Sizing Quality	Evaluates the similarity between the fragment pattern for the size standard dye specified in the size standard definition and the actual distribution of size standard peaks in the sample, calculates an interim SQ (a value between 0 and 1), then applies the broad peak weighting specified in the analysis method, as described in the *GeneMapper® ID-X Software Reference Guide*. **Note:** The GeneMapper *ID-X* Software does not genotype samples with SQ.

Allelic Ladder Quality Assessment

The quality value system performs an allelic ladder quality assessment to determine if a ladder is used in genotyping (to create bin offsets). Allelic ladder samples are analyzed before all other samples. An allelic ladder sample must have a SQ and a CGQ to be used for creating bin offsets. For an allelic ladder to have a CGQ, all the markers within the allelic ladder must pass the following rules:

Rule	Description
1	All ladder alleles specified in the panel used to analyze are detected.
2	In each marker, the peak height ratio of the first and second peak is greater than 50%. This rule eliminates allelic ladders if the stutter peak before the first true allele peak is labeled as an allele.
3	No spikes are detected above 20% (default) of the highest allele peak in the same dye color within the extended marker range. **Note:** Spike detection for allelic ladders is performed within each extended marker range (no gaps are present between markers; the end point of each marker is extended past the marker definition in the panel to the beginning of the next marker). **Note:** The Allelic Ladder Spike Cut-off value is user-definable in the Peak Quality tab of the analysis method.
4	The peak height ratio between the lowest and highest peak is equal to or greater than 15%.

Marker-Level Quality Assessments

Marker-level quality assessments indicate the quality of each marker in a sample and are displayed in the Genotypes table after analysis completes.

Acronym	Full Name	Description and Flags
OS	Off-scale	Indicates if any fluorescence signal within the marker exceeds the detection threshold of the instrument.
BIN	Out of Bin Allele	Indicates if labeled peaks do not fall inside bins. These peaks are labeled with OL (Off ladder).
PHR	Peak Height Ratio	Indicates if the peak height ratio between the lowest and highest peak is less than the Min Peak Height Ratio defined in the analysis method.
MPH	Max Peak Height	Indicates if any peak heights (in RFU) within the marker size range exceed the Max Peak Height value (in RFU) set in the analysis method.
LPH	Low Peak Height	Indicates if any peak heights (in RFU) within the marker size range are below the following thresholds set in the analysis method: • Homozygous Min Peak Height • Heterozygous Min Peak Height
AN	Allele Number	Indicates if the software detects no alleles, more than the Max Expected Alleles set in the analysis method (Peak Quality tab), or no X allele detected in amelogenin.
BD	Broad Peak	Indicates if the width of any peak exceeds the Max Peak Width (half height in base pairs) defined in the analysis method (Peak Quality tab).
CC	Control Concordance	Indicates if a positive, custom, or negative control produces the expected profile.
SPK	Marker Spike	• **Allelic ladders** – Indicates if spikes are detected within each extended marker range (no gaps are present between markers; the end point of each marker is extended past the marker definition in the panel to the beginning of the next marker). • **Samples** – Indicates if spikes are detected within a marker size range. The software uses a proprietary algorithm that detects spikes based on the peak morphology.
OVL	Overlapping Alleles	Indicates if a labeled peak (allele or artifact) falls within the size ranges of two neighboring markers.

Acronym	Full Name	Description and Flags
ACC	Amelogenin Cross Check	For analysis of samples that contain both Y and autosomal STRs. Indicates if Amelogenin and Y marker results are concordant. The same flag is displayed across Amelogenin and all markers with the Y Marker checkbox selected in the Panel Manager.

Genotype Quality Assessment

For samples, the quality value system assigns the GQ for each marker based on the individual marker quality flags. For allelic ladders, the quality value system assigns a GQ for each marker based on the allelic ladder quality requirements, as described in "Allelic Ladder Quality Assessment" on page 10. The GQ is used to determine the CGQ, and is displayed in the Genotypes table after analysis completes.

Acronym	Full Name	Description and Flags
GQ (samples)	Genotype Quality	Indicates the genotype quality of the marker in the sample. The genotype quality for a sample marker is determined based on the presence of labeled peaks detected (after filtering) and the GQ weighting specified in the analysis method. If no labeled peaks are detected (and the sample is not a negative control), the GQ is set to 0. If one or more labeled peaks are detected, the GQ is initially set to 1 with a final value determined by the GQ weighting of individual marker-level quality values as specified in the analysis method.
GQ (allelic ladders)	Genotype Quality	Indicates the genotype quality of the marker in the allelic ladder. The genotype quality for an allelic ladder marker is determined using system-defined quality rules (as described in the *GeneMapper® ID-X* Software *Reference Guide*) to ensure: • All expected peaks are present. • Peak height ratio of the first and second peak is greater than 50%. • No spikes are present in the extended marker range (within or between markers). • The peak height ratio between the lowest and highest peak is equal to or greater than 15%. **IMPORTANT!** If the Allelic Ladder GQ Weighting for Spikes is set to 0 (off) in the analysis method, the GQ may be , even if spikes are present in the allelic ladder.

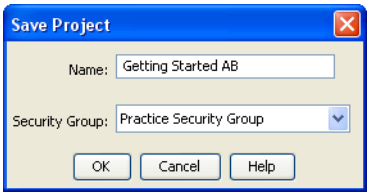
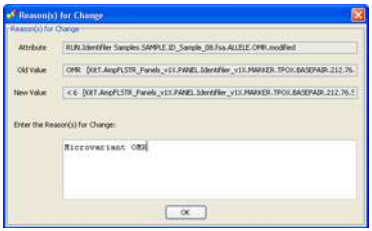
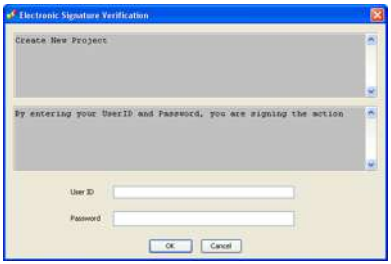
Sample-Level Quality Assessments

Sample-level quality assessments that indicate the quality of the entire sample are displayed in the Samples table after analysis completes.

Acronym	Full Name	Description and Flags
SOS	Sample Off-scale	Indicates if any fluorescence signal within the analysis range exceeds the detection threshold of the instrument.
MIX	Mixed Source	Indicates a potential mixed-source sample.
OMR	Outside Marker Range	Indicates if labeled peaks are detected between two marker size ranges defined in the panel.
SSPK	Sample Spike	• **Allelic ladders** – Indicates if spikes are detected within the sizing range. • **Samples** – Indicates if spikes are detected within or between two defined marker size ranges. Does not indicate if spikes are detected before the first marker or after the last marker. The software uses a proprietary algorithm that detects spikes based on the peak morphology.
CGQ (samples)	Composite Genotype Quality	Indicates overall sample genotype quality. Considers the individual marker GQ values.
CGQ (allelic ladders)	Composite Genotype Quality	Indicates overall allelic ladder quality. Considers the allelic ladder quality assessment (see page 10). **Note:** Allelic ladder samples with CGQ are not used to create bin offsets. **IMPORTANT!** If the Allelic Ladder GQ Weighting for Spikes is set to 0 (off) in the analysis method, the CGQ may be even if spikes are present in the allelic ladder.

GeneMapper® *ID-X* Software Security

The GeneMapper® *ID-X* Software contains a security system that includes the following features relevant to the procedures outlined in this guide:

Security System Component	Description
User accounts	A required component of the security system that allows only authorized users to access the software. This guide assumes that you log in using the Practice User account provided with the software (see the *GeneMapper® ID-X Software Administrator's Guide* for a description of user accounts). Chapter 2 provides detailed instructions for logging in.
Security groups	A component of the security system that restricts user access to specific data items (project, panels, analysis methods, and size standards.) When using the Practice User account, data is assigned to the Practice Security Group automatically (you are not prompted to select a security group). 
Audit trail	A component of the security system that keeps track of changes made and can require users to provide a reason for a change. The audit trail is set up by default to require a reason for change when you edit allele labels. If additional auditing is set up on your system, you may be prompted to specify a reason when you create or edit data items. 
E-signature	An optional component of the security system that requires users to provide a valid user name and password when they make a change. This guide assumes that E-signature *is not* set up on your system. If E-signature is set up on your system, you are prompted to provide your user name and password when you create or edit data items. 

For information on:

- Setting up security, audit trail, and E-signature on your system, see the *GeneMapper® ID-X Software Administrator's Guide*.
- Using security groups (and the impact that security groups have on the data you can access), using the audit trail, and using E-signature, see the *GeneMapper® ID-X Software Help.*

GeneMapper® *ID-X* Software Terms

Some common terms used in this guide are:

Term	Definition
allele	Variant form of a marker (locus).
allelic ladder	A sample that contains a set of alleles that are representative of those found in a particular STR marker. Allelic ladders are generated with the same primers as tested samples, providing a reference DNA size for each allele included in the ladder. Unknown samples are compared against them to determine their genotype.
AmpFℓSTR® Chemistry kit	Applied Biosystems® Human Identification PCR amplification kits for short tandem repeat (STR) analysis.
analysis method	A collection of user-defined settings that determine the sizing, genotyping and quality value algorithms used by the GeneMapper® *ID-X* Software to analyze sample files in a project.
bin	A fragment size (± 0.5 bp) that defines an allele within a marker.
bin set	A collection of bins (allele definitions), typically specific to a set of panels.
CODIS	The FBI Laboratory Combined DNA Index System. For more information, see: **http://www.fbi.gov/hq/lab/codis/index1.htm**
custom control	A positive amplification control other than the control DNA supplied with the AmpFℓSTR® kits.

Term	Definition
expert system	A computer program that: • Interprets alleles and DNA profiles • Uses knowledge acquired from Qualified Analysts • Is Rule-based • Acts as an “assistant” to Qualified Analysts • Automated–minimal human intervention • Is as good or better than human experts • Documents reasoning behind decisions • Does not require manual review of “passing” samples Source: National DNA Index System (NDIS) DNA Data Acceptable Standards, Appendix B, "Guidelines for Submitting Requests for Approval of an Expert System for Review of Offender Samples," 15 July 2004.
genotype	Allele designations for a genetic locus.
genotyping	Labeling of alleles based on allelic ladder bin comparisons and filtering of alleles based on analysis method settings.
lab reference profile	A genotyped profile of an analyst or other lab personnel.
marker	A genetic locus. A name, fragment size range in base pairs, dye color, repeat length, and physical allelic ladder alleles are defined for each marker.
negative control	A sample expected to generate no allele calls. Can be an extraction blank, reagent blank or an amplification negative control.
panel	A group of markers and properties (size ranges, dye label color, expected positive control genotypes). Each panel provided with the GeneMapper® *ID-X* Software represents a specific AmpFℓSTR® Chemistry kit.
positive control	A sample of known genotype. Each AmpFℓSTR® Chemistry kit includes a positive control. You can also run custom positive controls.
PQV (Process Quality Value)	Process quality values (PQVs) assess the quality of data at the sample and marker levels.
profile	The genotype (allele designations) of a sample.
project	In the GeneMapper® *ID-X* Software, a collection of sizing and genotyping results for a set of data.
run	The electrophoretic injection of a set of samples from a single plate (48- or 96-well) and the resulting sample files.
run folder	A folder containing a set of sample files from a capillary-electrophoresis run.

Term	Definition
sample files	Capillary-electrophoresis data files (*.fsa) generated by Data Collection Software.
size standard definition	A list of fragment sizes in base pairs that a size standard sample contains. Only those sizes required for accurate sizing are contained in the size standard definition.
stutter ratio	Percent value used to filter stutter peaks (remove labels).
traditional manual review	Visual inspection of samples regardless of quality.

For additional definitions, see the *GeneMapper® ID-X Software Help* (select **Glossary** in the Contents tab).

How to Use This Guide

Before You Start

IMPORTANT! Before using the procedures in this guide, make sure the GeneMapper® *ID-X* Software has been successfully installed and registered. See the *GeneMapper® ID-X Software Installation Guide* for more information.

When performing the procedures described in this guide, keep in mind the following:

- The steps in each chapter are designed to flow from start to finish, and from one chapter to the next
- Complete each chapter as a single unit before stopping your work, if possible
- Make sure you perform each step as it is described
- Carefully review any previously performed steps if you observe any differences between what is shown in this guide and what is displayed on your own system

User Account Requirements

You must use the Practice User account provided to perform the procedures in this guide.

Using the Guide with the Example Data Provided

Example data (.fsa) generated using the AmpFℓSTR® Identifiler® PCR amplification kit, and a reference project (.ser) containing analyzed lab reference samples, custom controls, and QC samples are installed with the GeneMapper® *ID-X* Software.

To perform the exercises described in this guide, use the example files (.fsa and .ser) located on your computer as shown below.

Install Configuration	File Location	
	Reference project (.ser)	**Sample files (.fsa)**
Client install	<*drive*>:\AppliedBiosystems\GeneMapperID-X\Client\Example Data\Projects	<*drive*>:\AppliedBiosystems\GeneMapperID-X\Client\Example Data\Identifiler Samples
Full install	<*drive*>:\AppliedBiosystems\GeneMapperID-X\Example Data\Projects	<*drive*>:\AppliedBiosystems\GeneMapperID-X\Example Data\Identifiler Samples

Note: The drive will vary depending on the installation of the GeneMapper® *ID-X* Software. The default installation drive is the Local Disk drive. See the *GeneMapper® ID-X Software Installation Guide* for more information on installation options.

Using the Guide as a Tutorial

This guide is a tutorial designed to help you follow a typical analysis workflow. Using the example data provided with the GeneMapper *ID-X* Software, follow the procedures in Chapters 2-7:

Chapter	Description
Chapter 2, Setting Up the Software	1. View default panels and bins 2. Create an analysis method 3. View default table and plot settings 4. Create new table and plot settings 5. View default size standard definitions 6. Add custom control and lab reference profiles to database 7. Set project options

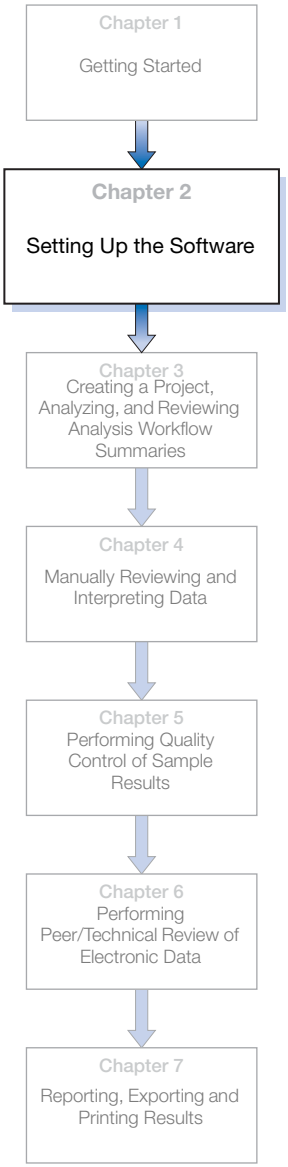
Chapter	Description
Chapter 3, Creating a Project, Analyzing, and Reviewing Analysis Workflow Summaries	1. Create a project and add samples 2. Analyze the data 3. Review the Analysis Requirement Summary 4. Review the Analysis Summary
Chapter 4, Manually Reviewing and Interpreting Data	1. View electropherograms 2. Interpret anomalies using quality value flags and details 3. Edit allele labels 4. Manually accept marker genotypes
Chapter 5, Performing Quality Control of Sample Results	1. Perform Sample Concordance check 2. Perform Sample Comparison check 3. Perform Lab Reference Comparison check 4. Perform Control/QC Sample Comparison check
Chapter 6, Performing Peer/Technical Review of Electronic Data	1. Use filtered tables to find edited samples 2. Use Label Edit Viewer to view edited labels 3. Manually accept sample profiles 4. View edit comments in tables
Chapter 7, Reporting, Exporting and Printing Results	1. Generate a custom report 2. Export reports and tables

For More Information

This guide contains basic procedures. It does not describe all features and parameters in the GeneMapper® *ID-X* Software. For detailed information on topics presented in this guide, see the *GeneMapper® ID-X Software Help*.

2

Setting Up the Software



This chapter covers:

- Overview. 22
- Step 1: Start the Software and Log In. 22
- Step 2: View Panels, Bins, and Stutter Settings 26
- Step 3: Create an Analysis Method. 29
- Step 4: Review Default Table Settings, Plot Settings, and Size Standards. 35
- Step 5: Import Lab Reference and Custom Control Profiles . 48
- Step 6: Set the Project Options 53

Overview

This chapter demonstrates how to prepare the GeneMapper® *ID-X* Software for analysis, using the example data set provided with the software.

In This Chapter

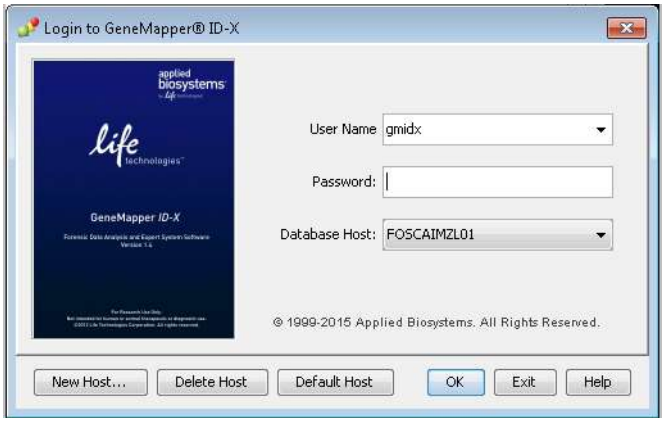
In this chapter you will learn how to:

- Start the software and log in
- Review the panels, bins, and stutter files provided
- Create analysis methods
- Review the table settings, plot settings and size standards provided
- Create new table settings and plot settings
- Import lab reference and custom control profiles
- Set project options

Step 1: Start the Software and Log In

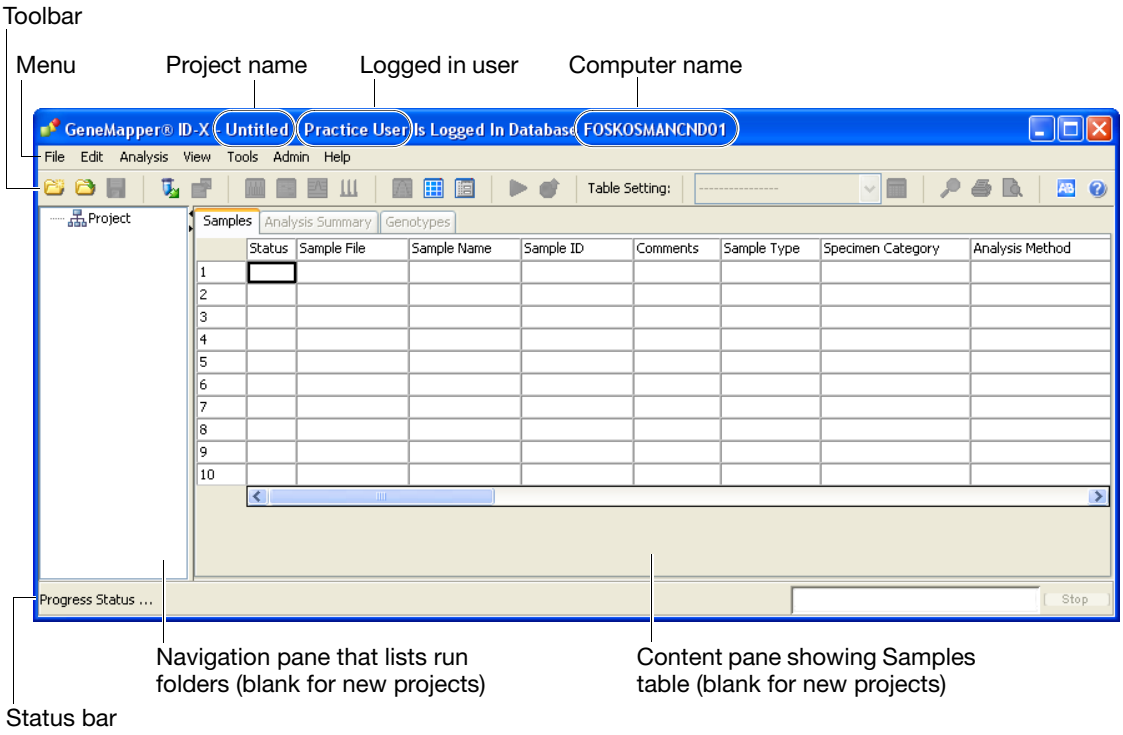
Starting the Software and Logging In

1. Double-click (GeneMapper® ID-X) on the desktop to launch the software.
2. In the Login to GeneMapper® ID-X dialog box, enter **Practice User** for User Name and **password** for Password, then click **OK**.



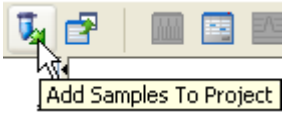
Note: If you have logged in before, you can select the user name from the drop-down list.

The Project window opens.



Resizing and Exploring the Project Window

1. Adjust the window to display as many of the table columns as possible:
   a. Click (Maximize) in the upper right corner of the Project window to expand the window to occupy the full area of the screen.
   b. Resize columns by dragging the separating lines:
      - Position the pointer over the line separating two columns until the pointer changes to sizing arrows.
      - Click-drag the sizing arrows. Dragging to the left narrows the left-hand column. Dragging to the right widens the left-hand column.
2. Place the pointer over toolbar buttons to display tooltips that explain the function of the button.

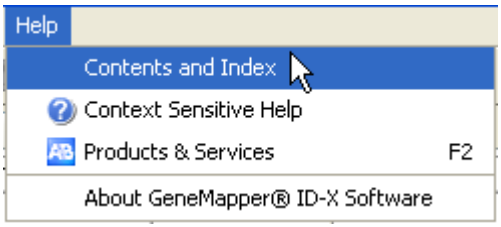


Exploring the Context-Sensitive Help System

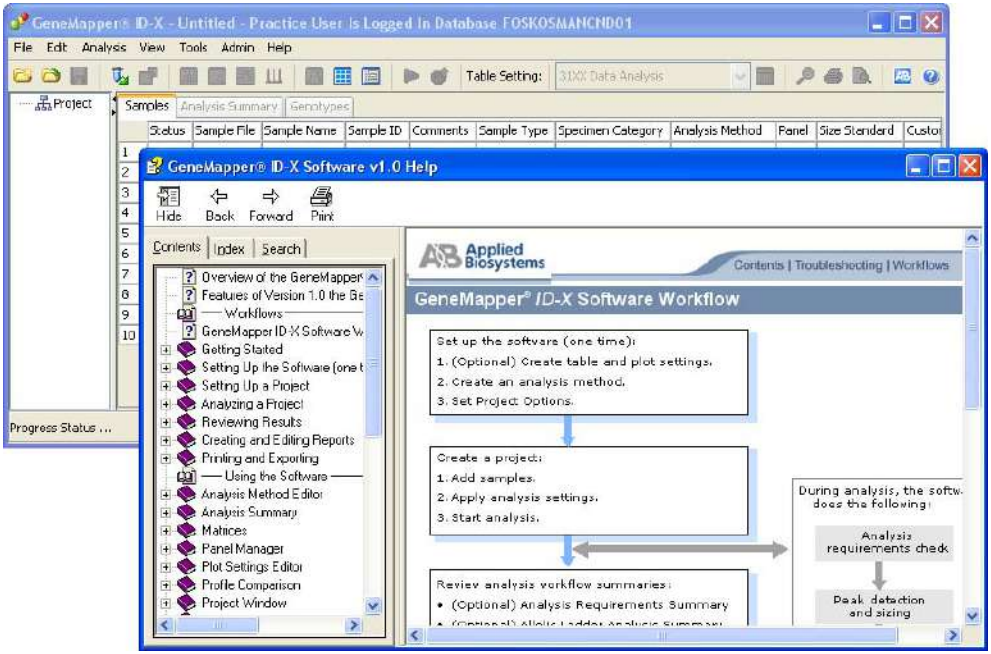
The GeneMapper® *ID-X* Software context-sensitive Help system provides immediate access to detailed information regarding software views, functions and troubleshooting guidelines.

To access the Help system from the Project window:

1. Click the **Help** menu, then select **Contents and Index**.



Help opens in a separate window.



Note: You can also access the Help system anywhere in the software by pressing **F1**, by clicking ? in the toolbar of the Project window, or by clicking the **Help** button in a window, tab or dialog box to access help topics specific to that particular feature of the user interface. You can then click on the internal links within the specific help topic to navigate to related topics.

2. Click ☒ (Close) to close the Help window and return to the Project window.

Step 2: View Panels, Bins, and Stutter Settings

Overview

Before analyzing data, the software must have access to AmpFℓSTR® kit details such as marker size ranges and dyes, allele sizes, and stutter ratios. The files that contain this information are called Panel, Bin, and Stutter files, respectively.

IMPORTANT! The panel, bin and stutter values shown in this section are configured specifically for use with AmpFℓSTR® kit data. We recommend that you use the provided panels and bins when analyzing AmpFℓSTR® data from your laboratory, unless your laboratory has validated alternative values.

When to Import

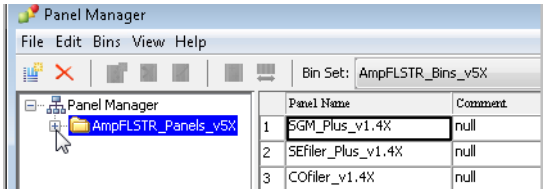
As part of the GeneMapper® *ID-X* Software installation process, the panel, bin and stutter files are automatically imported into the GeneMapper® *ID-X* Software database.

Note: You can import new panel, bin and stutter files whenever updated versions are provided.

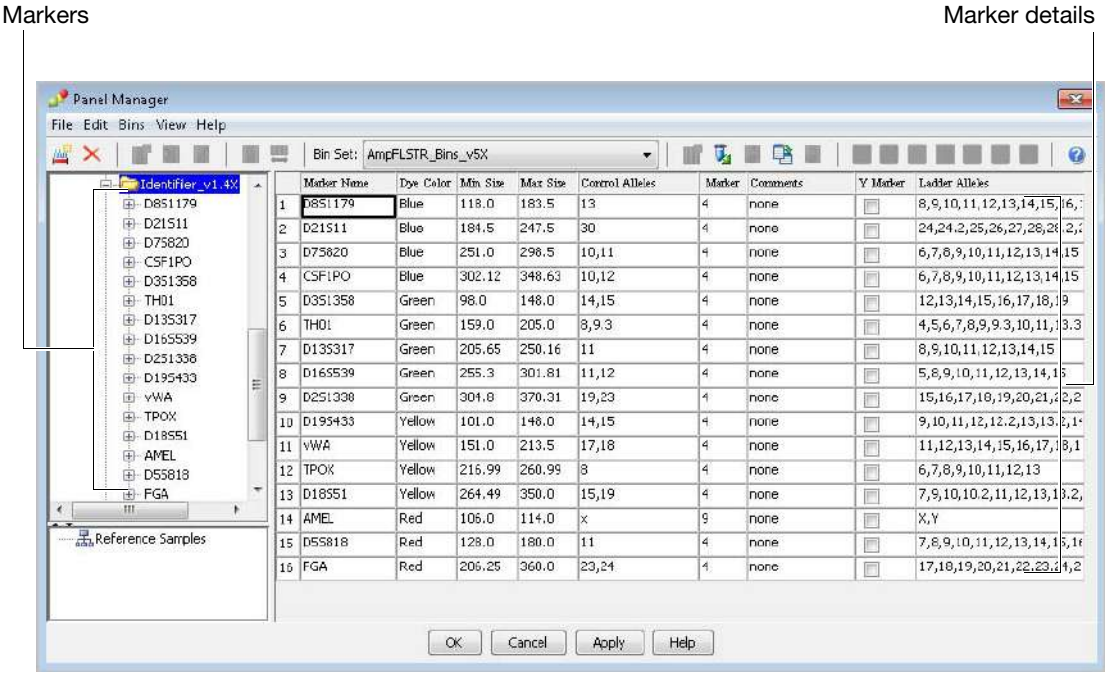
Viewing Panel, Bin, and Stutter Settings

1. If not already started, launch the GeneMapper® *ID-X* Software (see page 22).
2. In the Project window toolbar, click (Panel Manager).

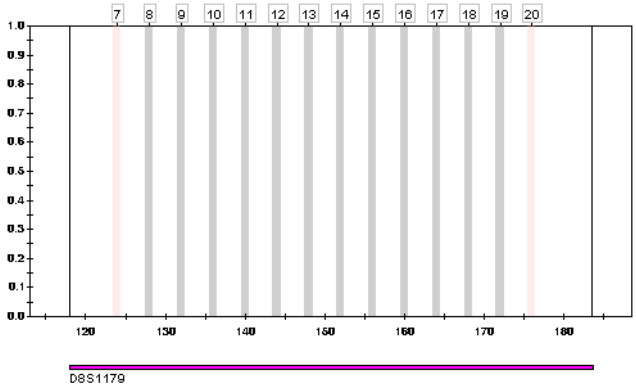
3. In the navigation pane of the Panel Manager, select the **AmpFLSTR_Panels_v5X** kit folder, then click ⊞ to expand its contents.



4. Double-click the **Identifiler_v5X** panel folder. The markers found in the Identifiler kit are listed in the navigation pane and the marker details are displayed in the content pane.



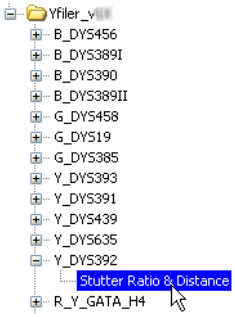
5. Double-click the **D8S1179** marker in the navigation pane. A plot showing the bins for this marker is displayed in the content pane.



Markers can include two bin types: physical (represent alleles present in the allelic ladder sample) and virtual (represent alleles not present in the allelic ladder sample). The software displays all physical bins in grey, and all virtual bins (except for CODIS bins) in pink. In this example, the D8S1179 marker contains two virtual bins, 7 and 20, displayed in pink in the content pane.

6. In the navigation pane, click ⊞ to expand the Yfiler_v5X panel folder.

7. Click ⊞ to expand the Y_DYS392 marker, then select **Stutter Ratio & Distance**.



The marker-specific stutter ratios defined in the stutter file are displayed in the content pane. In this example, both Minus and Plus stutter ratios are specified for this marker.

Please enter the stutter filter(s) for Y_DYS392 marker here.If left blank, the global stutter filter will be applied.

Minus Stutter

	Ratio	From Distance	To Distance
1	0.1622	2.25	3.75
2			
3			
4			

Plus Stutter

	Ratio	From Distance	To Distance
1	0.0790	2.25	3.75
2			
3			
4			

New Edit Delete

Note: From this window, you can apply up to four minus and four plus stutter ratios per marker, and edit the default stutter percentages provided with the GeneMapper® *ID-X* Software.

8. Click **OK** to close the Panel Manager window.

Step 3: Create an Analysis Method

Overview

Analysis methods define the peak detection, sizing, genotyping, and quality assessment parameters applied during analysis of sample data.

In This Section

In this section, you will create a new analysis method, configured specifically for use with the example data provided with the GeneMapper® *ID-X* Software, and with the procedures outlined in this guide.

IMPORTANT! The values used in this guide may not be suitable for analyzing data generated in your laboratory. You must optimize and validate these values during internal verification.

Creating the Analysis Method

1. In the Project window toolbar, click (GeneMapper ID-X Manager).
2. Select the **Analysis Methods** tab, then click **New**.
3. Complete the tabs of the Analysis Method Editor for this example analysis method as described in Table 1 on page 30, and use the *GeneMapper® ID-X Software Help* to learn more about the purpose of specific settings and the effects of changing these settings as needed.

Table 1 Analysis method settings for the example data (Identifiler® samples)

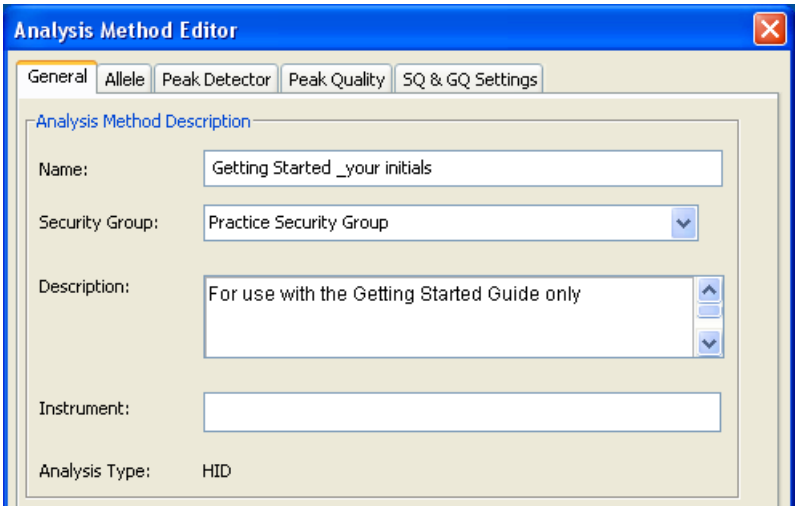
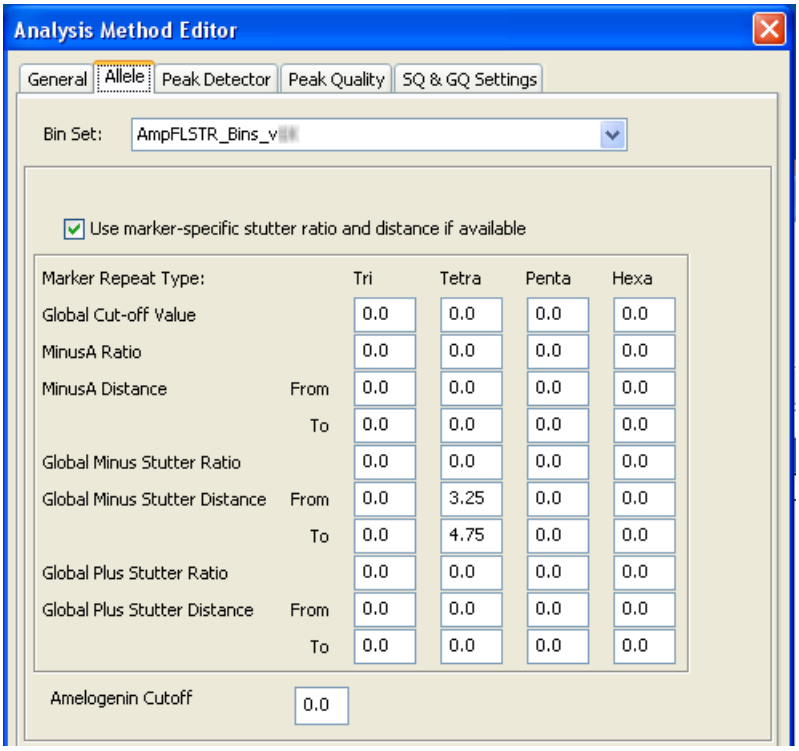
Tab	Settings
General	Enter the settings as shown below:  **Note:** When following the procedures in this guide, We recommend that you add your initials to the names of data objects you create so you can distinguish your data objects from similar objects that may be saved to the same GeneMapper® *ID-X* Software database by other laboratory personnel when performing the same steps in this guide. **Note:** This analysis method is assigned to the Practice Security Group automatically (you are not prompted to select a security group), and is only available if you log in to the software with the Practice User account. You will verify the Project Options set for the Practice User account in “Step 6: Set the Project Options” on page 53.

Table 1 Analysis method settings for the example data (Identifiler® samples) *(continued)*

Tab	Settings
Allele	Select the bin set and review the default settings as shown below:  **Note:** This analysis method specifies marker-specific stutter ratios. In this case, the stutter ratios viewed in the Panel Manager will be applied. However, a Global Cut-off Value and a Global Minus Stutter Ratio may be applied for single-source samples to minimize background labeling that could cause samples to be flagged as low quality.

Analysis Method Editor

General | Allele | Peak Detector | Peak Quality | SQ & GQ Settings

Bin Set: AmpFLSTR_Bins_v

☑ Use marker-specific stutter ratio and distance if available

Marker Repeat Type:		Tri	Tetra	Penta	Hexa
Global Cut-off Value		0.0	0.0	0.0	0.0
MinusA Ratio		0.0	0.0	0.0	0.0
MinusA Distance	From	0.0	0.0	0.0	0.0
	To	0.0	0.0	0.0	0.0
Global Minus Stutter Ratio		0.0	0.0	0.0	0.0
Global Minus Stutter Distance	From	0.0	3.25	0.0	0.0
	To	0.0	4.75	0.0	0.0
Global Plus Stutter Ratio		0.0	0.0	0.0	0.0
Global Plus Stutter Distance	From	0.0	0.0	0.0	0.0
	To	0.0	0.0	0.0	0.0

Amelogenin Cutoff 0.0

Table 1 Analysis method settings for the example data (Identifiler® samples) *(continued)*

Tab	Settings
Peak Detector	Enter the settings as shown below:

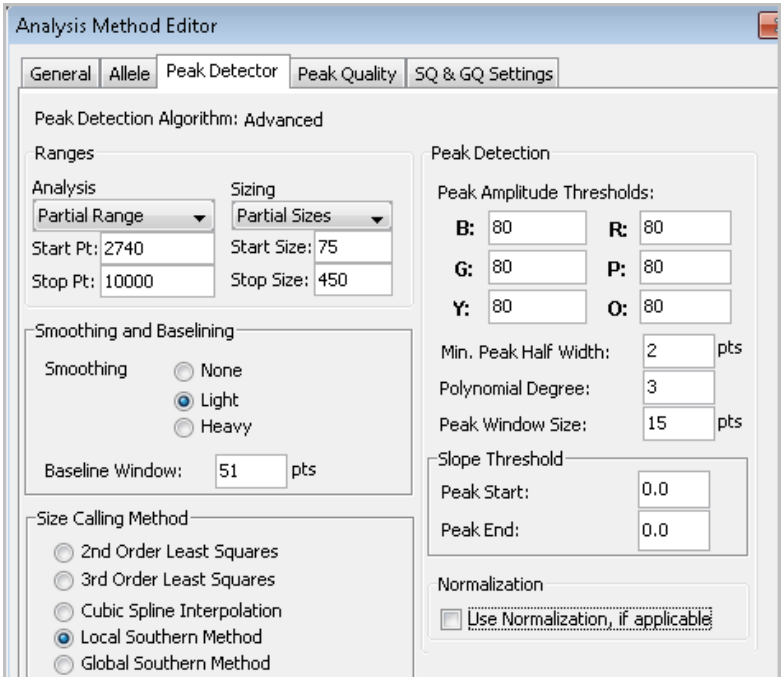


Table 1 Analysis method settings for the example data (Identifiler® samples) *(continued)*

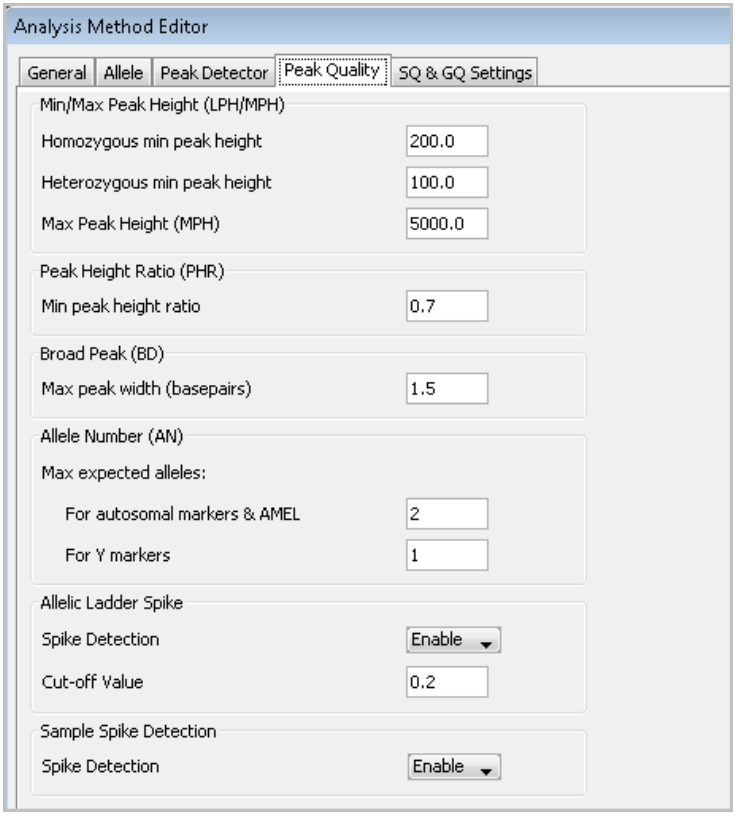
Tab	Settings
Peak Quality	If a marker does not meet a PQV threshold value set in this tab, the GeneMapper® *ID-X* Software will set the marker PQV flag to yellow (Check). Review the default PQV threshold settings shown below. Click **Help** at the bottom of the tab and navigate to the *Peak Quality* help topic to learn more about the parameters presented in this tab. 

Table 1 Analysis method settings for the example data (Identifiler® samples) *(continued)*

Tab	Settings
SQ & GQ Settings	The values entered in this tab affect the calculation of SQ and GQ. Review the default SQ and GQ settings shown below. Click **Help** at the bottom of the tab and navigate to the *SQ & GQ Settings Tab* help topic to learn more about the parameters presented in this tab.

4. After completing all tabs, click **Save** to save your changes and close the Analysis Method Editor dialog. The GeneMapper ID-X Manager remains open.

Step 4: Review Default Table Settings, Plot Settings, and Size Standards

Overview

In this section, you will review the default table, plot, and size standard settings provided with the GeneMapper® *ID-X* Software. The default settings are designed to support a logical and efficient data analysis and review workflow using the example data provided with the software and for use when analyzing data generated in your laboratory. Use of the default settings is demonstrated throughout this guide.

Note: You can modify the default table, plot, and size standard settings or create new settings to support individual laboratory workflows.

Review Default Table Settings

Overview

Table settings determine the content (columns) displayed in or exported from the Samples and Genotypes tables. The default table settings shown in this section include specific columns required to perform different workflow tasks.

Reviewing Table Settings

1. Open the GeneMapper ID-X Manager if not already open (see page 30).
2. In the GeneMapper ID-X Manager, select the **Table Settings** tab.
3. Click a table row to select the table setting to review, then click **Open**.
4. Review the default table settings provided using Table 2 on page 36.
5. After you have finished reviewing the default table settings, click **OK** to close the Table Settings Editor. The GeneMapper ID-X Manager remains open.

Defaults Provided

The following table settings are installed with the GeneMapper® *ID-X* Software.

Table 2 Default table settings

Table Setting		Column Settings	
Name	**Purpose**	**Samples Table**	**Genotypes Table**
310 Data Analysis	Used to set up table data to analyze sample files generated on an 310 Genetic Analyzer.	Displays analysis setting columns and sample-level quality values.	Displays marker and allele columns, and marker-level quality values.
31XX Data Analysis	Used to set up table data to analyze sample files generated on 3100 Series and 3130 Series Genetic Analyzers.	Same as 310 Data Analysis table setting (see entry above), except the Matrix column is not displayed.	Same as 310 Data Analysis table setting (see entry above).
3500 Data Analysis and 3500 Data Analysis 6_dye	Used to set up table data to analyze sample files generated on 3500/3500xL Series Genetic Analyzers.	Same as 310 Data Analysis table setting (see entry above), except the Matrix column is not displayed.	Same as 310 Data Analysis table setting (see entry above).
CODIS Export	Used to enter data in the appropriate columns of the Samples table for exporting a CODIS-supported CMF file.	Displays the columns that may be used to populate CODIS-compatible fields when the table is exported in CMF file format.	**Note:** No action is required on the Genotypes tab when exporting a CMF file.
Import Reference Profiles	Used to import lab reference and custom control profiles into the GeneMapper® *ID-X* Software database.	Displays the Profile ID column and the UD7 column (User-defined 7) in which you specify Custom Control or Lab Reference.	**Note:** No action is required on the Genotypes tab when importing reference profiles.
Known sample export	Used to export a known contributor profile for later import into the Mixture Analysis Tool.	Displays the Sample File and Sample Name columns which are required for Known sample import into the Mixture Analysis Tool.	Displays marker and allele columns.
View CGQ Overrides	Used to quickly identify samples that have been manually accepted (this includes samples with and without allele edits).	Displays only samples with CGQ override flag and sample-level quality values.	Displays marker and allele columns, edit comments and marker-level quality values.

Table 2 Default table settings *(continued)*

Table Setting		Column Settings	
Name	**Purpose**	**Samples Table**	**Genotypes Table**
View Edited Samples	Used to quickly identify samples that have one or more allele edits.	Displays only samples with allele edits and sample-level quality values.	Displays only markers with edits, edit comments and marker-level quality values.
Traditional Allele Table	Used to export data into an allele table using Combined Table Export.	Displays only the sample information required for an allele table export format.	Displays only the marker and allele information required for an allele table export format.
	Note: The allele table export format resembles that created in Genotyper® Software. The table is compatible with spreadsheet software such as Microsoft® Excel®.		
View Unedited Samples	Used to quickly identify samples that have not been manually manipulated (edited or overridden).	Displays only samples without label edits, or GQ or CGQ override flags and sample-level quality values.	Displays only markers without allele edits and marker-level quality values.
Yfiler Haplotype DB Export	Used to export the appropriate columns using Combined Table Export for upload into the Yfiler® Haplotype Database.	Displays only the sample information required for export in Yfiler® Haplotype Database format.	Displays only the sample and marker information required for export in Yfiler® Haplotype Database format.
VALID_GMIDX_ TableSetting-1.0	Used to export the appropriate columns for importing tabular data into VALID™ Software.	Displays only the sample information and run information required for a VALID™ software-compatible format.	Displays only the marker information required for a VALID™ software-compatible format.

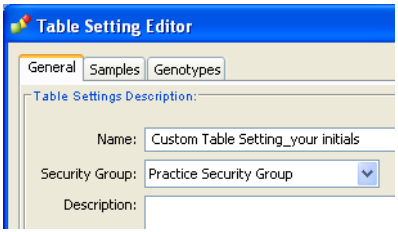
	1	2	3	4
1	Sample Name	D8S1179	D21S11	D7S820
2	Sample 01	14,16	29,31	9
3	Sample 02	14,16	29,31	9
4	Sample 03	15,16	31.2	11,12
5	Sample 04	13,14	30	8,10

Note: The columns in the Genotypes table displayed at the bottom of the Samples plot are determined by the table setting selected in the Project window.

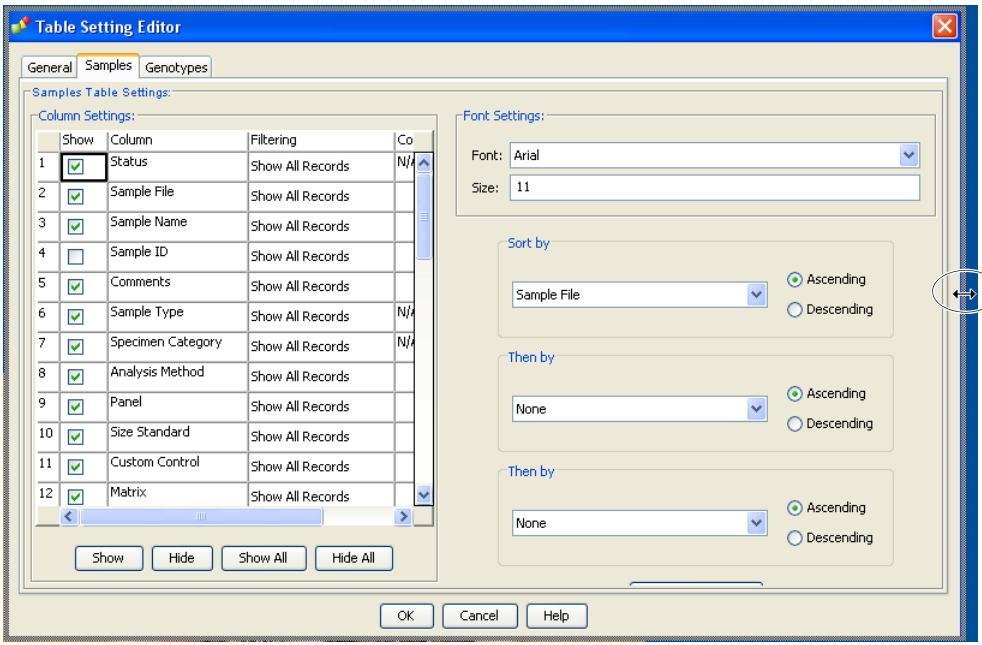
Creating a New Table Setting

With custom table settings, you can adjust your view of the Samples and Genotypes tables to show or hide specific columns, and apply filters to display only specific samples.

1. Open the GeneMapper ID-X Manager if not already open (see page 30).
2. In the GeneMapper ID-X Manager, select the **Table Settings** tab, then click **New** to open the Table Setting Editor.
3. Enter the following information in the General tab:



4. Select the **Samples** tab, then expand the window (click-drag the window border) to display the Column, Filtering and Content columns in the Column Settings list:

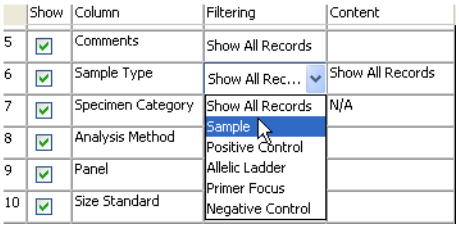


Click-drag border to resize window

5. Click **Hide All** to deselect all of the default table column selections in the Column Settings list.

6. Select to **Show** (☑) the individual Samples table columns you wish to display using this custom table setting.

7. Click the **Filtering** field for the Sample Type row, then select **Sample** from the drop-down list.



Note: When you view the Samples table using this table setting, the Samples table displays only sample files of sample type Sample, and not Allelic Ladder or Control sample types. You can then export this filtered view of the Samples table (as described in Chapter 7).

8. Select additional filters to apply to the remaining columns in the Samples table, if desired.

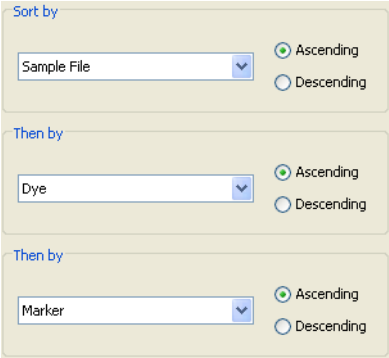
9. In the Sort by drop-down list, select **Run Date & Time**.

Note: Each sample file records the run date and time for that individual sample. When you view the Samples table using this table setting, the sample files sort by order of injection rather than the default sort option of by Sample File.

10. Select the **Genotypes** tab, then follow the same procedures outlined in steps 5 through 8 above to select the columns to display and filter in the Genotypes table.

Note: The settings you specify in the Genotypes tab determine the columns displayed in the Genotypes table in the Project window, and the Genotypes table in the Samples plot.

11. Verify the default sort options for the Genotypes table.



Note: The GeneMapper® *ID-X* Software executes the selected sort options in succession. By default, markers are sorted within a sample by dye color (B, G, Y, R), then by size.

12. Click **OK** to apply your changes and close the Table Setting Editor. The GeneMapper ID-X Manager remains open.

Review Default Plot Settings

Overview

Plot settings determine the number of panes, headers, labels and tables displayed in the Samples and Genotypes plot windows. The default plot settings shown in this section include a specific set of display elements required to perform different analysis workflow tasks. These elements are designed for efficient data review.

Reviewing Plot Settings

1. Open the GeneMapper ID-X Manager if not already open (see page 30).
2. In the GeneMapper ID-X Manager, select the **Plot Settings** tab.
3. Click on a table row to select the plot setting you wish to review, then click **Open**.
4. Review the default plot settings provided using Table 3 on page 41.

5. Click **OK** to close the Plot Settings Editor after reviewing all plot settings. The GeneMapper ID-X Manager will remain open.

Defaults Provided

The following plot settings are installed with the GeneMapper® *ID-X* Software.

Table 3 Default plot settings

Plot Setting		
Name	**Purpose**	**Display Settings**
Check LIZ Size Standard	Used to display the GeneScan™ LIZ® size standard in the same format as the Check GS500 Macro in the Genotyper® Software templates.	Displays the GeneScan™ LIZ® size standard fragments with labels per sample in separate electropherogram panes.
Check ROX Size Standard	Used to display the GeneScan™ ROX™ size standard in the same format as the Check GS500 Macro in the Genotyper® Software templates.	Displays the GeneScan™ ROX™ size standard fragments with labels per sample in separate electropherogram panes.
Data Interpretation and Data Interpretation 6_dye	Used during manual review of sample data, to enable quick interpretation of anomalies and marker-level quality values.	Displays the electropherogram plots for the selected sample(s), the Genotypes table, and the Quality Value Details (QVD) pane. **Note:** If small markers and low-molecular weight microvariant alleles are not displayed in plots, Edit plot settings to decrease the starting size range (for example, to 60 bp).
Mixture Analysis Defaults	Used for Mixture Analysis.	Includes the sample-level PQV MIX (Mixed Source) and selected and unselected genotype combinations.
Overlay LIZ Dye	Used to perform sizing precision checks with the GeneScan™ LIZ® size standards.	Overlays all selected size standard fragments in one electropherogram pane, and displays the Sizing table.
Overlay ROX Dye	Used to perform sizing precision checks with the GeneScan™ ROX™ size standards.	Overlays all selected size standard fragments within a project in one electropherogram pane, and displays the Sizing table.
Sizing Data	Used to display data in a format similar to the GeneScan™ Software plots.	Displays all dyes per sample in one electropherogram pane, and the Sizing table.

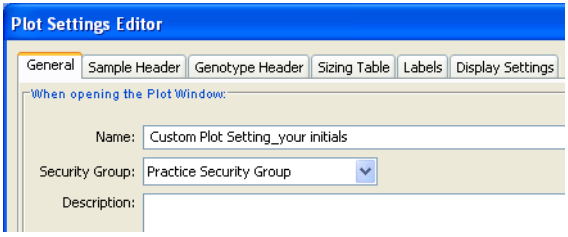
Table 3 Default plot settings *(continued)*

Plot Setting		
Name	**Purpose**	**Display Settings**
Traditional Genotype Plot and Traditional Genotype Plot 6_dye	Used to display data in a format similar to the Genotyper® Software plots.	Displays each dye for a sample in a separate electropherogram pane.
View Label Edits	Used to display allele edits for the selected sample(s) in a table below the electropherogram for electronic data review.	Displays the electropherogram plots for the selected sample(s), and the Label Edit Viewer table.

Creating a New Plot Setting

Custom plot settings save frequently used combinations of display elements to minimize the time spent adjusting display settings during manual review.

1. Open the GeneMapper ID-X Manager if not already open (see page 30).
2. In the GeneMapper ID-X Manager, select the **Plot Settings** tab, then click **New**.
3. Enter the following information in the General tab of the Plot Setting Editor:



4. Select the **Sample Header** tab, then select the columns to display in the Samples plot header.

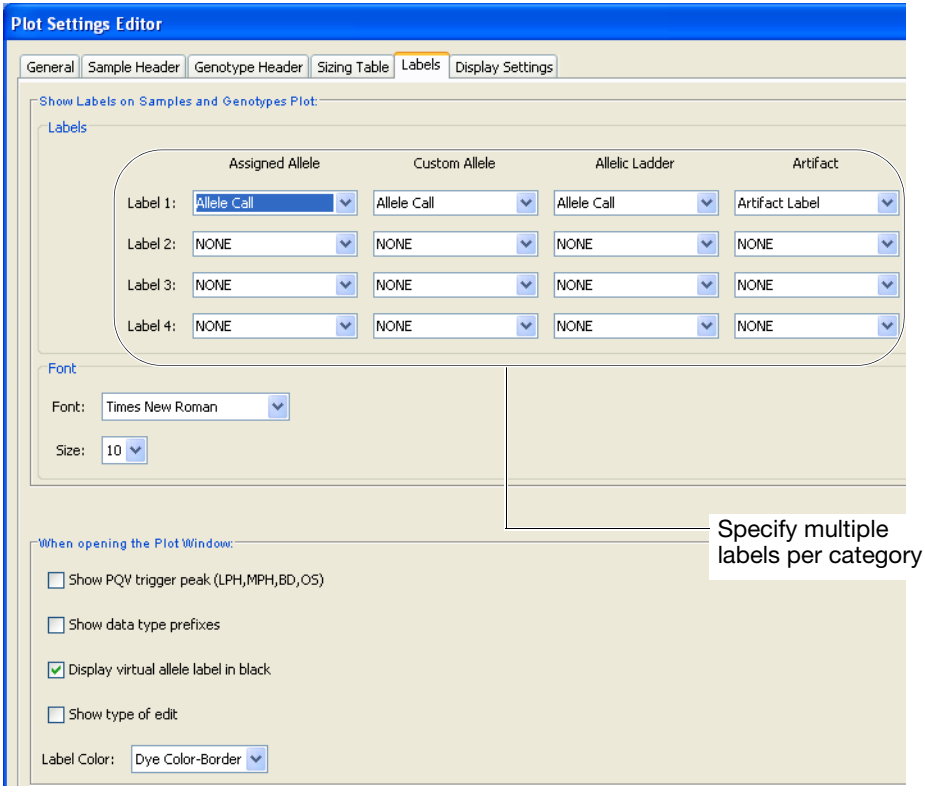
5. Select the **Genotype Header** tab, then select the columns to display in the Genotypes plot header.

Note: The columns in the Genotypes table displayed at the bottom of the Samples plot are determined by the table settings selected in the Project window.

---

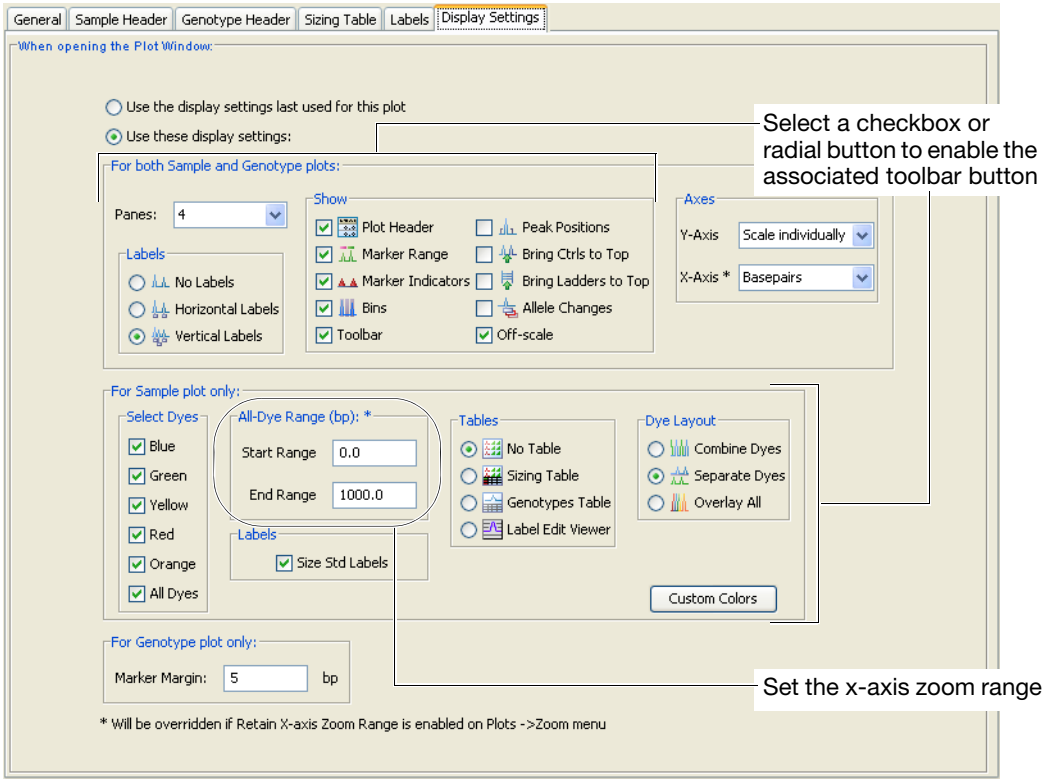
6. Select the **Sizing Table** tab, then select the columns to display and the font and font size to use for the Sizing table displayed at the bottom of the Samples plot.

7. Select the **Labels** tab, then select the label information to display for the detected peaks in the Samples and Genotypes plots:



This tab allows you to choose a different set of labels for different types of samples and different types of peak labels. For instance, you may choose to only display the allele call for all allelic ladder samples, but you want to display both the allele call and height on all other sample types and have them both be displayed in the same plot window. Each peak can have up to four labels.

8. Select the **Display Settings** tab, then specify settings:



Click **Help** at the bottom of the tab and navigate to the *Display Settings Tab* help topic to learn more about the parameters presented in this tab.

9. Click **OK** to apply your changes and close the Plot Setting Editor. The GeneMapper ID-X Manager remains open.

Review Default Size Standards

Overview

A size standard definition file provides a list of fragment sizes in base pairs and the dye color associated with a particular size standard. During peak detection and size-calling, the GeneMapper® *ID-X* Software matches an observed fragment peak from the size standard run with the sample with a corresponding size in the definition file.

Reviewing Size Standard Settings

1. Open the GeneMapper ID-X Manager if not already open (see page 30).
2. In the GeneMapper ID-X Manager, select the **Size Standards** tab.
3. Click a table row to select the size standard setting to review, then click **Open**.
4. Review the default size standard settings provided using Table 4 on page 46.
5. When you are finished reviewing all size standard settings, click **Done** to close the GeneMapper ID-X Manager.

Note: You can also create a new size standard definition using the GeneMapper ID-X Manager.

Defaults Provided

The following default size standard definition files are provided with the GeneMapper® *ID-X* Software for analysis of AmpFℓSTR® kit data:

Table 4 Default size standards

Size Standard	Description	When to Use
CE_G5_HID_GS500	Includes fragments present in the GeneScan™ 500 LIZ® size standard (75 to 450-bp), excluding the 250-bp fragment.	Use with data run with the GS500 LIZ® Size Standard.

Table 4 Default size standards *(continued)*

Size Standard	Description	When to Use
CE_F_HID_GS500 (75-400)	Includes fragments present in the GeneScan™ 500 ROX™ size standard (75 to 400-bp), excluding the 250-bp fragment.	Use with data run with the GS500 ROX™ Size Standard and all AmpFℓSTR® 4-dye kits (except the SGM Plus® kit).
CE_F_HID_GS500 (75-450)	Includes fragments present in the GeneScan™ 500 ROX™ size standard (75 to 450-bp), excluding the 250-bp fragment.	Use with data run with the GS500 ROX™ Size Standard and the AmpFℓSTR® SGM Plus® kit.
GS600_LIZ, GS600_LIZ (60-460), and GS600_LIZ (80-400)	Includes fragments present in the GeneScan™ 600 LIZ® size standard in the following ranges: 80 to 460-bp, 60 to 460-bp, and 80 to 400-bp.	Use with data run with the GS600 LIZ® Size Standard.
GS600_LIZ+Normalization_(60-460), and +Normalization_GS600_LIZ (80-400)	Includes fragments present in the GeneScan™ 600 LIZ® size standard in the following ranges: 60 to 460-bp, and 80 to 400-bp.	Use with data generated on a 3500 Series instrument (with the normalization function enabled) and run with the GS600 LIZ® Size Standard.

Step 5: Import Lab Reference and Custom Control Profiles

Overview

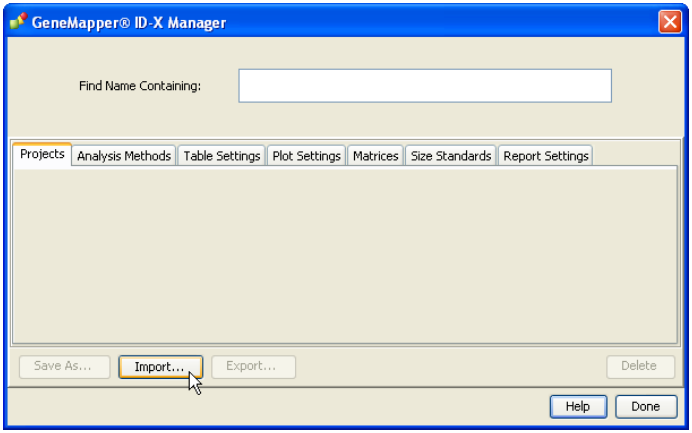
In this section, you will import a project that contains analyzed lab reference samples, custom controls, and QC samples that are used to illustrate the quality control features of the GeneMapper® *ID-X* Software (see Chapter 5). This project has been edited to remove any off-ladder (OL) labels.

Note: For information on importing profiles from a .txt or .csv file instead of a project, refer to the *GeneMapper® ID-X Software Help*.

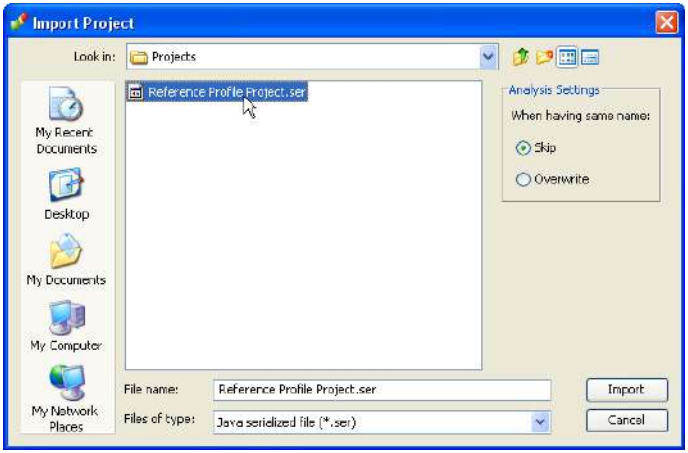
IMPORTANT! Before adding your own lab reference and custom control samples using the procedure described below, review the samples manually and edit allele labels as needed to ensure that the profile is accurate. Profiles that include OL labels are not imported into the Profile Manager. Profiles that include numeric allele labels on peaks that are not true DNA peaks will affect concordance results.

Importing the Reference Project

1. Open the GeneMapper ID-X Manager if not already open (see page 30).
2. In the GeneMapper ID-X Manager, select the **Projects** tab, then click **Import**.



3. Navigate to and select **Reference Profile Project.ser**, then click **Import**.



Note: The reference project file path shown above is for Full install configurations only. For the reference project file path for Client install configurations, see "Using the Guide with the Example Data Provided" on page 18.

4. Make sure the Practice Security Group is selected, then click **OK**.

5. Click **Done** to close the GeneMapper ID-X Manager.

Adding Profiles to the Software Database

1. In the Project window, click (Open Project).
2. Select **Reference Profile Project**, then click **OK**.
3. From the Table Setting drop-down list, select **Import Reference Profiles**. The Sample table view changes to display only those columns required to add reference profiles to the GeneMapper® *ID-X* Software database.

   Note: All genotypes and edits are saved with the imported project. This project does not require re-analysis.

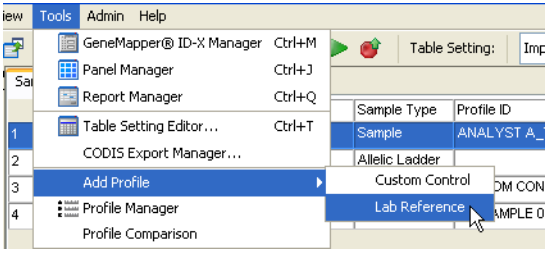
4. In the Profile ID column of the Samples tab, click each cell, then enter the Profile ID names as shown below.

Samples | Analysis Summary | Genotypes

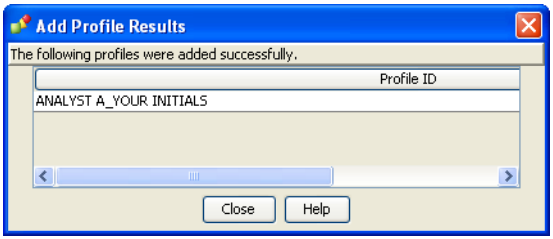
	Status	Sample File	Sample Name	Sample Type	Profile ID
1		Analyst A.fsa	Analyst A	Sample	ANALYST A_YOUR INITIALS
2		ID_AllelicLadder.fsa	Ladder	Allelic Ladder	
3		ID_CustomControl.fsa	Custom Control	Sample	CUSTOM CONTROL_YOUR INITIALS
4		QC_Sample_01.fsa	QC Sample 01	Sample	QC SAMPLE 01_YOUR INITIALS

Note: Profiles are stored in the GeneMapper® *ID-X* Software database under Profile ID, not Sample Name.

5. Select the **Analyst A.fsa** row, then select **Tools ▸ Add Profile ▸ Lab Reference**.



6. Click **Close** in the Add Profile Results dialog box to save the assigned lab reference profile to the GeneMapper® *ID-X* Software database.

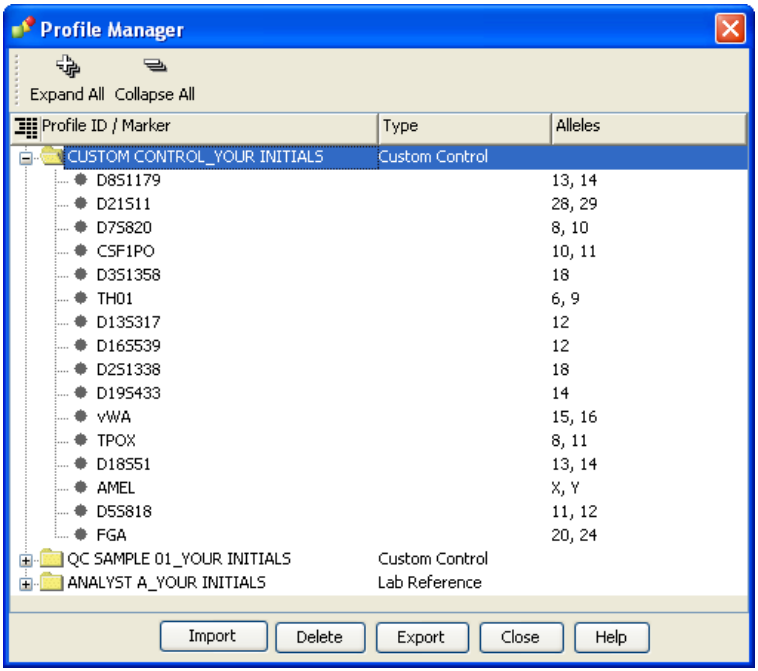


7. Shift-click to select the **ID_CustomControl.fsa** and **QC_Sample_01.fsa** rows, then select **Tools ▸ Add Profile ▸ Custom Control**.

8. Click **Close** in the Add Profile Results dialog box to save the assigned custom control profiles to the GeneMapper® *ID-X* Software database.

Viewing Profiles in the Profile Manager

1. In the Project window, select **Tools ▸ Profile Manager**.
2. View the list of profiles in the Profile Manager window. Click ⊞ to expand at least one Profile ID to view the genotypes stored in the GeneMapper® *ID-X* Software database.



3. Click **Close** to close the Profile Manager window and return to the Project window.

Step 6: Set the Project Options

Overview

Project Options are user-specific project preferences that allow you to specify default security and analysis options for your user account.

In This Section

In this section, you will set the software to:

- Automatically assign a security group (available selections determined by user account) when you create a project.
- Automatically assign analysis settings when you add samples to the project.
- Display the Analysis Requirements Summary (ARS) to identify any conditions that could prevent analysis.
- Automatically disregard low-quality allelic ladders and proceed with the analysis of run folders containing one or more passing allelic ladders.
- Display the Analysis Summary, which provides a snapshot of the analysis status of all samples in the project and the quality status of allelic ladders, controls, and samples.

Note: These options are suggested for an optimized, efficient data review workflow. However, you can modify project options as needed to meet your laboratory workflow requirements.

Setting Project Options

Project options are associated with the user account currently logged in to the GeneMapper® *ID-X* Software (in this example, the Practice User).

Note: Set project options when you obtain your personal or lab-specific user account. For more information on the GeneMapper® *ID-X* Software security system, see the *GeneMapper® ID-X Software Administrator's Guide*.

1. Select **File ▸ Project Options**.
2. Complete the Options tabs for the Practice User as described in Table 5 on page 54, using the *GeneMapper® ID-X Software Help* as a guide.

Table 5 Practice User project options settings

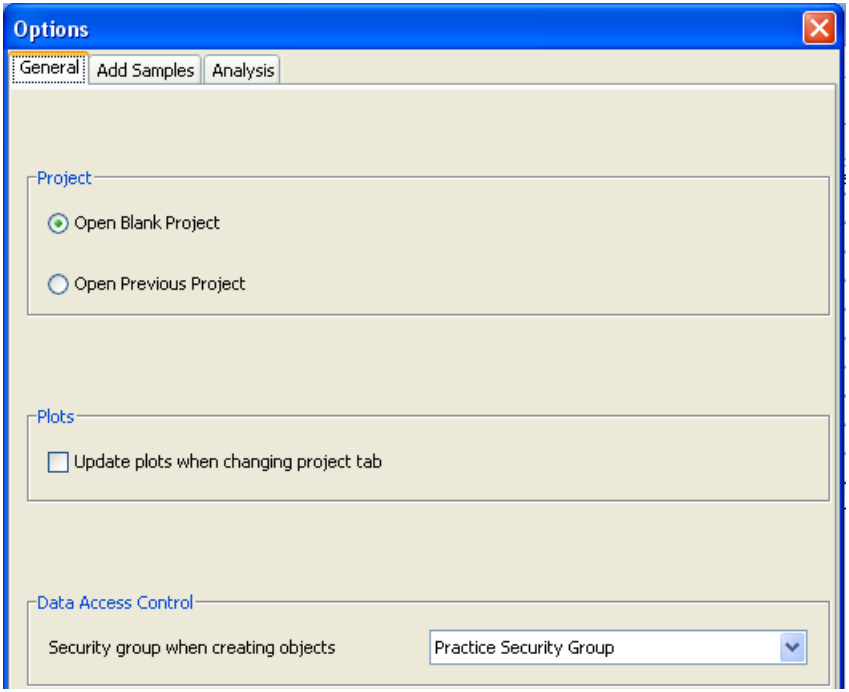
Tab	Settings
General	Verify the settings as shown below: 

Table 5 Practice User project options settings *(continued)*

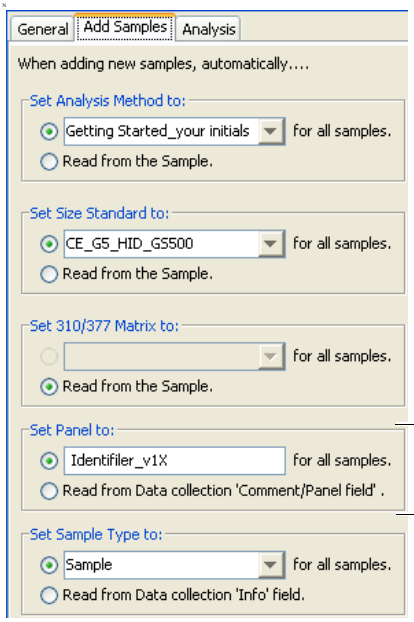
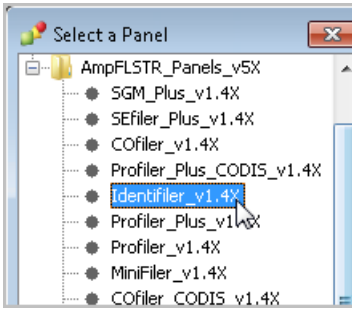
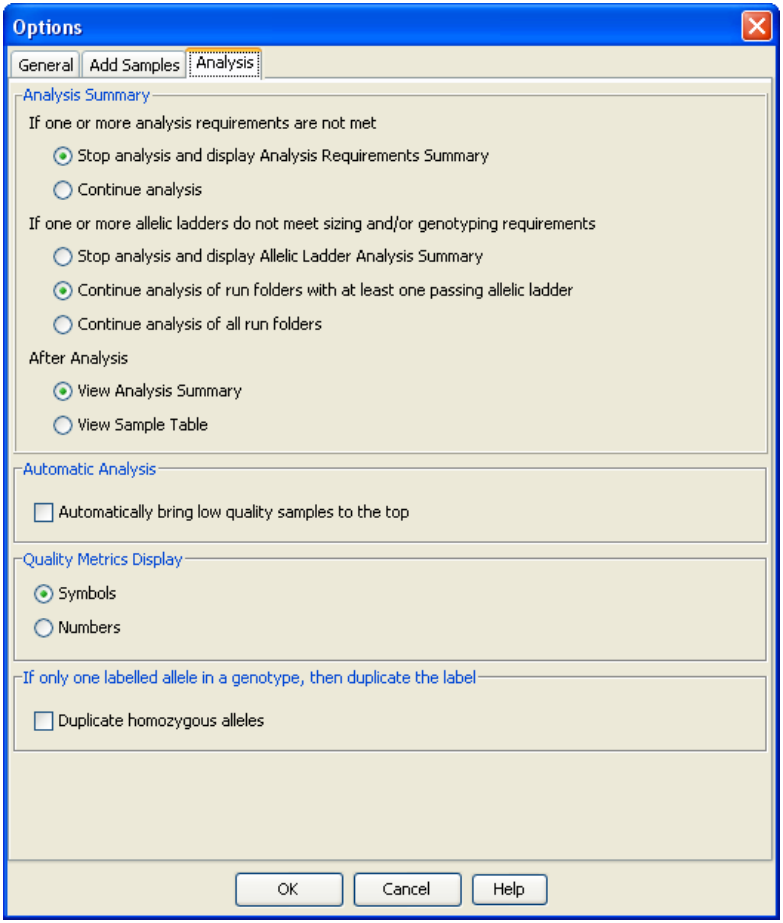
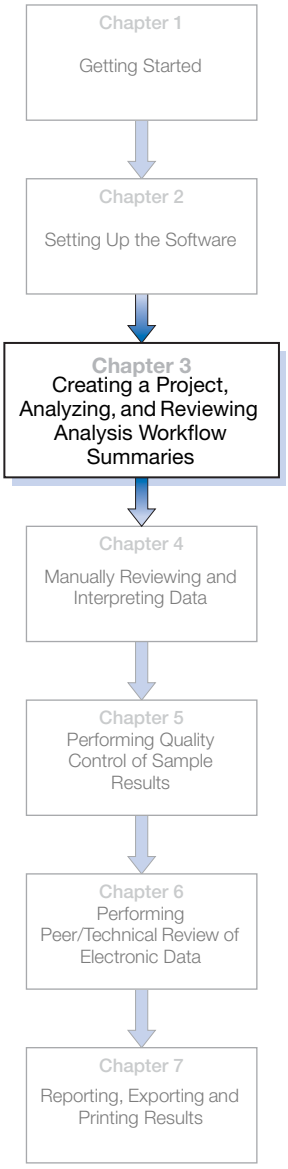
Tab	Settings
Add Samples	Enter the settings as shown below:  To set a panel for all samples: • Click in the text field to open the Select a Panel dialog • Click ⊞ to view the available AmpFℓSTR® panels, then select the **Identifiler_v1.4X** panel  • Click **OK** to close the dialog

Table 5 Practice User project options settings *(continued)*

Tab	Settings
Analysis	Click **Help** at the bottom of the tab to learn more about the options presented in the Analysis Summary area of this tab, then enter the settings as shown below: 

3. Click **OK** after completing all tabs to close the Options dialog and return to the Project window.

3 Creating a Project, Analyzing, and Reviewing Analysis Workflow Summaries



This chapter covers:

- Overview. 58
- Step 1: Create a Project and Add Samples 58
- Step 2: View Sample Information and Raw Data 61
- Step 3: Select Analysis Settings and Start Analysis 65
- Step 4: Review the ARS and Correct the Requirements 66
- Step 5: Analyze the Data 68
- Step 6: Review the Analysis Summary. 70

Overview

During analysis, the GeneMapper® *ID-X* Software performs the following tasks, based on the analysis settings and project options set up in Chapter 3:

- Analysis requirements check
- Data analysis (peak detection and sizing, allele-calling)
- Allelic ladder and sample quality assessment
- Analysis workflow summary generation

This chapter will demonstrate these tasks using the example data set provided with the software.

In This Chapter

In this chapter, you will learn how to:

- Create a project and add samples
- View sample details
- Select analysis settings
- Analyze the data
- Review the analysis workflow summaries (Analysis Requirements Summary, Analysis Summary)

Step 1: Create a Project and Add Samples

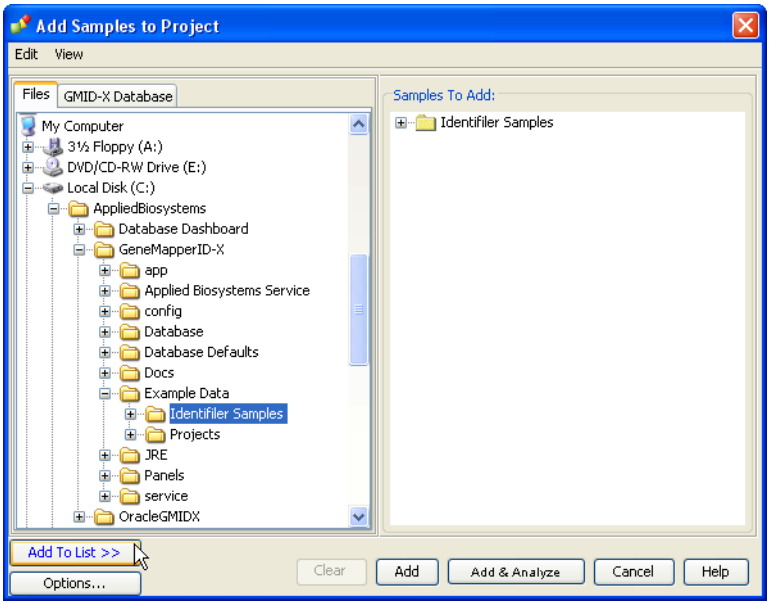
Creating a New Project

If a project is already open in the Project window, click (New Project).

Note: If you have a previous project open you may be prompted to save changes. Click **Yes**.

Adding Samples from Sample Files

1. In the new Project window, click (Add Samples to Project).
2. In the Files tab of the Add Samples to Project dialog box, navigate to the folder containing the example data, select the **Identifiler Samples** folder, then click **Add to List**.



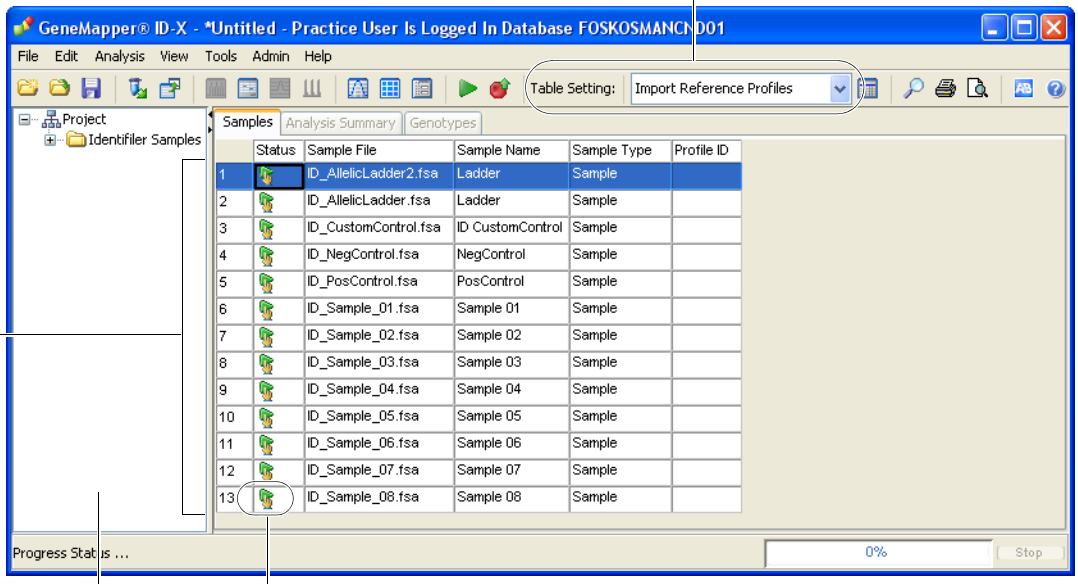
Note: The example data file path shown above is for Full install configurations only. For the example data file path for Client install configurations, see "Using the Guide with the Example Data Provided" on page 18.

3. Click **Add**. When you add samples from sample (.fsa) files to a project:
   - You specify the location of the .fsa files on the hard drive or a network drive.
   - The sample files remain in their original location on the drive, and are not stored in the GeneMapper® *ID-X* Software project or database.

- The GeneMapper® *ID-X* Software reads the information it needs from the .fsa files. No information is written back to the original sample files.
- The added samples are displayed in the Samples table in the content pane of the Project window. The run folder from which you added the samples is displayed in the navigation pane.

Sample table lists all files in selected run folder (only one run folder in this example). Analysis settings are loaded by default as specified in Project Options.

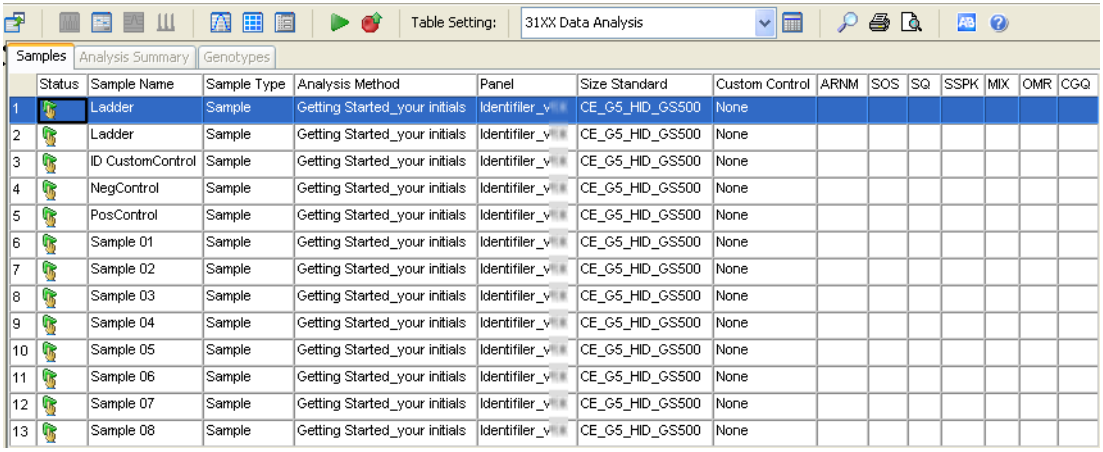
Table Setting selected determines the columns displayed in the Samples table



Navigation pane lists run folders for added samples

indicates samples have not been analyzed

4. In the Project window, select **31XX Data Analysis** from the Table Setting drop-down list. Only the columns needed for analysis of 3130 Series Genetic Analyzer data are displayed in the Samples table.



	Status	Sample Name	Sample Type	Analysis Method	Panel	Size Standard	Custom Control	ARNM	SOS	SQ	SSPK	MIX	OMR	CGQ
1		Ladder	Sample	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	None							
2		Ladder	Sample	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	None							
3		ID CustomControl	Sample	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	None							
4		NegControl	Sample	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	None							
5		PosControl	Sample	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	None							
6		Sample 01	Sample	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	None							
7		Sample 02	Sample	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	None							
8		Sample 03	Sample	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	None							
9		Sample 04	Sample	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	None							
10		Sample 05	Sample	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	None							
11		Sample 06	Sample	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	None							
12		Sample 07	Sample	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	None							
13		Sample 08	Sample	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	None							

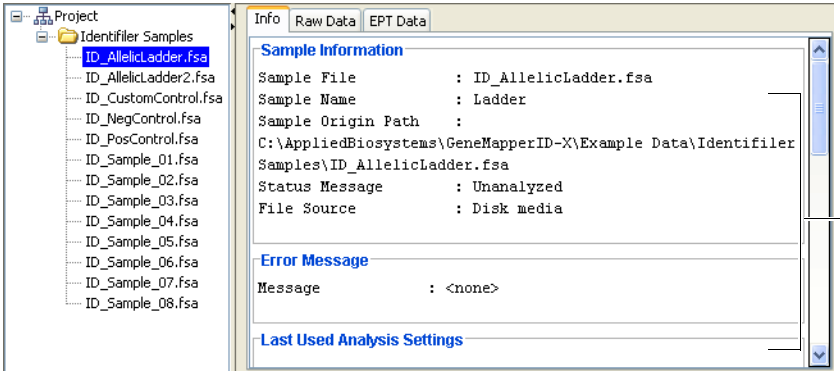
Step 2: View Sample Information and Raw Data

You have access to the following sample information from the Project window:

- Sample file, analysis and run parameters
- Raw data
- Electrophoresis, power and temperature (EPT) data

Viewing Sample Information

1. Click ⊞ to expand the Identifiler Samples folder in the navigation pane, then select the **ID_AllelicLadder.fsa** sample. The Info tab for the selected sample is displayed in the content pane:



The Sample Information, Error Message and Last Used Analysis Settings areas are updated each time you analyze

2. Click the vertical scroll bar at the right of the Info tab window to review the following *sample-specific* information presented in this tab:

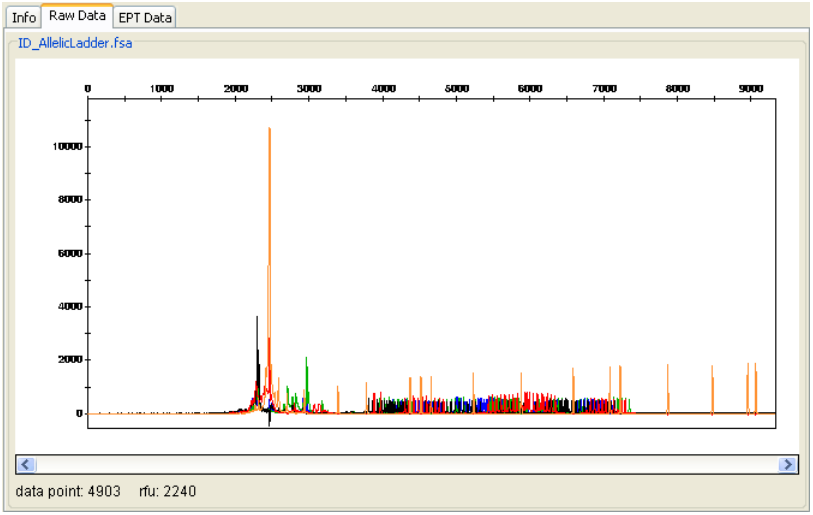
Info Type	Info Listed for Selected Sample
Sample Information	• Sample file name and sample name • Sample origin path and file source • Status message indicating any changes made to the sample in the Samples table
Error Message	• Errors encountered during analysis (if any)
Last Used Analysis Settings	• Last settings used for analysis **Note:** This area will be blank if the sample Status is (Unanalyzed). After analysis, the settings last used to analyze the sample file are displayed in this tab regardless of the analysis settings selected in the Samples table.

Info Type	Info Listed for Selected Sample
Run Information	• Instrument user name • Instrument name and Data Collection version • Run date (in yyyy:dd:mm) and time (in hr:min:sec) • Run duration • Total data points **Note:** All sample files created during one injection on a multi-capillary instrument (a set of 4 or 16 capillaries) will have the same run date, run time, and injection time.
Data Collection Settings	• Run voltage and injection voltage (in volts) • Injection duration (in milliseconds) • Laser power (in mW) and temperature (in °C) • Run module and run protocol name • Dye set name • Polymer lot number and expiration date • Results Group name • 3500 data: Consumables lot, type, expiration date, and installation date.
Capillary Information	• Length and number of capillaries • Capillary number used for injection

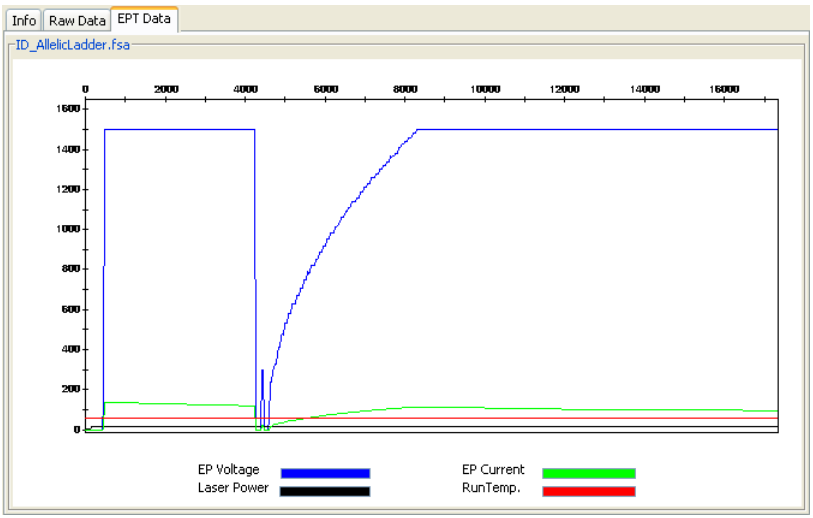
Viewing Raw Data

You can use the raw data view of a sample to help evaluate any anomalies, the causes of poor size-calling, and to determine the start and stop points for analysis.

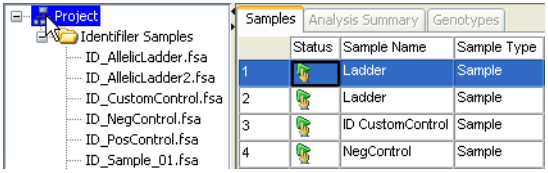
1. Select the **Raw Data** tab in the content pane. The raw data plot for the ID_AllelicLadder.fsa sample is displayed.



2. Select the **EPT Data** tab in the content pane. The EPT plot is displayed.



3. Select the **Project** node in the navigation pane to return to the Samples table view.



Step 3: Select Analysis Settings and Start Analysis

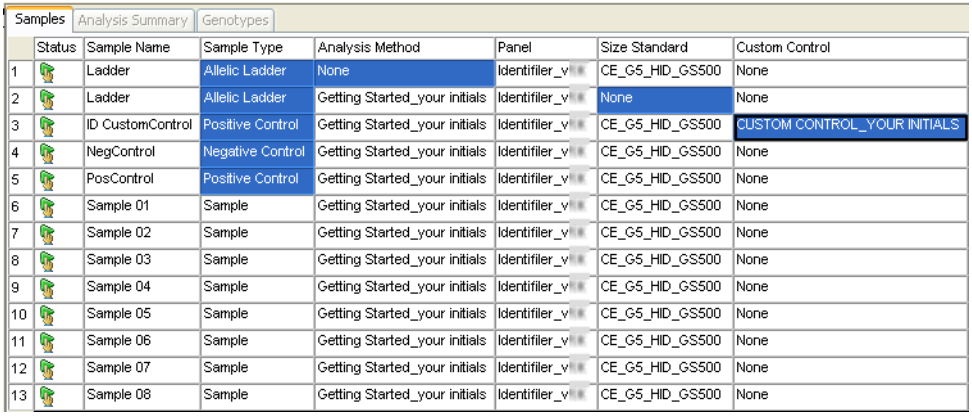
Analysis settings include the Analysis Method, Size Standard, Panel, and Sample Type selections needed to perform analysis. Based on the Project Options you set in Chapter 2, the samples you added to the Samples table automatically have analysis settings specified.

Selecting Analysis Settings

Note: In this section, you will intentionally alter some of the analysis settings in the Samples tab to trigger the display of the ARS when you start the analysis.

1. In the Project window, make sure **31XX Data Analysis** is selected from the Table Setting drop-down list.

2. Enter the Samples tab analysis settings as shown below:



Samples | Analysis Summary | Genotypes

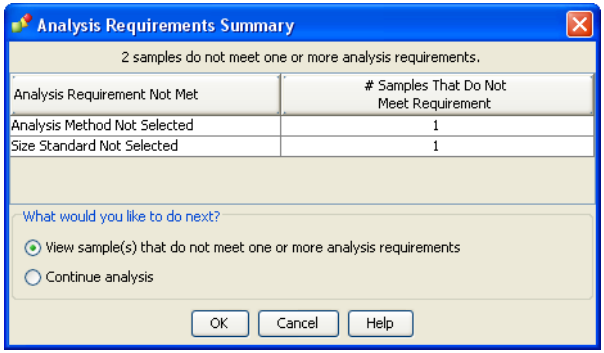
	Status	Sample Name	Sample Type	Analysis Method	Panel	Size Standard	Custom Control
1		Ladder	Allelic Ladder	None	Identifiler_v	CE_G5_HID_GS500	None
2		Ladder	Allelic Ladder	Getting Started_your initials	Identifiler_v	None	None
3		ID CustomControl	Positive Control	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	CUSTOM CONTROL_YOUR INITIALS
4		NegControl	Negative Control	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	None
5		PosControl	Positive Control	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	None
6		Sample 01	Sample	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	None
7		Sample 02	Sample	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	None
8		Sample 03	Sample	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	None
9		Sample 04	Sample	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	None
10		Sample 05	Sample	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	None
11		Sample 06	Sample	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	None
12		Sample 07	Sample	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	None
13		Sample 08	Sample	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	None

Starting Analysis

Click (Analyze).

When analysis is started, the software identifies any conditions that may prevent analysis or cause unexpected results, sets a flag for Analysis Requirements Not Met (ARNM) PQV and displays the ARS if chosen in the Project Options. In this example, we have chosen to display the ARS.

The Analysis Requirements Summary dialog box opens because at least one sample in the project does not meet one or more analysis requirements.



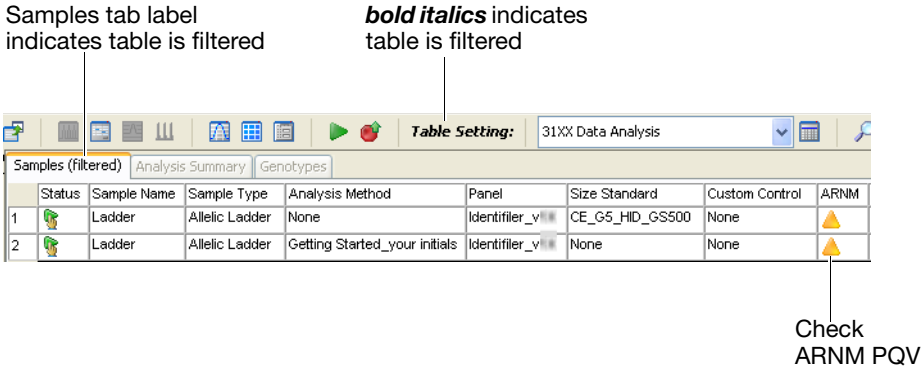
Step 4: Review the ARS and Correct the Requirements

Reviewing the ARS

From the ARS, you may view the samples that do not meet the analysis requirements or you can continue with analysis. In this section, you will view the flagged samples in the example project.

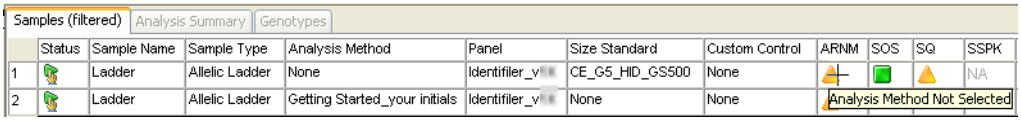
1. Keep the default selection in the *What would you like to do next?* area of the ARS, then click **OK**.

   The Samples table opens. If one or more analysis requirements are not met, the ARNM PQV is set to . Based on the analysis settings changes you made in step 2 on page 65, note that only the allelic ladder samples in the example project are listed with a ARNM flag in the Samples table.



When you view the Samples table from the ARS, the Samples table is filtered. The current table setting is applied to determine which columns are displayed, but only the samples in the category selected are listed. In this example, samples that do not meet the analysis requirements are listed. The filtered condition of the table is indicated by the status of the Samples tab label (filtered) and the Table Setting label (***bold italics***).

2. Place the pointer over a ARNM flag to display a tooltip with analysis requirement information for each sample in the filtered Samples table.



Note that the analysis Status for the samples is still (Unanalyzed).

Correcting the Analysis Requirements

To correct the unmet requirements listed in the ARS, change the analysis settings in the Samples table back to their original values:

1. Change the Analysis Method for the first Ladder sample to **Getting Started**.
2. Change the Size Standard for the second Ladder sample to **CE_G5_HID_GS500**.

   The updated Samples table analysis settings should be:

Samples (filtered) | Analysis Summary | Genotypes

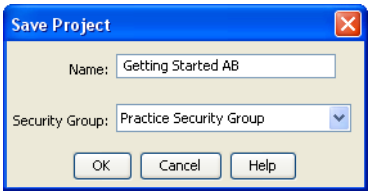
	Status	Sample Name	Sample Type	Analysis Method	Panel	Size Standard	Custom Control
1		Ladder	Allelic Ladder	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	None
2		Ladder	Allelic Ladder	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	None

Step 5: Analyze the Data

Analyzing the Data

In this section, you will analyze with all analysis requirements satisfied and display the Analysis Summary.

1. Click (Analyze).
2. Complete the fields in the Save Project dialog box:
   a. For Name, type **Getting Started <your initials>**. For example, **Getting Started AB**.
   b. Verify that the Practice Security Group is selected from the drop-down list.



   c. Click **OK** to save the project to the GeneMapper® *ID-X* Software database and start analysis.

Viewing Analysis Progress

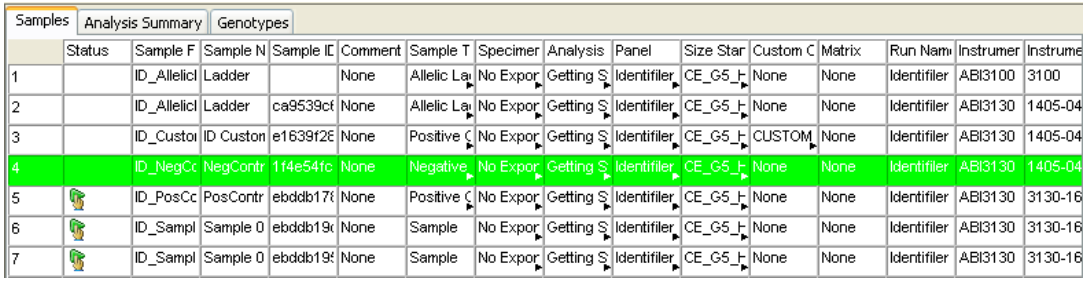
After saving the project to the database:

- The project name (entered in step 2a on page 68) appears in the title bar of the Project window. For example:

- The software then begins analysis.

Note the following after analysis begins:

- Allelic ladder sample types are analyzed before all other sample types in the project. The sample currently being analyzed is highlighted in green in the Samples table:



Samples | Analysis Summary | Genotypes

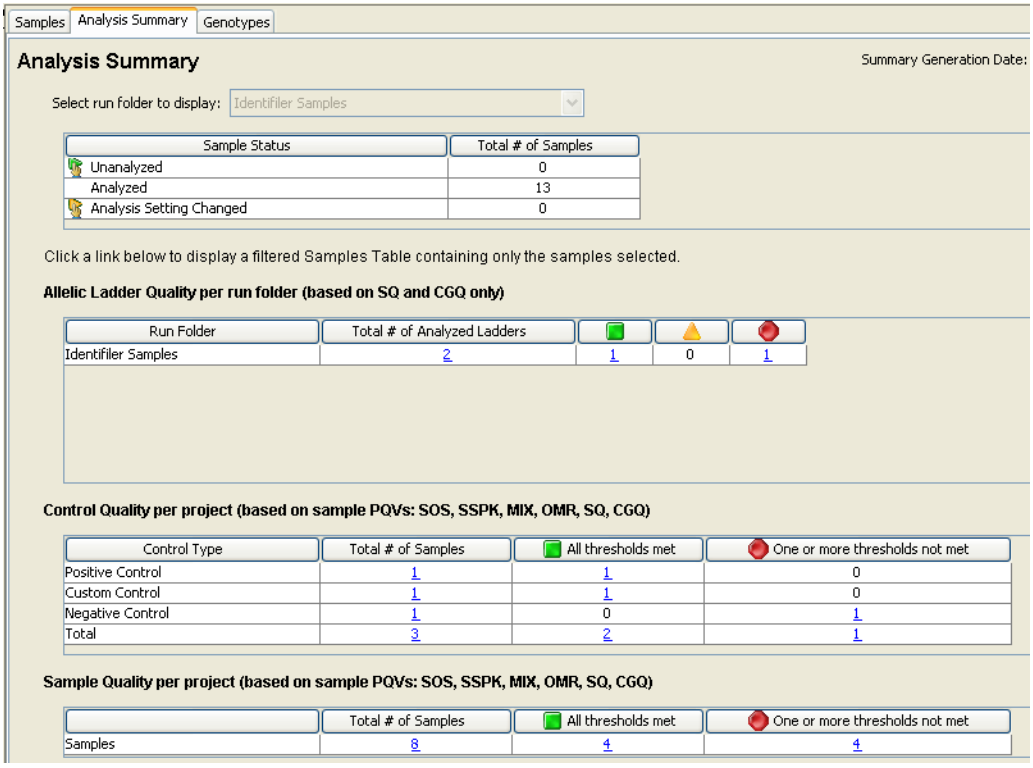
	Status	Sample F	Sample N	Sample ID	Comment	Sample T	Specimen	Analysis	Panel	Size Star	Custom C	Matrix	Run Nam	Instrumer	Instrume
1		ID_Allelicl	Ladder		None	Allelic La	No Expor	Getting S	Identifiler	CE_G5_H	None	None	Identifiler	ABI3100	3100
2		ID_Allelicl	Ladder	ca9539cf	None	Allelic La	No Expor	Getting S	Identifiler	CE_G5_H	None	None	Identifiler	ABI3130	1405-04
3		ID_Custo	ID Custon	e1639f2E	None	Positive C	No Expor	Getting S	Identifiler	CE_G5_H	CUSTOM	None	Identifiler	ABI3130	1405-04
4		ID_NegCo	NegContr	114e54fc	None	Negative	No Expor	Getting S	Identifiler	CE_G5_H	None	None	Identifiler	ABI3130	1405-04
5		ID_PosCo	PosContr	ebddb17(	None	Positive C	No Expor	Getting S	Identifiler	CE_G5_H	None	None	Identifiler	ABI3130	3130-16
6		ID_Sampl	Sample 0	ebddb19(	None	Sample	No Expor	Getting S	Identifiler	CE_G5_H	None	None	Identifiler	ABI3130	3130-16
7		ID_Sampl	Sample 0	ebddb19	None	Sample	No Expor	Getting S	Identifiler	CE_G5_H	None	None	Identifiler	ABI3130	3130-16

- Analysis progress is displayed at the bottom of the Project window:

Step 6: Review the Analysis Summary

About the Analysis Summary Tab

Based on the Project Options you set in Chapter 2, the Analysis Summary tab is displayed when analysis is complete.



For efficient data evaluation, the Analysis Summary tab:

- Provides a summary of the analysis status for all or a subset of samples in the project
- Displays an overview of allelic ladder, control, and sample quality
- Visually separates passing samples from samples that do not meet one or more quality thresholds

- Provides interactive links to specific categories of samples (passing/check/low quality, allelic ladder/control/sample)

IMPORTANT! Refer to your own laboratory protocol to determine the samples (allelic ladder, positive control, negative control, custom control, and unknown) that require manual review when analyzing data generated in your laboratory.

Briefly review the features and areas of the Analysis Summary tab as described below, then continue to Chapter 4, where you will review the functions of this tab in more detail.

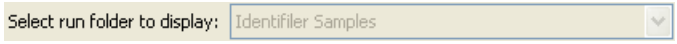
Features of the Analysis Summary Tab

Note the following features of the Analysis Summary tab:

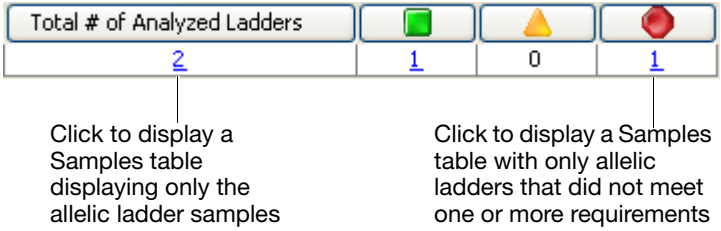
- You can display an Analysis Summary for the entire project or for an individual run folder in the project by selecting a run folder from the drop-down list at the top of the tab

  Note: If the project contains only one run folder, this selection is dimmed.

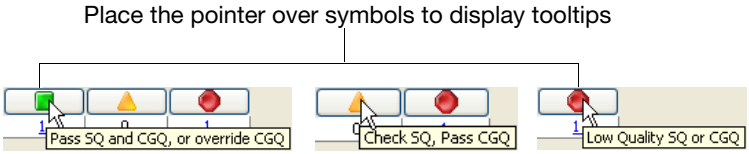
  In this example, Identifiler Samples is the only run folder in the Getting Started project:



- Blue links take you to a Samples table displaying only the samples of interest



- Tooltips explain symbols



Areas of the Analysis Summary Tab

Analysis Status

The Analysis status area at the top of the Analysis Summary tab indicates that all samples in the Getting Started project are analyzed.

Sample Status	Total # of Samples
Unanalyzed	0
Analyzed	13
Analysis Setting Changed	0

Allelic Ladder Quality

The Allelic Ladder Quality area indicates that of the two allelic ladder samples analyzed in the Getting Started project, one met all of the allelic ladder quality requirements ( ) and one did not ( ).

Run Folder	Total # of Analyzed Ladders			
Identifiler Samples	2	1	0	1

You can use the allelic ladder quality results to determine the samples that require manual review. For example, if each run folder contains at least one passing allelic ladder and the positive and custom controls meet all quality value thresholds, visual inspection of the allelic ladders may not be required. Depending on validation, you can proceed directly to evaluation of the controls or samples in the project.

Control Quality

The Control Quality area of the Analysis Summary indicates that of the three controls analyzed in the Getting Started project, the positive and custom controls met all quality thresholds ( ) and generated the expected profile, but the negative control did not.

Control Type	Total # of Samples	All thresholds met	One or more thresholds not met
Positive Control	1	1	0
Custom Control	1	1	0
Negative Control	1	0	1
Total	3	2	1

When validated, this area of the Analysis Summary may eliminate the need for you to visually inspect the control samples if they fall under the green All Thresholds Met column. Controls in this column have met all sample-level and marker-level quality values, including

Control Concordance (CC). This means that the sample genotypes for positive and custom controls match the expected known profile without any anomalies. For negative controls, this means that there were no peaks detected above the peak amplitude threshold.

You can also use the control quality results to verify allelic ladder quality. For example, allelic ladders may meet all requirements, but if their migration rate differs from the sample migration rate, samples and controls may contain OL calls. However, controls that met all thresholds (and therefore do not contain OL calls), may indicate consistent migration rates for allelic ladders and samples.

Sample Quality

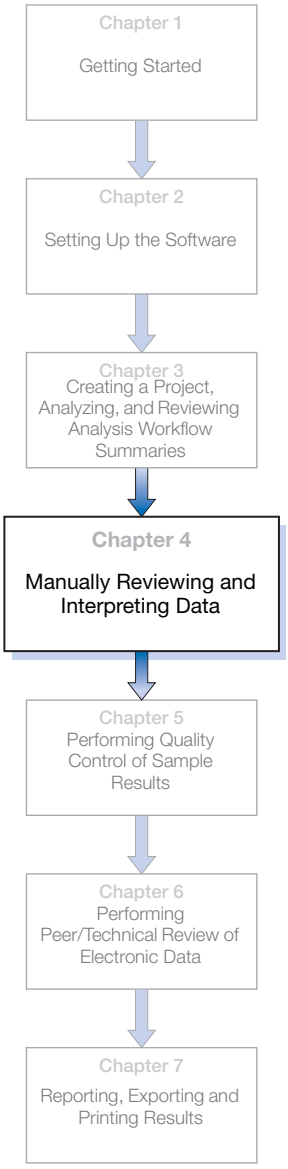
The Sample Quality area of the Analysis Summary indicates that of the eight samples analyzed, four met all quality thresholds ( ) and four did not ( ).

	Total # of Samples	All thresholds met	One or more thresholds not met
Samples	8	4	4

In Chapter 4, you will use the blue links to visually inspect certain categories of samples.

4

Manually Reviewing and Interpreting Data



This chapter covers:

- Overview. 76
- Step 1: Examine Allelic Ladder Quality. 76
- Step 2: Examine Control Quality 85
- Step 3: Examine Sample Quality 90
- Step 4: Review the Low-Quality Sample Results. 92
- Step 5: View Additional Plot Settings 131
- Step 6: Delete Samples from the Project 135
- Step 7: Save the Project 136

Overview

The Analysis Summary discussed in Chapter 3 guides you directly to different types of samples while following a logical data interpretation workflow, regardless of whether the software is being used as an expert system or not. In a traditional manual review workflow, you would most likely visually inspect all unknown sample electropherograms. In a validated expert system workflow, you would only view those samples that did not meet one or more quality value thresholds.

Note: Refer to your own laboratory protocol to determine the controls and samples that require manual review.

In This Chapter

In this chapter, you will review the example data analyzed in Chapter 3 and learn how to:

- Use the Analysis Summary tab and filtered Samples table to examine allelic ladder, control and sample quality
- Investigate sample-level PQVs, and marker-level PQVs using the QVD pane
- Review sample plots and edit peak labels
- Adjust plot displays to determine the source of artifacts

Note: Some steps performed in this chapter are included only to demonstrate the use of certain features in the GeneMapper® *ID-X* Software and may not be a part of your routine analysis workflow.

Step 1: Examine Allelic Ladder Quality

Overview

The order of information displayed in the Analysis Summary is designed to direct you to evaluate the allelic ladder quality first. The designation is based on a set of allelic ladder requirements. If there is at least one passing allelic ladder per run folder and the positive and custom controls meet all quality value thresholds as displayed in the Analysis Summary, you may not require visual inspection of the

allelic ladders and may proceed directly to evaluation of the controls or samples in the project (depending on validation). However, for the purposes of this guide, you will walk through the process for manually reviewing allelic ladder samples.

Viewing Allelic Ladder Quality Status

In Chapter 3, the Allelic Ladder Quality area of the Analysis Summary tab indicates that of the two allelic ladder samples analyzed, one has met all allelic ladder requirements ( ) and one has not ( ).

Allelic Ladder Quality per run folder (based on SQ and CGQ only)

Run Folder	Total # of Analyzed Ladders			
Identifiler Samples	2	1	0	1

Note: Low-quality ( ) allelic ladders are not used to create bin offsets.

Examining the Low-Quality Allelic Ladder

1. If the Getting Started project is not already open, click (Open Project) in the Project window.

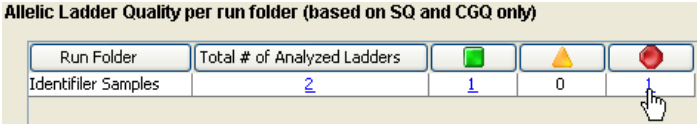
   Note: If you have a previous project open you may be prompted to save changes. Click **Yes**.

2. In the Open Project window, select the **Getting Started <your initials>** project, then click **OK**. The Getting Started project opens in the Project window.

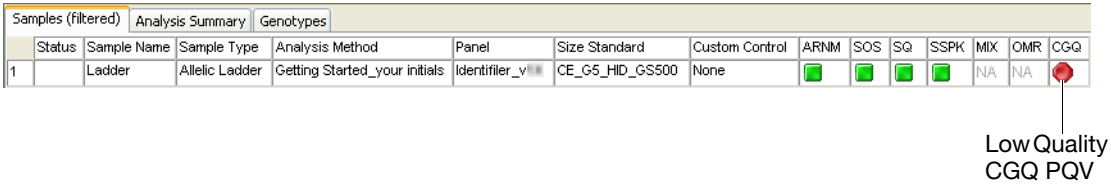
3. In the Samples table, verify that **31XX Data Analysis** is selected from the Table Setting drop-down list.

4. Verify that the Analysis Summary tab is selected in the content pane.

5. In the Allelic Ladder Quality area of the Analysis Summary tab, click the link for the allelic ladder sample.

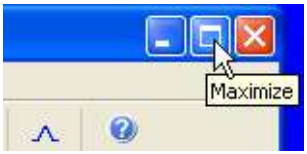


The filtered Samples table displays the selected sample. Note that this low-quality allelic ladder sample has a CGQ PQV.

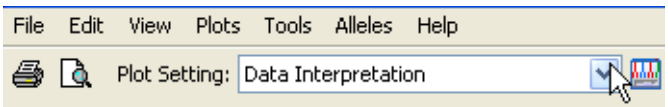


6. Select the **Ladder** sample in the filtered Samples table, then click (Display Plots). The plot for the selected allelic ladder sample opens in the Samples plot.

7. Click (Maximize) in the top-right corner of the Samples plot to maximize the display.

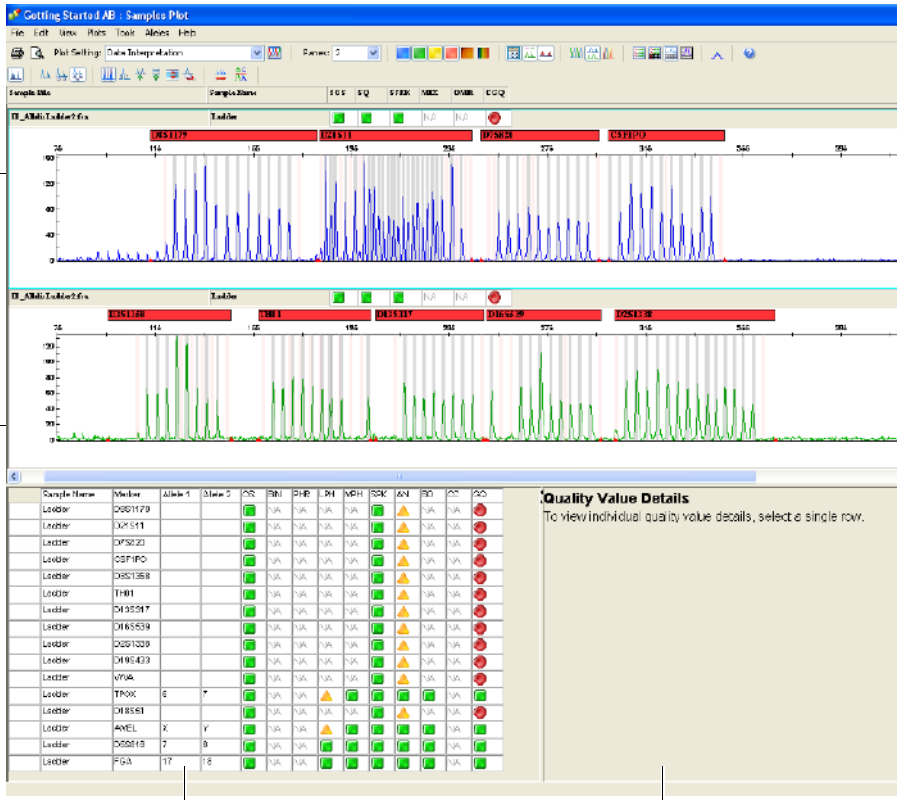


8. Select **Data Interpretation** from the Plot Setting drop-down list.



Based on the display options of this default plot setting (see Chapter 2), the Samples plot displays:

- Two electropherogram (dye) panes in a single view
- Each pane zoomed to the 75 - 450 base pair range
- Each of four dyes (blue, green, yellow, red) in a separate pane
- The Genotypes table and QVD pane displayed under the electropherograms

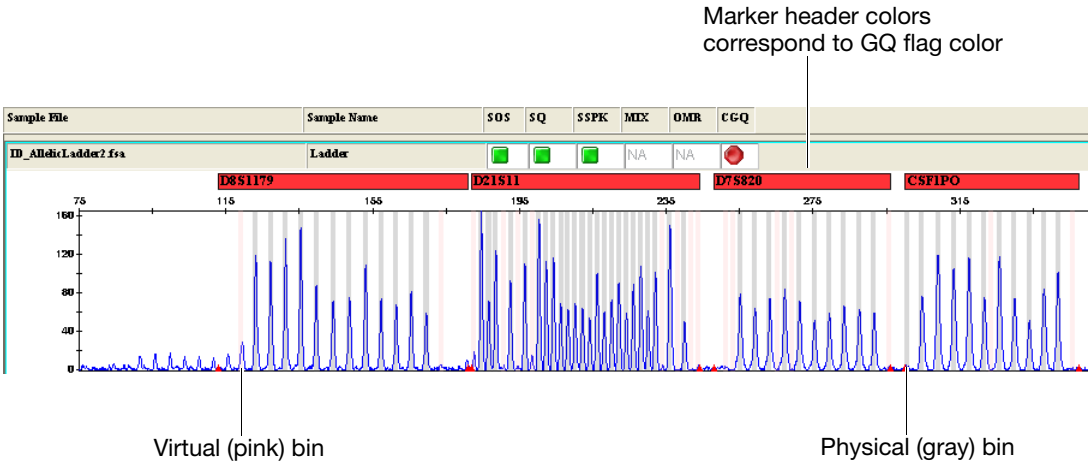


Electropherogram (dye) panes

Genotypes table

QVD pane

Note the appearance of the marker headers and bins:



The marker headers are color-coded (green, yellow, red) to reflect the GQ flag color. Bins are displayed according to bin type (see Chapter 2), with virtual bins in pink and physical bins in gray.

Note: For more information on virtual and physical bins, see the *GeneMapper® ID-X Software Reference Guide*.

9. To investigate the reason why a particular marker failed the allelic ladder quality assessment, click the red **D8S1179** marker header in the blue dye pane to display quality assessment information in the QVD pane for this marker.

Note: The columns displayed in the Genotypes table are determined by the table setting selected in the Project window.

The QVD pane displays the allelic ladder quality requirement that was not met for the selected marker. In this example, one or more of the expected allelic peaks fell below the peak amplitude threshold set for the D8S1179 marker.

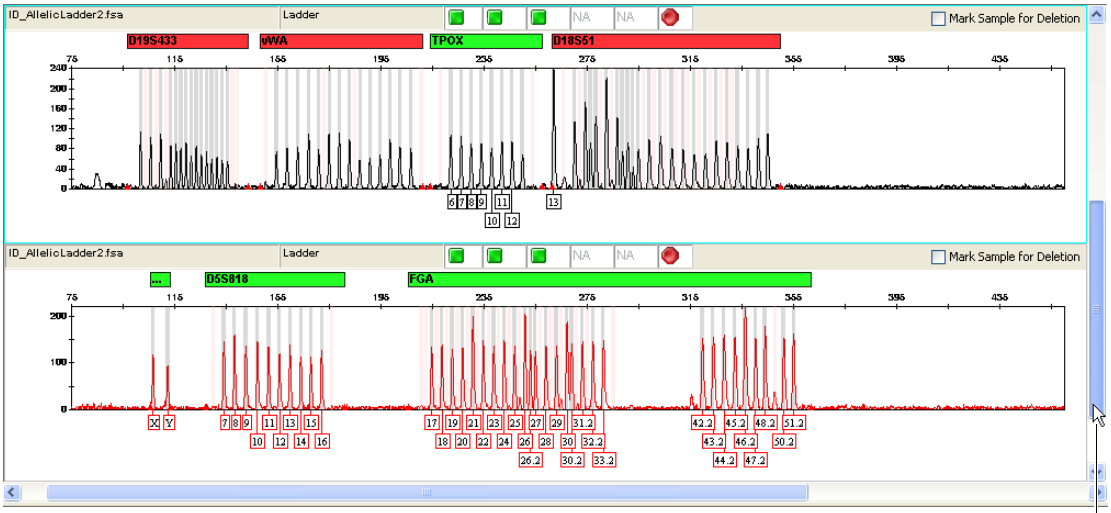
Quality Value Details

GQ = 0.0

One or more allelic ladder requirements have not been met. See below:
- One or more peaks were not detected

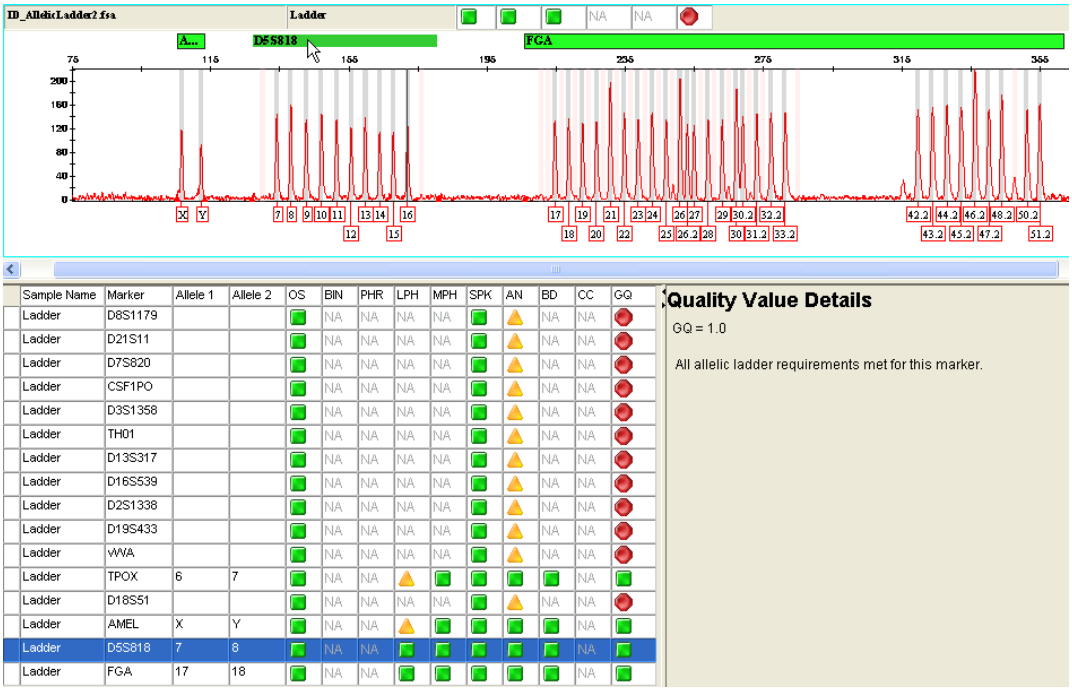
Due to the above reason(s), this ladder has not been used to generate bin offsets.

10. Click the vertical scroll bar at the right of Samples plot until you can see the D5S818 marker in the red dye pane.



Click the scroll bar to display the next electropherogram

11. Click the green **D5S818** marker header to display the quality assessment details for this marker.

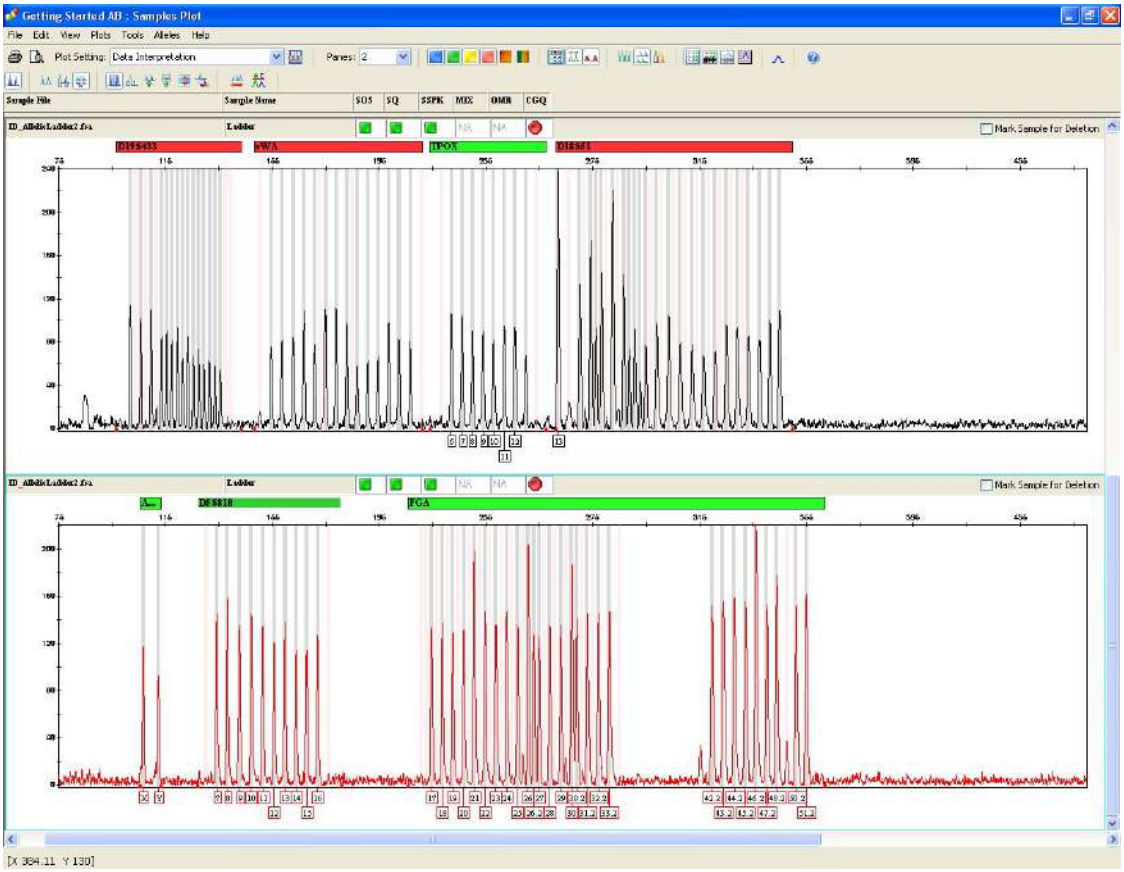


Sample Name	Marker	Allele 1	Allele 2	OS	BIN	PHR	LPH	MPH	SPK	AN	BD	CC	GQ
Ladder	D8S1179				NA	NA	NA	NA			NA	NA	
Ladder	D21S11				NA	NA	NA	NA			NA	NA	
Ladder	D7S820				NA	NA	NA	NA			NA	NA	
Ladder	CSF1PO				NA	NA	NA	NA			NA	NA	
Ladder	D3S1358				NA	NA	NA	NA			NA	NA	
Ladder	TH01				NA	NA	NA	NA			NA	NA	
Ladder	D13S317				NA	NA	NA	NA			NA	NA	
Ladder	D16S539				NA	NA	NA	NA			NA	NA	
Ladder	D2S1338				NA	NA	NA	NA			NA	NA	
Ladder	D19S433				NA	NA	NA	NA			NA	NA	
Ladder	vWA				NA	NA	NA	NA			NA	NA	
Ladder	TPOX	6	7		NA	NA						NA	
Ladder	D18S51				NA	NA	NA	NA			NA	NA	
Ladder	AMEL	X	Y		NA	NA						NA	
Ladder	D5S818	7	8		NA	NA						NA	
Ladder	FGA	17	18		NA	NA						NA	

The QVD pane indicates that all allelic ladder quality requirements are met for the selected marker. Based on the calculated GQ values shown in the QVD pane, this marker is flagged as Passing quality ( ). However, since one or more markers within this allelic ladder sample do not meet all quality requirements, the allelic ladder is classified as Low Quality ( ) and it will not be used to create bin offsets.

12. Click (No Table) in the Samples plot toolbar to hide the Genotypes table and QVD pane, and view only the dye panes.

Note: This toolbar selection does not alter the selected plot settings.



13. Select **4** from the Panes drop-down list to display all dyes in the Samples plot.

    While this particular ladder would not be usable (since several expected peaks are missing), if another ladder had broad peaks or a spike but was still genotyped accurately, you may override CGQ for the ladder, which will then automatically apply the bin offsets from that ladder. This procedure is recommended only if you do not have another passing ladder.

14. Select **File ▸ Close Plot Window** to return to the Project window.

Step 2: Examine Control Quality

Overview

In this section, you will use the features of the Analysis Summary tab and the Samples plot to manually verify the quality of the control samples analyzed in the Getting Started project in Chapter 3.

Viewing Control Quality Status

Select the **Analysis Summary** tab in the Project window.

In Chapter 3, the Control Quality area of the Analysis Summary tab indicates that of the three control samples analyzed, two were of Passing quality ( ) and one was of Low Quality ( ).

If a control has met all thresholds, the expected profile was obtained with no other anomalies detected. Therefore, you may not be required to visually inspect the control samples. However, the next two sections demonstrate how to manually review the control samples.

Examining a Passing Control

1. In the Control Quality area of the Analysis Summary tab, click the link for the Custom Control sample.

Control Quality per project (based on sample PQVs: SOS, SSPK, MIX, OMR, SQ, CGQ)

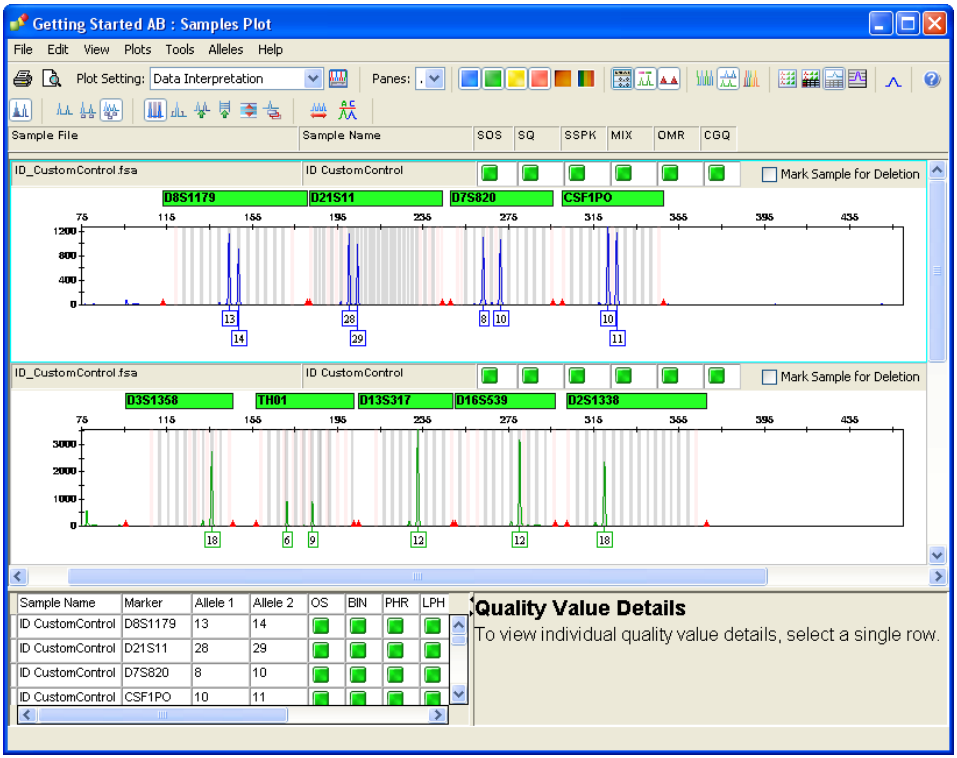
Control Type	Total # of Samples	All thresholds met	One or more thresholds not met
Positive Control	1	1	0
Custom Control	1	1	0
Negative Control	1		1
Total	3	2	1

The filtered Samples table displays the selected sample. Note that all sample-level PQVs for this custom control sample are .

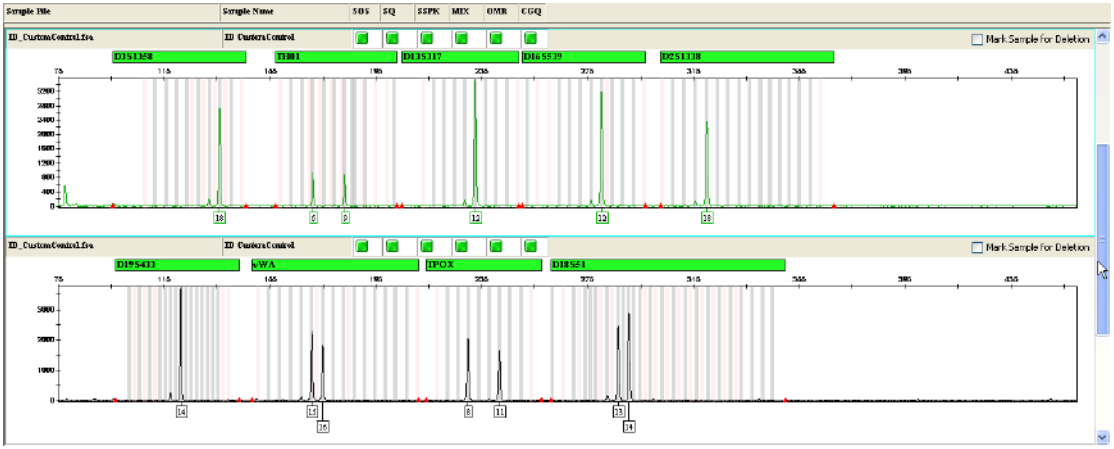
Samples (filtered) | Analysis Summary | Genotypes

	Status	Sample Name	Sample Type	Analysis Method	Panel	Size Standard	Custom Control	ARNM	SOS	SQ	SSPK	MIX	OMR	CGQ
3		ID CustomControl	Positive Control	Getting Started_your initials	Identifiler_v1X	CE_G5_HID_GS500	CUSTOM CONTROL_YOUR INITIALS							

2. Select the **ID CustomControl** sample in the filtered Samples table, then click (Display Plots). The plot for the selected custom control sample opens in the Samples plot.



3. To investigate the quality of this control sample, click the vertical scroll bar at the right of the Samples plot to scroll through all panes. Note that all of the marker headers are colored green, which indicates that this custom control sample has met all marker-level PQV thresholds.



Note: For traditional manual review, if you are required to visually inspect the control samples (not rely solely on PQVs), the color-coded marker headers may help reduce the amount of time spent on this task. If all markers are green, you know that this control produced the expected profile and no other anomalies were detected. If a marker header is yellow or red, you know those are the markers where anomalies were detected.

4. Click the green **D8S1179** marker header in the blue dye pane to display the quality assessment details for this marker. Based on the calculated GQ value shown in the QVD pane, this marker is flagged as Passing quality ( ).

Passing CC PQV

Sample Name	Marker	Allele 1	Allele 2	OS	BIN	PHR	LPH	MPH	SPK	AN	BD	CC	GQ
ID CustomControl	D8S1179	13	14										
ID CustomControl	D21S11	28	29										
ID CustomControl	D7S820	8	10										

Quality Value Details

GQ = 1.0

All quality value thresholds have been met for this marker.

Note the CC PQV, indicating that this marker contains the expected genotype for that marker.

Note: The profile for each sample in the project designated as a custom control is compared against the custom positive control profile stored in the Profile Manager. You added the CUSTOM CONTROL profile ID to the Profile Manager in Chapter 2.

5. Select **File ▸ Close Plot Window** to return to the Project window.

Examining a Low-Quality Control

1. Select the **Analysis Summary** tab in the Project window.
2. In the Control Quality area of the Analysis Summary tab, click the link for the Negative Control sample.

Control Quality per project (based on sample PQVs: SOS, SSPK, MIX, OMR, SQ, CGQ)

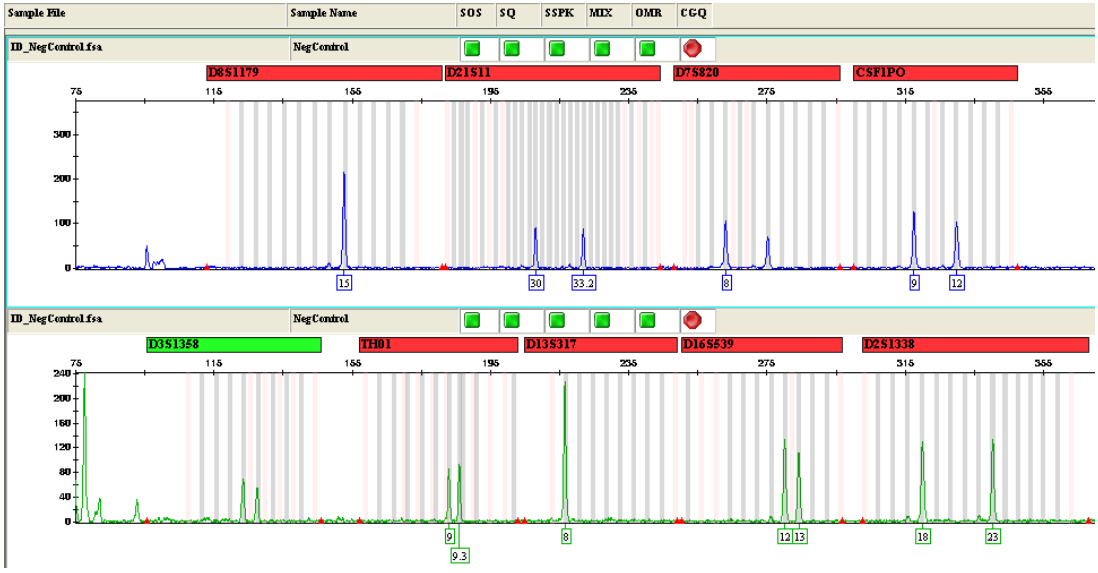
Control Type	Total # of Samples	All thresholds met	One or more thresholds not met
Positive Control	1	1	0
Custom Control	1	1	0
Negative Control	1	0	1
Total	3	2	

The filtered Samples table displays the selected sample. Note that this negative control sample shows a CGQ PQV.

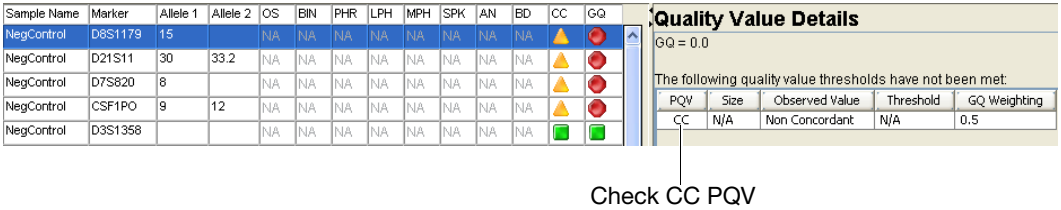
Samples (filtered) | Analysis Summary | Genotypes

	Status	Sample Name	Sample Type	Analysis Method	Panel	Size Standard	Custom Control	ARNM	SOS	SQ	SSPK	MIX	OMR	CGQ
4		NegControl	Negative Control	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	None							

3. To investigate the anomalies detected in this sample, select the **NegControl** sample in the filtered Samples table, then click (Display Plots). The plot for the selected negative control sample opens in the Samples plot. Note that several of the marker headers are colored red and there are peaks detected in the sample.



4. Click the red **D8S1179** marker header in the blue dye pane to display the quality assessment details for this marker. Based on the calculated GQ value shown in the QVD pane, this marker is flagged as Low Quality ( ).



Check CC PQV

Note the CC PQV for this marker, indicating that this marker does not contain the expected result. Since peaks are detected in several markers of this negative control, this negative control is flagged as low quality.

Note: You will use the Profile Comparison tool in Chapter 5 of this guide to help determine the possible contributor to this negative control profile.

5. Select **File ▸ Close Plot Window** to return to the Project window.

Step 3: Examine Sample Quality

Overview

In this section, you will use the features of the Analysis Summary tab and the Samples plot to manually verify the quality of the unknown samples analyzed with the Getting Started project in Chapter 3.

Viewing Sample Quality Status

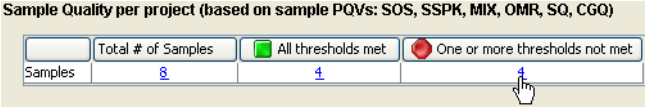
Select the **Analysis Summary** tab in the Project window.

In Chapter 3, the Sample Quality area of the Analysis Summary tab indicates that of the eight samples analyzed, four met all quality thresholds ( ) and four did not ( ).

For the purposes of this guide, you will examine only those samples that did not meet one or more thresholds. Examination of passing samples would follow the same workflow.

Examining the Low-Quality Samples

1. In the Sample Quality area of the Analysis Summary tab, click the link for the Samples.



2. The filtered Samples table displays the selected samples. Note the and sample-level PQVs for these low-quality samples.

Samples (filtered) | Analysis Summary | Genotypes

	Status	Sample Name	Sample Type	Analysis Method	Panel	Size Standard	Custom Control	ARNM	SOS	SQ	SSPK	MIX	OMR	CGQ
10		Sample 05	Sample	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	None							
11		Sample 06	Sample	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	None							
12		Sample 07	Sample	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	None							
13		Sample 08	Sample	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	None							

From the sample-level PQVs shown, you can conclude that:

- **Sample 05** contains a spike (SSPK) within a marker size range or another minor anomaly within the marker size range was detected in addition to spike between two markers (CGQ)
- **Sample 06** is a potential mixture (MIX)
- **Sample 07** has a SQ, which indicates there is a resolution issue or a problem with the size standard peak detection
- **Sample 08** contains an unexpected peak in between two markers (OMR) and other anomalies within one or more marker size ranges (CGQ)

Follow the procedures outlined in "Step 4: Review the Low-Quality Sample Results" on page 92 to individually review the quality of these unknown samples.

Step 4: Review the Low-Quality Sample Results

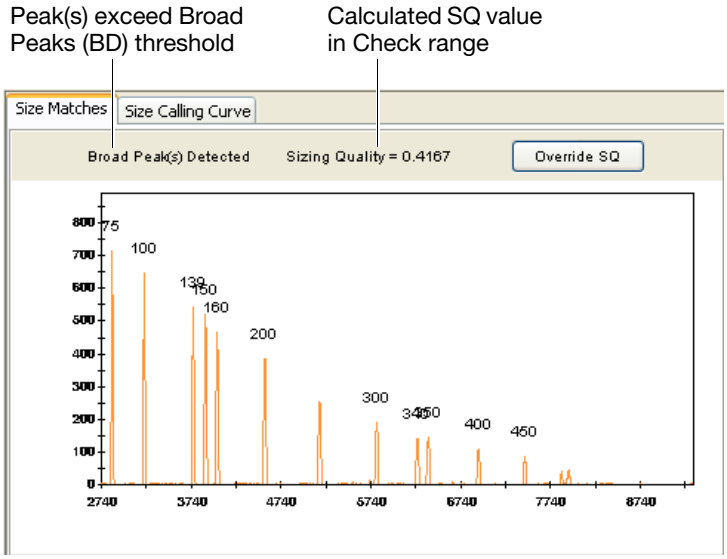
Examining the ⚠ SQ Sample Results

A ⚠ SQ PQV indicates that the sizing quality is below the passing range specified in the analysis method. This example illustrates:

- How to view SQ using the Size Match Editor
- The SQ PQV flag

To examine a ⚠ SQ PQV:

1. Select **Sample 07** in the Samples table, then click (Size Match Editor) to view the peak assignments for the size standard peaks in this sample.



Note the following in the Size Match Editor:

- All size standard peaks are present and labeled correctly, as compared to the fragment sizes stored in the CE_G5_HID_GS500 size standard definition file set for this sample
- The calculated SQ value, 0.4167 for this sample, is within the Check ( ) range set in the analysis method (default value = 0.74 – 0.26)
- A "Broad Peak(s) Detected" message indicates there are peaks present in this sample that exceed the Broad Peak (BD) Max Peak Width threshold set in the analysis method

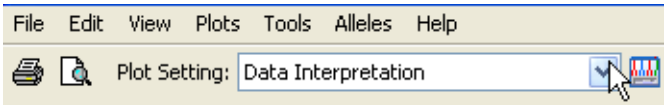
In this example, the SQ is due to the broad peaks.

Note: For more information on the calculation of SQ, see the *GeneMapper® ID-X Software Reference Guide*.

2. Click **OK** to close the Size Match Editor.

Displaying the Low-Quality Sample Plots

1. Shift-click to select all samples in the filtered Samples table, then click (Display Plots).
2. Verify that **Data Interpretation** is selected from the Plot Setting drop-down list.



3. The plots for all of the selected samples are shown in the Samples plot.

Sample Name	Marker	Allele 1	Allele 2	OS	BIN	PHR	LPH	MPH	SPK	AN	BD	CC	GQ
Sample 05	D8S1179	15	16									NA	
Sample 05	D21S11	31.2				NA						NA	
Sample 05	D7S820	11	12									NA	
Sample 05	CSF1PO	12				NA						NA	
Sample 05	D3S1358	16				NA						NA	

The first two panes display the blue dye and green dye for the first sample selected in the Samples table (in this example, Sample 05).

Note: The selection order in the Samples table determines the display order in the Samples plot.

Examining the ⚠ SSPK Sample Results

A ⚠ SSPK (Sample Spike) PQV indicates that one or more spike peaks have been detected within a marker range or between two markers. This example illustrates:

- Automatic labeling of spikes
- The Marker Spike (SPK) and Peak Height Ratio (PHR) PQV flags
- How to confirm a spike in raw data

- How to override GQ
- How to override CGQ

To investigate a marker with a yellow or red marker header bar:

1. Click the yellow **D2S1338** marker header in the green dye pane of Sample 05 to display the quality assessment details for this marker.

Check PHR and SPK PQVs

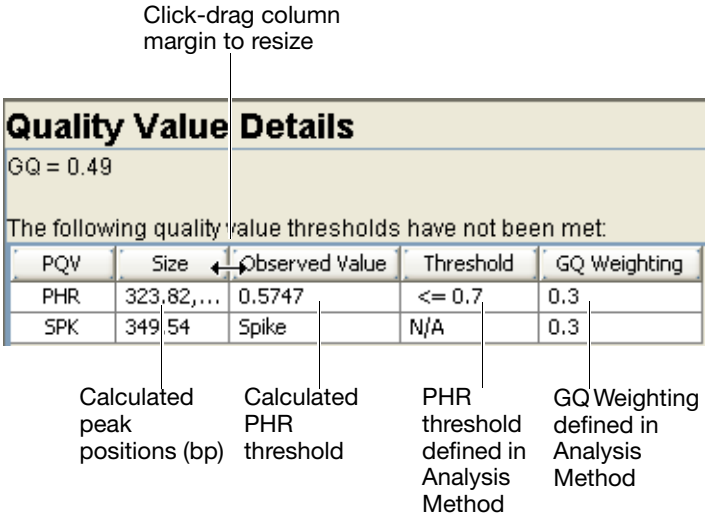
Note the following:

- There is a peak labeled as “Spike” present in this marker and the marker-level SPK PQV is triggered, indicating that one or more spikes are detected within the marker size range (in this case, one spike was detected)

Note: This peak is automatically labeled as a Spike artifact and displayed with a pink label border by the GeneMapper® *ID-X* Software. Peaks labeled as artifacts are not considered true alleles by the software and are not listed in the Genotypes table.

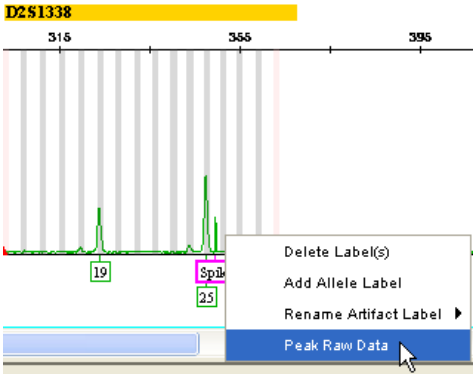
- The marker-level ⚠ PHR PQV is also triggered for this marker, indicating that the peak height ratio calculated between the lowest and highest allele peaks within the marker is less than the Min Peak Height Ratio threshold defined in the analysis method (in this example, 0.7)

2. Click-drag the Size column margin in the QVD pane to the right to expand its contents and view the peak sizes contributing to the ⚠ PHR and SPK PQVs for this marker.



Note: The SPK and PHR PQV status are used to determine the GQ PQV. Based on the calculated GQ value shown in the QVD pane, this marker is flagged as Check (⚠).

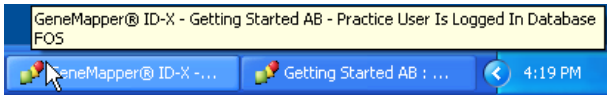
3. In the green dye pane, left-click the **Spike** artifact label to select the peak, then right-click the label to open a drop-down menu containing peak edit actions.



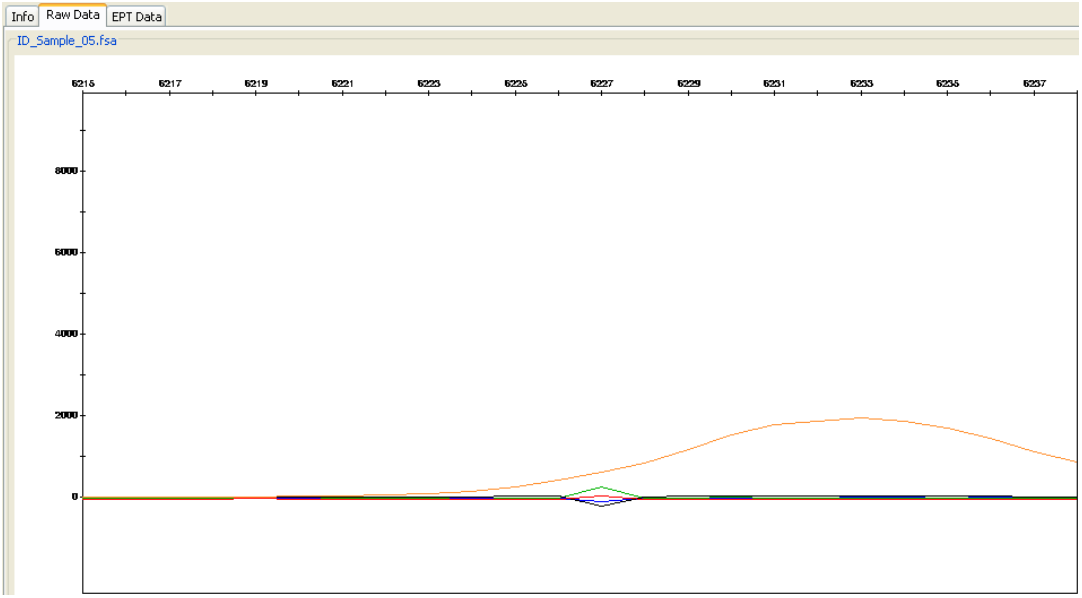
IMPORTANT! You must left-click the label before you right-click the label.

4. To confirm the spike peak, select **Peak Raw Data** from the drop-down menu to open the raw data plot for this peak in the Project window.

5. To view the raw data plot, click the **GeneMapper® ID-X - Getting Started <your initials>** taskbar button.



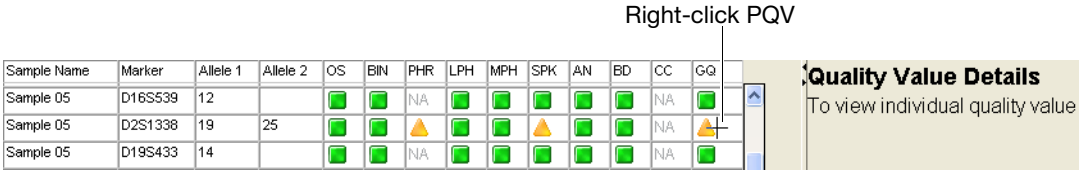
6. Review the raw data and peak morphology to confirm that the selected peak is a spike.



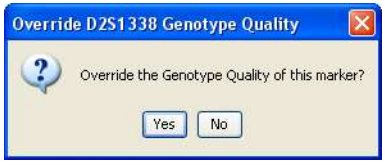
Note: Once you have validated the spike detection PQVs using your own data, you may not be required to view the raw data to confirm a spike peak.

7. To return to the Samples plot, click the **Getting Started <your initials> : Samples Plot** taskbar button.

8. Right-click the ⚠ GQ PQV in the D2S1338 marker row highlighted in the Genotypes table.



9. Click **Yes** in the dialog box to override the genotype quality for this marker.

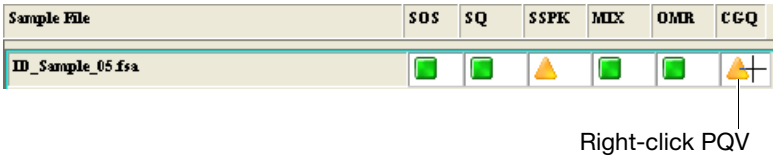


Note that after overriding:

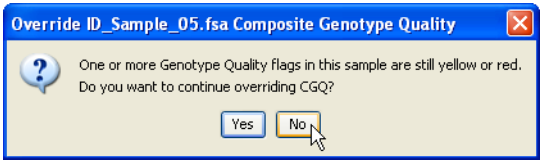
- The GQ PQV changes to
- All other PQVs for the marker turn gray and maintain their original shape ( ) to indicate that the marker has been overridden
- The marker header turns green D2S1338

Note: Overriding GQ allows you to manually accept the genotype for a particular marker and also provides evidence that the marker was visually inspected by the analyst.

10. To accept the entire sample profile for Sample 05, you can override its CGQ PQV. To do so, first right-click the CGQ PQV for Sample 05 in the genotypes header of the Samples plot.



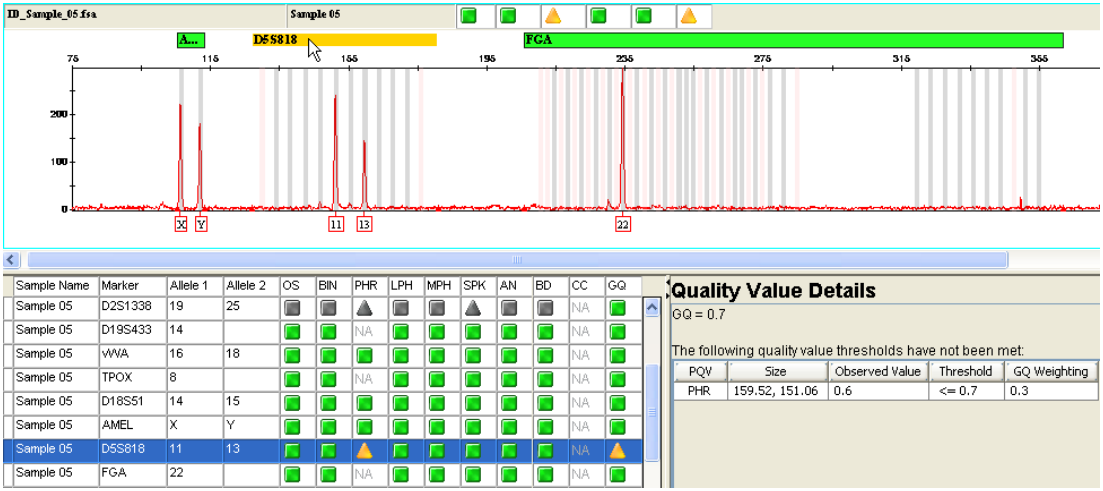
The next dialog box displays a message indicating that there are still or marker-level GQ PQVs present in this sample. Unless you have viewed the rest of the markers and confirmed the genotypes, you should not continue with the CGQ override.



Click **No** to return to the Samples plot and confirm the genotypes for the remaining markers.

11. Click the vertical scroll bar at the right of the Samples plot to scroll through the two remaining dye panes (yellow and red) for Sample 05. Note that there is a yellow D5S818 marker header in the red dye pane. It was this marker that triggered the Override CGQ warning message seen in step 10 on page 99.

12. Click the yellow **D5S818** marker header to display the quality assessment details for this marker.



Note the marker-level ⚠ PHR PQV in the Genotypes table for this marker. Based on the calculated PHR value shown in the QVD pane, this marker is flagged as Check (⚠).

13. Right-click the ⚠ GQ PQV in the highlighted row of the Genotypes table, then click **Yes** in the dialog box to override the genotype quality for this marker.

14. Now that all GQ PQVs are for Sample 05, click **Yes** in the next dialog box to override the CGQ for this sample.

Note that after overriding, the CGQ PQV changes to (Manually Overridden).

Note: Overriding CGQ allows you to manually accept the entire profile for a particular sample and also provides evidence that the sample was visually inspected by the analyst.

You have now completed reviewing Sample 05. Another use for CGQ override will be demonstrated in Chapter 6 for review purposes.

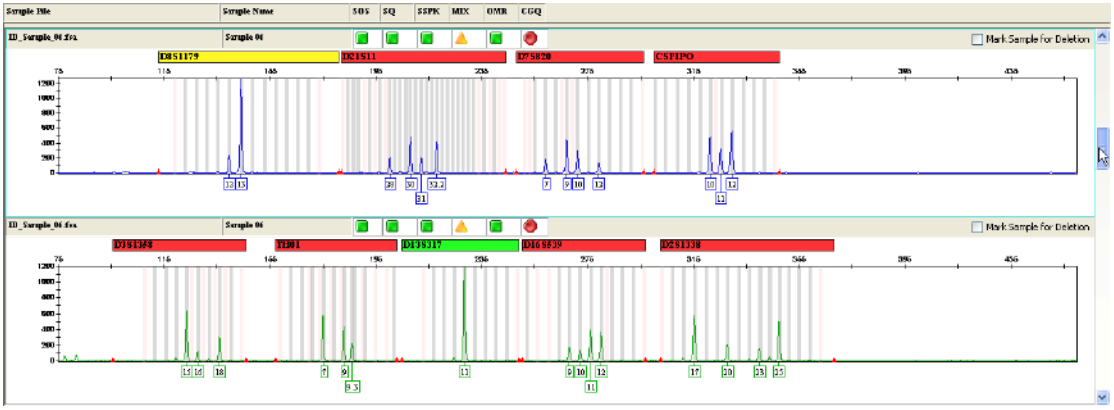
Examining the MIX Sample Results

A Mixed Source (MIX) PQV indicates a potential mixed-source sample. This example illustrates:

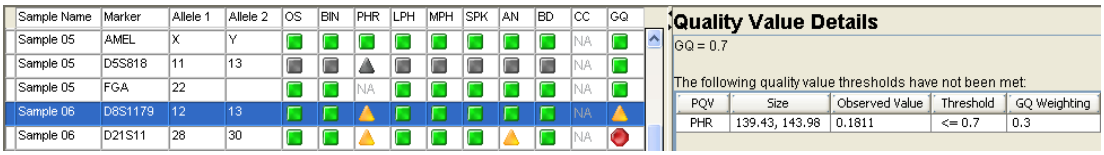
- How to select peaks and view their calculated peak height ratios
- The marker-level PQVs triggered in mixed samples
- The PHR and Allele Number (AN) PQV flags

To investigate the cause of the yellow MIX PQV flag:

1. Click the vertical scroll bar at the right of the Samples plot until you can see the first two dye panes (blue and green) for Sample 06.



2. Click the yellow **D8S1179** marker header in the blue dye pane to display the quality assessment details for this marker.



Sample Name	Marker	Allele 1	Allele 2	OS	BIN	PHR	LPH	MPH	SPK	AN	BD	CC	GQ
Sample 05	AMEL	X	Y									NA	
Sample 05	D5S818	11	13									NA	
Sample 05	FGA	22				NA						NA	
Sample 06	D8S1179	12	13									NA	
Sample 06	D21S11	28	30									NA	

Quality Value Details

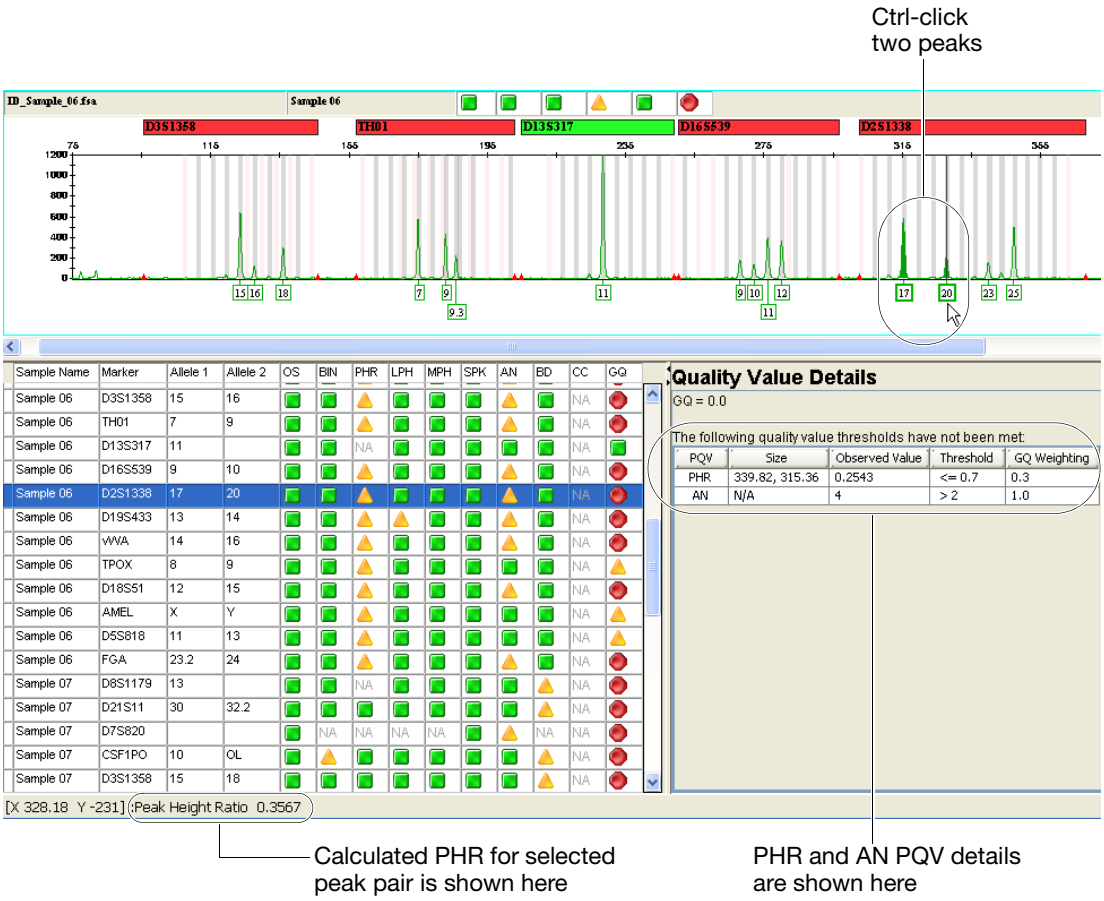
GQ = 0.7

The following quality value thresholds have not been met:

PQV	Size	Observed Value	Threshold	GQ Weighting
PHR	139.43, 143.98	0.1811	<= 0.7	0.3

Note the marker-level ⚠ PHR PQV in the Genotypes table for this marker. Based on the calculated PHR value shown in the QVD pane, this marker is flagged as Check (⚠).

3. Ctrl-click the **17** and **20** allele peaks in the green dye pane for marker D2S1338 to select them both. The software automatically calculates a peak height ratio (PHR) value for the selected peaks and displays the results in the status bar at the bottom of the Samples plot.



Note the following:

- The calculated PHR is less than the minimum PHR threshold of 0.7 specified in the analysis method
- The ⚠ AN PQV in the Genotypes table for this marker, indicating the number of alleles detected is greater than the Max Expected Alleles defined in the analysis method

Note: The AN and PHR PQV status is used to determine the GQ PQV. Based on the observed AN and PHR values shown in the QVD pane, this marker is flagged as Low Quality ( ).

4. Repeat step 3 on page 103 for any of the other peak pairs in this sample.

   You have now completed reviewing Sample 06. This sample was expected to be from a single source. You will use the Profile Comparison tool in Chapter 5 to help determine the potential contributor(s) to this mixture.

Note: For more information on the conditions that must be met for a sample to be considered a potential mixture, see the *GeneMapper® ID-X Software Reference Guide*.

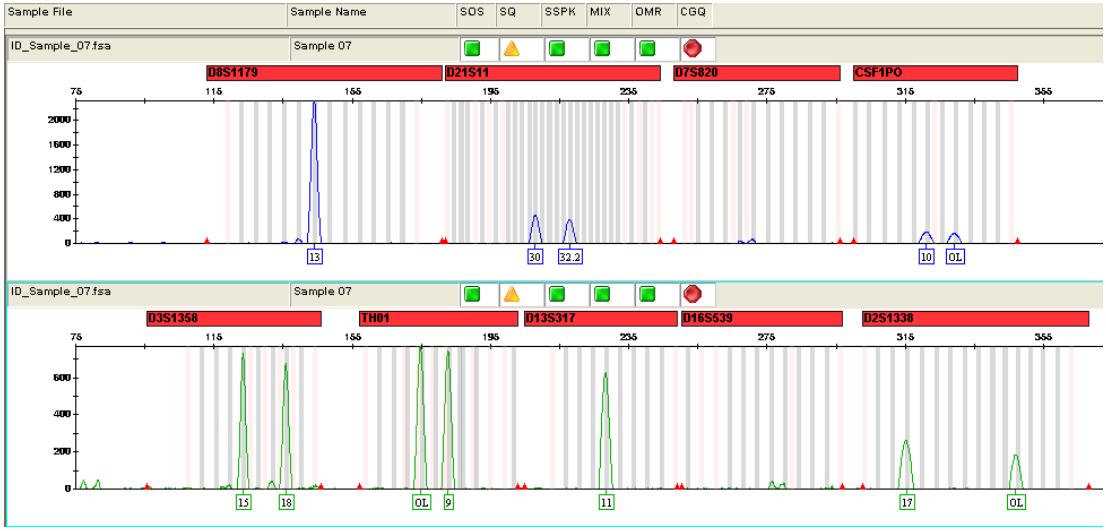
Examining the ⚠ SQ Sample Results

This example illustrates:

- How to view PQV trigger labels
- The Broad Peak (BD) PQV flag
- How to delete allele labels for a sample
- How to add a reason for change to the audit trail
- How to mark a sample for deletion from the project

To investigate this low-quality sample:

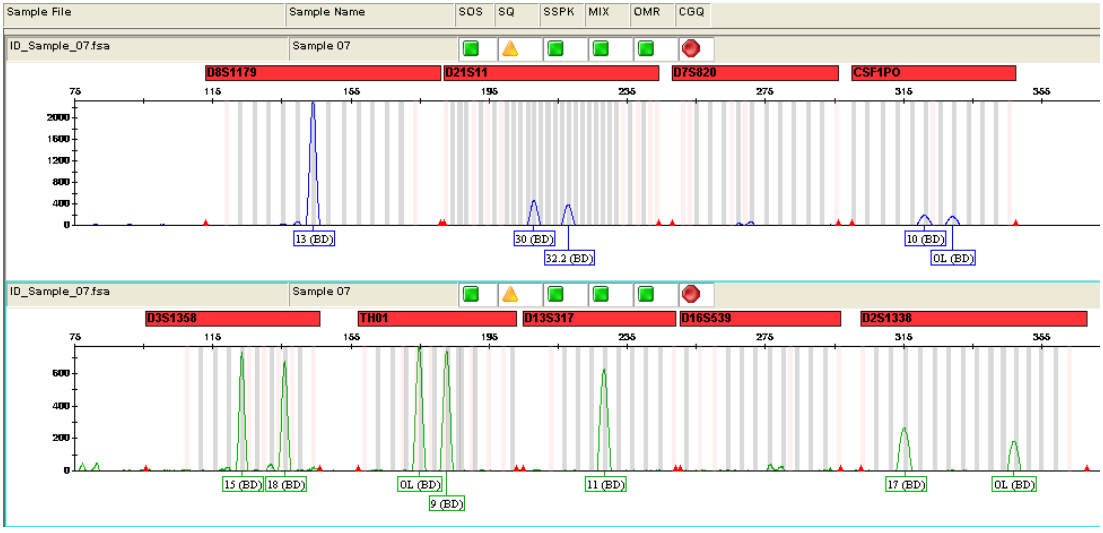
1. Click the vertical scroll bar at the right of the Samples plot until you can see the first two dye panes (blue and green) for Sample 07.



Note: This is the same sample you viewed using the Size Match Editor earlier in this chapter (see "Examining the SQ Sample Results" on page 92).

2. In the Samples plot toolbar, click (PQV Trigger Peak) to add PQV suffixes to peak labels for peaks that cause any of the following PQVs: LPH, MPH, BD, OS.

Note: Displaying the PQV trigger peak labels can help you quickly find the exact peaks that triggered specific PQV flags.



In this example, (BD) is appended to the label of each peak that exceeds the Max Peak Width threshold.

3. Click (PQV Trigger Peak) again to turn off the PQV trigger labels.

4. Click the red **D3S1358** marker header to display the quality assessment details for this marker. Note the marker-level ⚠ BD PQV in the Genotypes table, and the Max Peak Width threshold and observed values related to this PQV in the QVD pane.

The genotype profile for this sample is not reliable and should not be reported. Therefore, from the Samples plot, you can choose to:

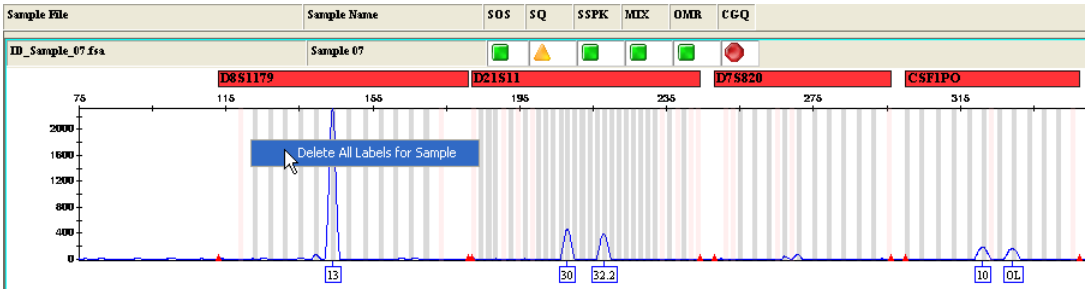
- Delete all labels for a sample

  or

- Mark the sample for deletion from the project

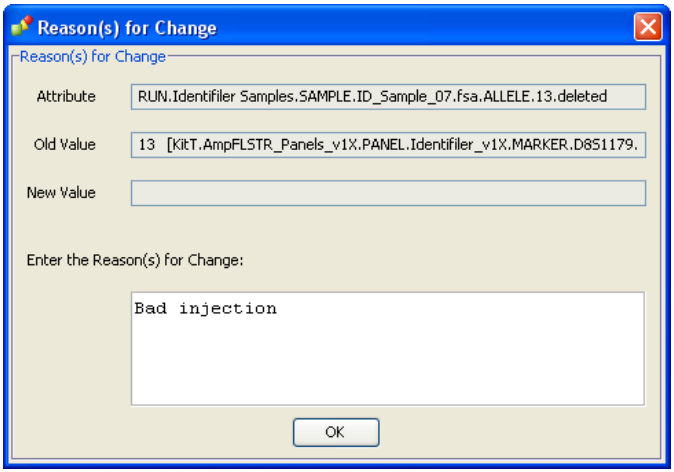
The following steps 5 through 8 demonstrate these two options.

5. To delete all labels for Sample 07, right-click in any pane for this sample, then select **Delete All Labels for Sample**.



6. Click **Yes** in the Delete All Labels dialog box.

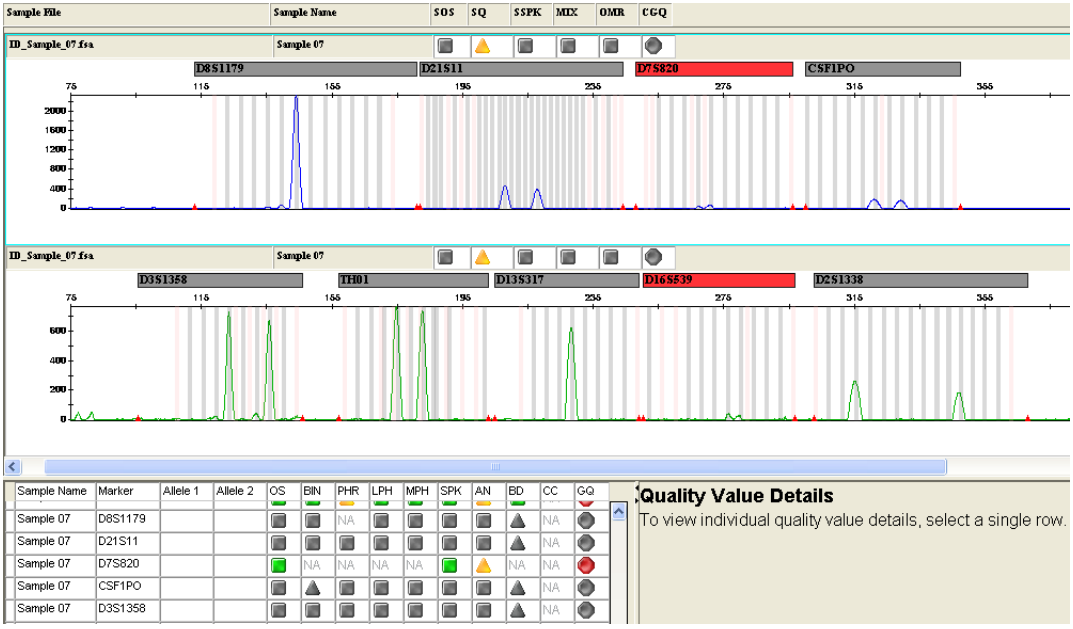
7. Enter **Bad injection** in the Reason(s) for Change dialog box (recorded in the audit trail), then click **OK**. You can also press **Alt+O** to close the dialog box.



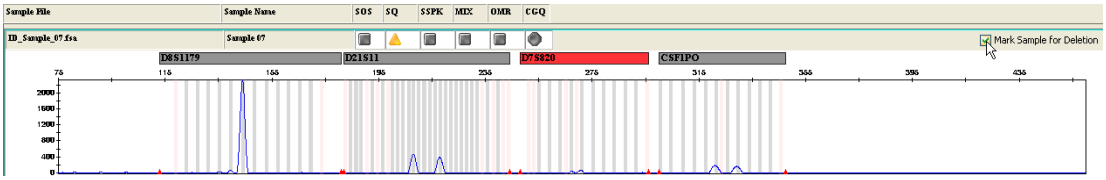
Note: By default, a reason for change audit trail entry is required for all label edits.

Note that after deleting all allele labels for Sample 07:

- All sample-level and marker-level PQVs for the sample turn gray and maintain their original shapes ( ) to indicate that the sample has been edited
- Affected marker header colors change to gray
- The sample will still be listed in the Samples and Genotypes table, but no allele values are reported



8. To delete Sample 07 from the Getting Started project, select **Mark Sample for Deletion** at the top right of any dye pane for this sample.



Note: The remaining panes associated with this sample are automatically marked for deletion by the software.

The sample will be deleted from the project when the Samples plot is closed. For now, keep the Samples plot open to continue reviewing the last low-quality sample results in this project.

You have now completed reviewing Sample 07.

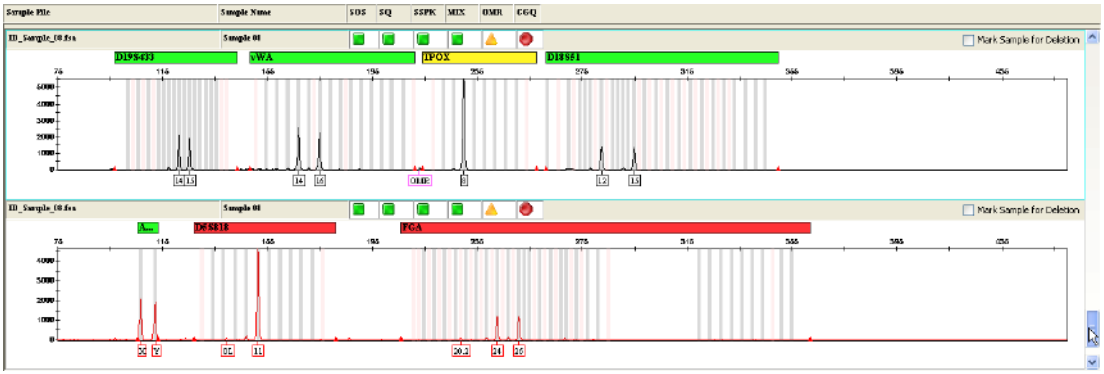
Examining the ⚠ OMR Sample Results

A ⚠ Outside Marker Range (OMR) PQV indicates that one or more peaks were detected between two marker size ranges specified in the panel for the marker. This example illustrates:

- Automatic labeling of OMR peaks
- The Out of Bin Allele (BIN), Low Peak Height (LPH), and Max Peak Height (MPH) PQV flags
- How to change an artifact label to an allele label and assign it to a selected marker
- How to save a custom artifact label for future use
- How to change an allele label to an artifact label

To investigate the source of the OMR peak and other potential anomalies:

Click the vertical scroll bar at the right of the Samples plot until you can see the last two dye panes (yellow and red) for Sample 08.



Note the following:

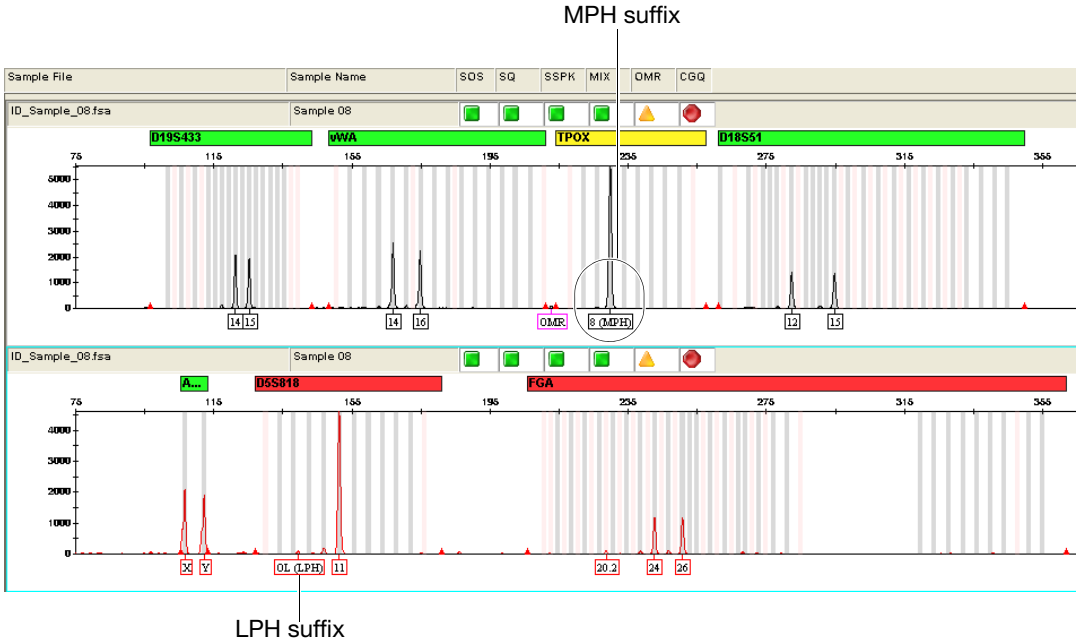
- There is a peak labeled OMR present between the vWA and TPOX markers
- The marker header for TPOX is yellow
- The marker headers for D5S818 and FGA are red
- There is a peak labeled OL present in the D5S818 marker
- There are three labeled peaks present in the FGA marker

Follow the procedures in the following sections (on pages 112 to 130) to individually review the quality of these markers.

You will first investigate the OMR peak between vWA and TPOX and the quality of the TPOX marker.

To examine TPOX marker and OMR peak:

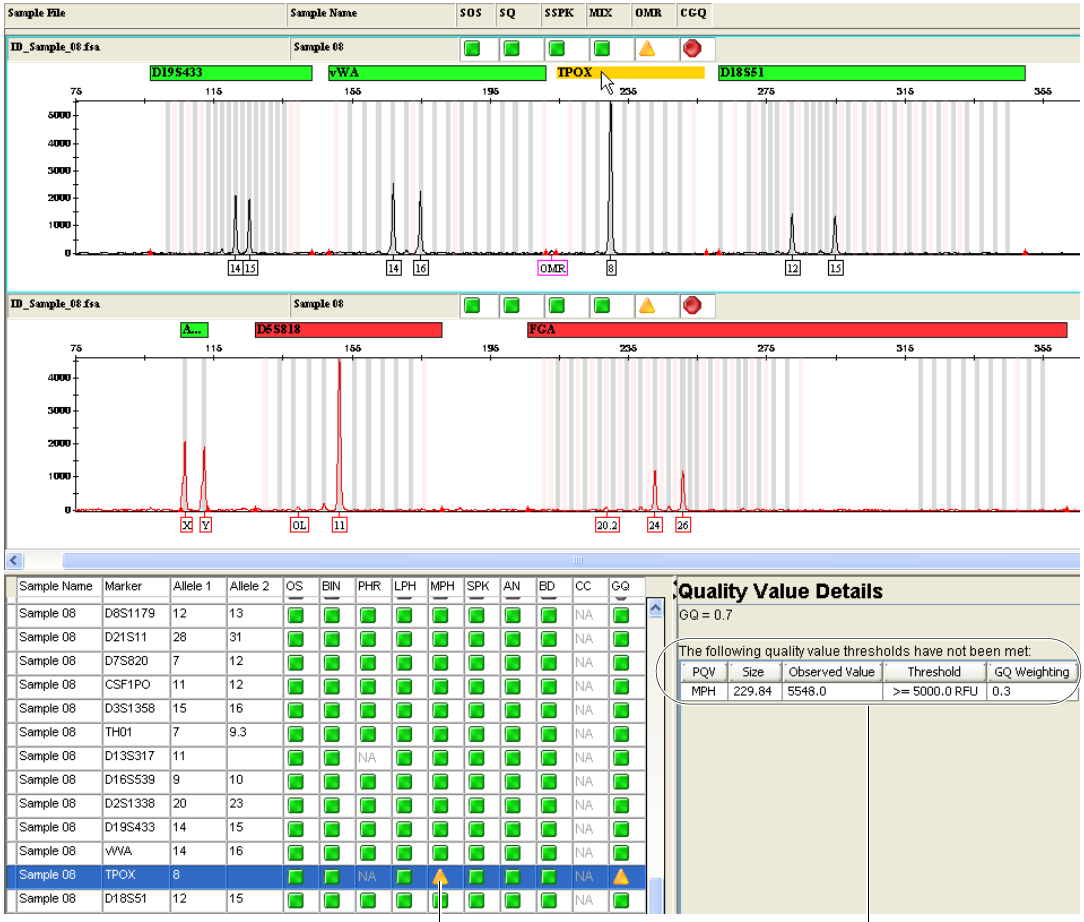
1. In the Samples plot toolbar, click (PQV Trigger Peak) to add PQV suffixes to peak labels for peaks that cause any of the following PQVs: LPH, MPH, BD, OS.



In this example, (MPH) in TPOX and (LPH) in D5S818 are appended to the label of each peak that does not meet the thresholds related to these PQVs set in the analysis method.

2. Click (PQV Trigger Peak) again to turn off the PQV trigger labels.

3. Click the yellow **TPOX** marker header to display the quality assessment details for this marker. Note the marker-level MPH PQV in the Genotypes table for this marker, and the Max Peak Height threshold (in RFU) and observed values related to this PQV in the QVD pane.



Check MPH PQV for selected marker

Observed Max Peak Height values and accepted thresholds are shown here

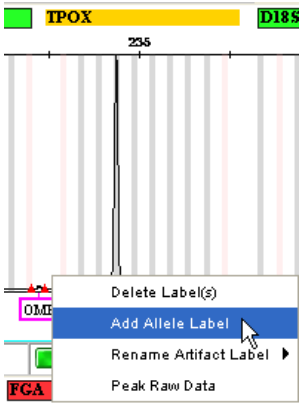
4. With the TPOX marker header still selected, left-click the **OMR** label.

Sample Name	Marker	Allele 1	Allele 2	OS	BIN	PHR	LPH	MPH	SPK	AN	BD	CC	GQ
Sample 08	D7S820	7	12									NA	
Sample 08	CSF1PO	11	12									NA	
Sample 08	D3S1358	15	16									NA	
Sample 08	TH01	7	9.3									NA	
Sample 08	D13S317	11				NA						NA	
Sample 08	D16S539	9	10									NA	
Sample 08	D2S1338	20	23									NA	
Sample 08	D19S433	14	15									NA	
Sample 08	vWA	14	16									NA	
Sample 08	TPOX	8				NA						NA	
Sample 08	D18S51	12	15									NA	

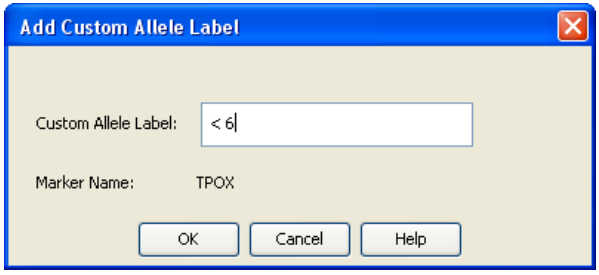
Note: This peak is automatically labeled as an OMR artifact by the GeneMapper® *ID-X* Software. Peaks labeled as artifacts are not listed in the Genotypes table. In some rare cases, an OMR artifact peak may be a true allele; you may need to verify the classification of an OMR peak if you encounter one in your own data.

For this example, you will assume that this OMR artifact peak is indeed a true allele solely to learn how to add an allele label to an artifact peak and assign it to a selected marker (although you are aware this is not a true DNA peak).

5. Right-click the **OMR** label, then select **Add Allele Label**.

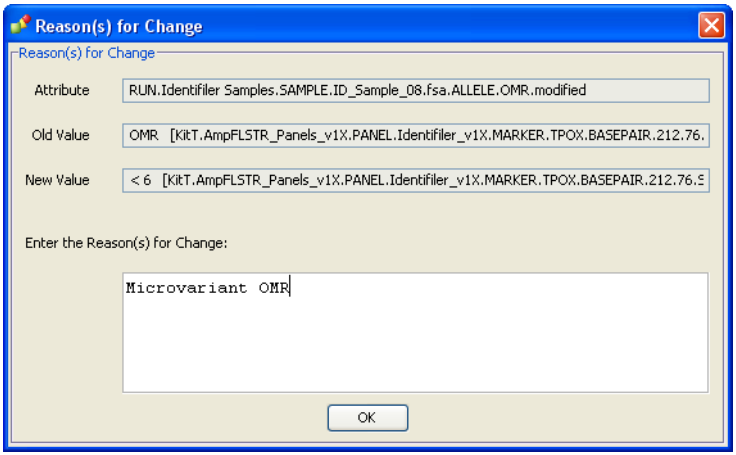


6. In the Add Custom Allele Label dialog box, enter **< 6**, then click **OK**. Note that the TPOX marker is selected for this allele since this was the marker selected before adding the allele.



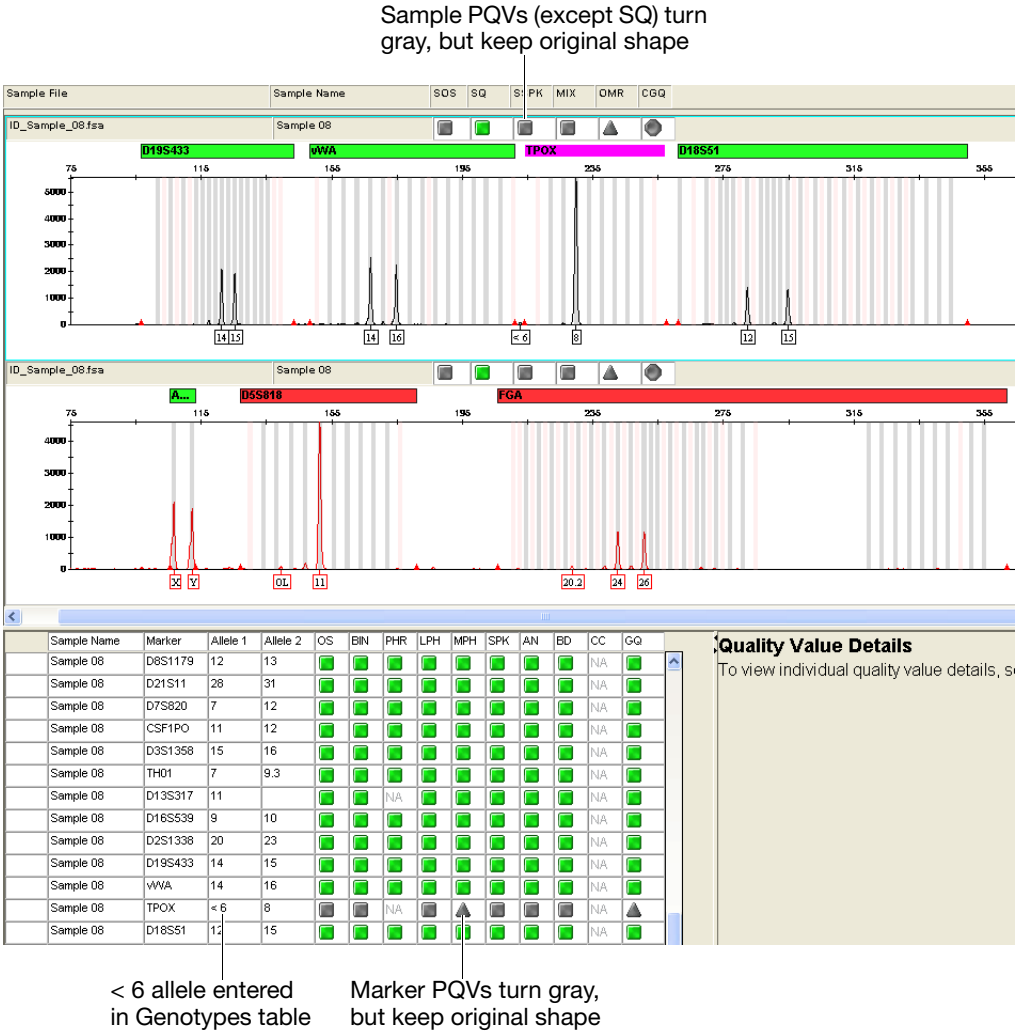
Note: This custom allele will be added to the TPOX marker and entered as an allele in the Genotypes table.

7. In the Reason(s) For Change dialog box, enter **Microvariant OMR**, then click **OK**. You can also press **Alt+O** to close the dialog box.



Note that after editing:

- All sample-level PQVs (except SQ) for the sample turn gray ( ) to indicate that the sample has been edited
- All marker-level PQVs for the marker turn gray and maintain their shape ( ), to indicate that the marker has been edited
- The allele is listed in the Genotypes table
- The allele label changes text and color from pink to black
- The marker header color changes from yellow to gray (pink if selected)



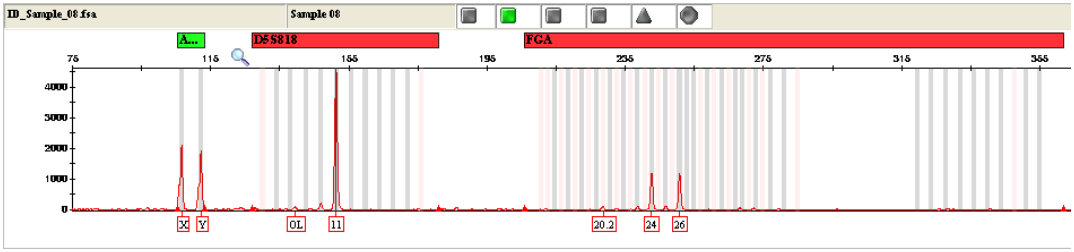
8. Select the **TPOX** marker row in the Genotypes table, then right-click its ▲ GQ PQV.

9. Click **Yes** in the dialog box to override the genotype quality and manually accept the current genotype (<6, 8) for the TPOX marker. The GQ PQV changes to ■ and the marker header turns green.

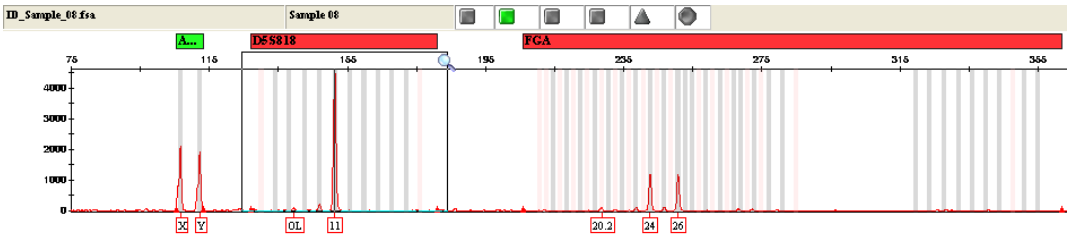
You have completed reviewing the TPOX marker. You will now investigate the OL peak present in the D5S818 marker.

To examine the cause of an extra peak:

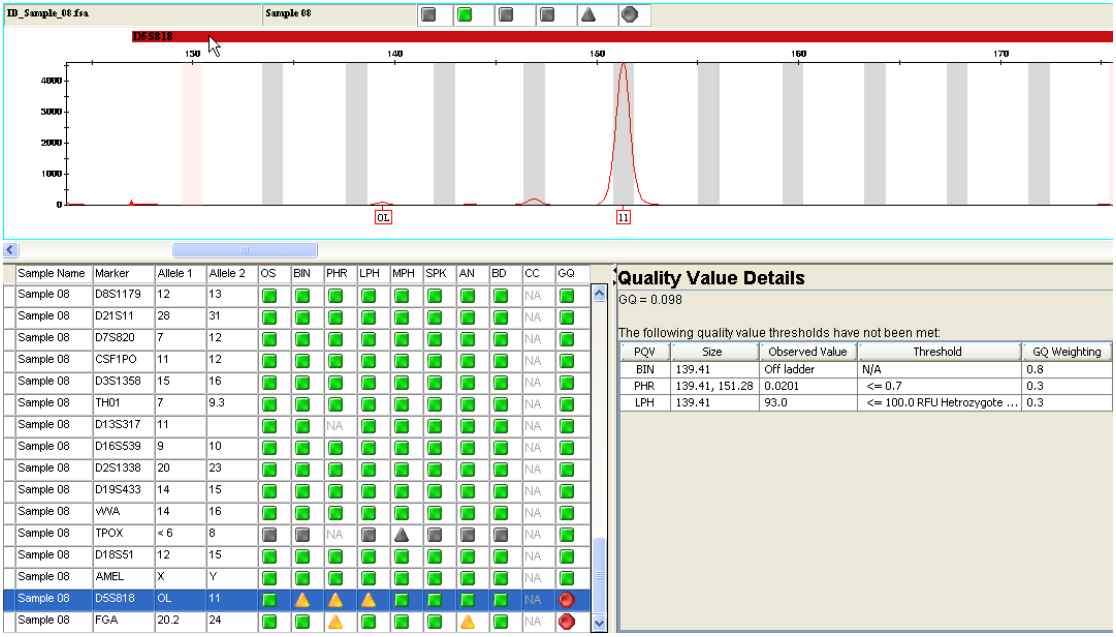
1. In the red dye pane for Sample 08, place the pointer next to the X-axis below the red D5S818 marker header until the pointer changes to a 🔍.



2. Click-drag the pointer to the right to create a box that includes the area to the left of the D5S818 marker, then release to zoom in on this marker, as shown below.



3. Click the red **D5S818** marker header to display the quality assessment details for this marker. Note the marker-level BIN PQV in the Genotypes table for this marker.

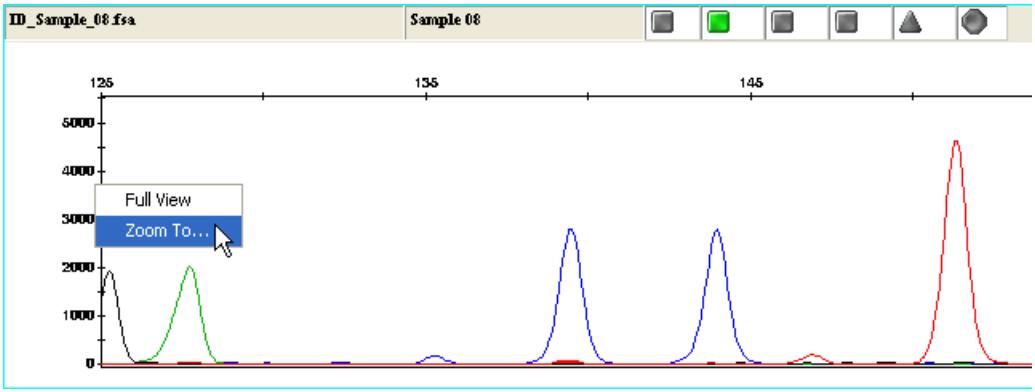


Note: A BIN PQV indicates that one or more labeled peaks are detected outside an offset bin. These peaks are automatically labeled as OL peaks by the software. For information on bin offsetting, see the *GeneMapper® ID-X Software Reference Guide*.

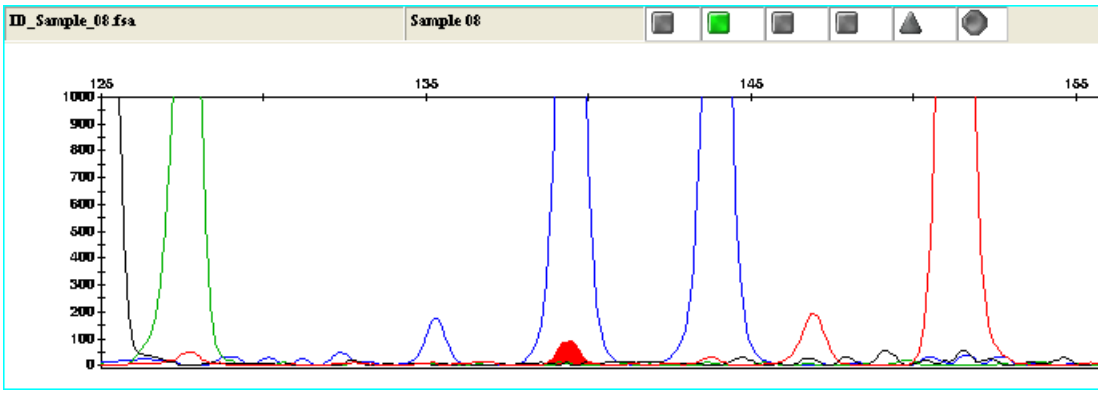
4. Click the **OL** peak label to select the OL peak.

5. Click (Combine Dyes) in the Samples plot toolbar to display all dye colors for the sample in one pane.

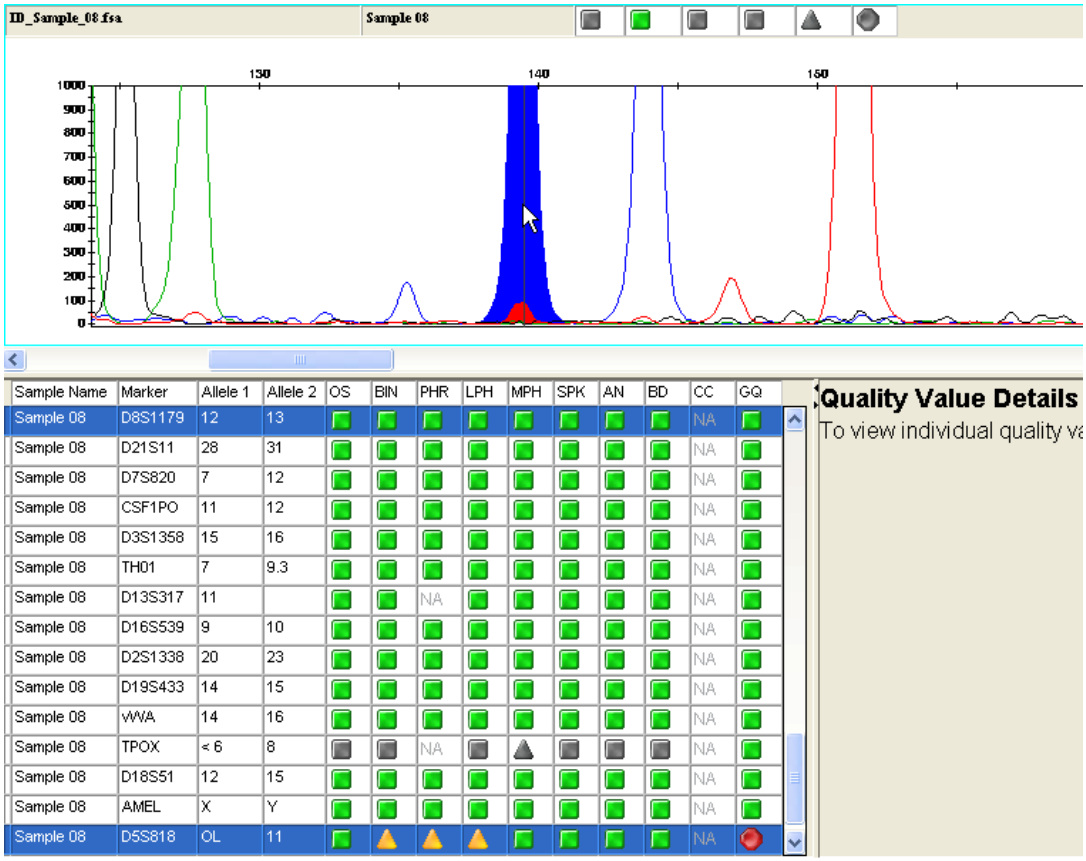
6. Place the pointer next to the Y-axis until the pointer changes to a 🔍, then right-click and select **Zoom to**.



7. Enter **1000** in the Y-Axis Zooming dialog box, then click **OK** to zoom in on the OL peak. Note that the red OL peak is located under a larger blue dye peak.

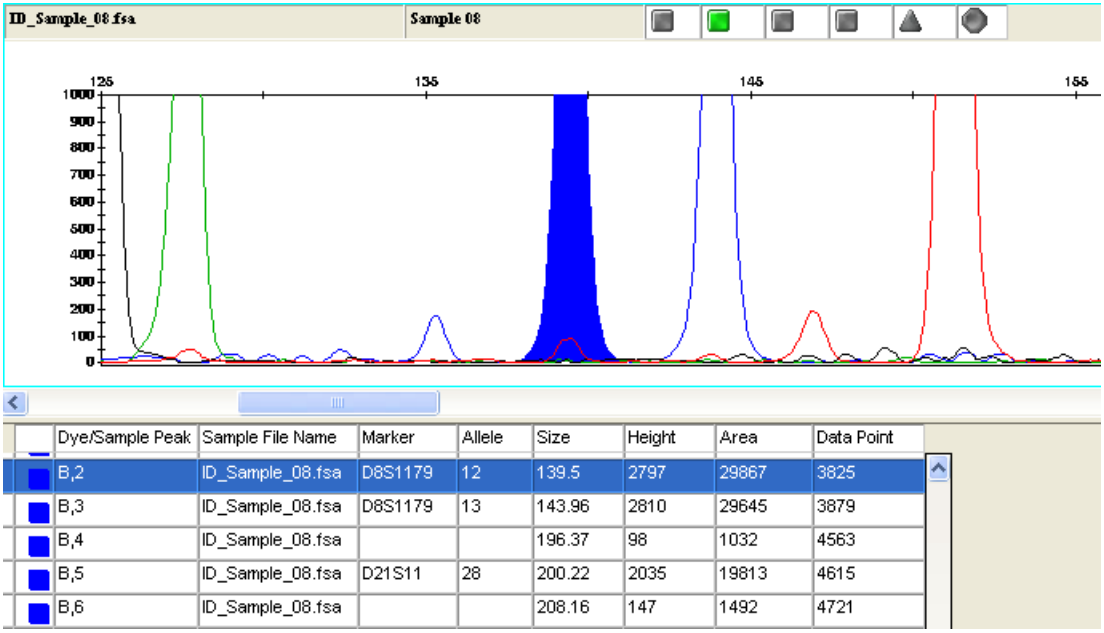


8. Ctrl-click the overlaying blue dye peak to select it (the red OL peak should still be highlighted).



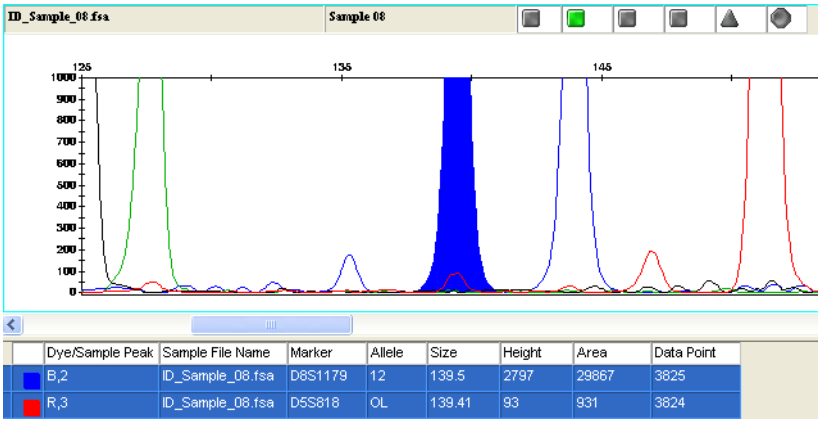
Sample Name	Marker	Allele 1	Allele 2	OS	BIN	PHR	LPH	MPH	SPK	AN	BD	CC	GQ
Sample 08	D8S1179	12	13									NA	
Sample 08	D21S11	28	31									NA	
Sample 08	D7S820	7	12									NA	
Sample 08	CSF1PO	11	12									NA	
Sample 08	D3S1358	15	16									NA	
Sample 08	TH01	7	9.3									NA	
Sample 08	D13S317	11				NA						NA	
Sample 08	D16S539	9	10									NA	
Sample 08	D2S1338	20	23									NA	
Sample 08	D19S433	14	15									NA	
Sample 08	vWA	14	16									NA	
Sample 08	TPOX	< 6	8			NA						NA	
Sample 08	D18S51	12	15									NA	
Sample 08	AMEL	X	Y									NA	
Sample 08	D5S818	OL	11									NA	

9. Click (Sizing Table) in the Samples plot toolbar to display the Sizing table, which contains the following peak sizing information for all detected peaks in the sample: size (in base pairs), peak height, peak area, and data point.



Note: The columns displayed in the Sizing table are determined by the plot setting selected in the Samples plot.

10. With the blue dye and red OL peaks still selected, press **Ctrl+G** to display only the selected peaks in the Sizing table.

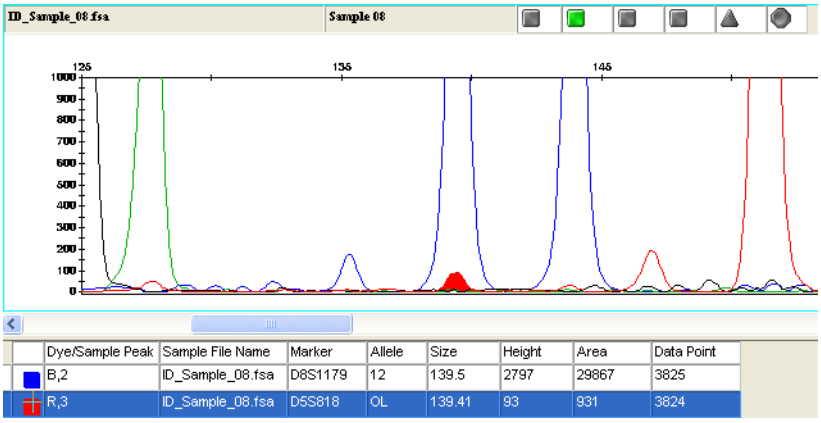


The sizing data indicates that the red OL peak could be caused by spectral pull-up.

You will change the software-generated OL peak label to a custom artifact label in the next steps.

To change the OL peak label to a custom artifact label:

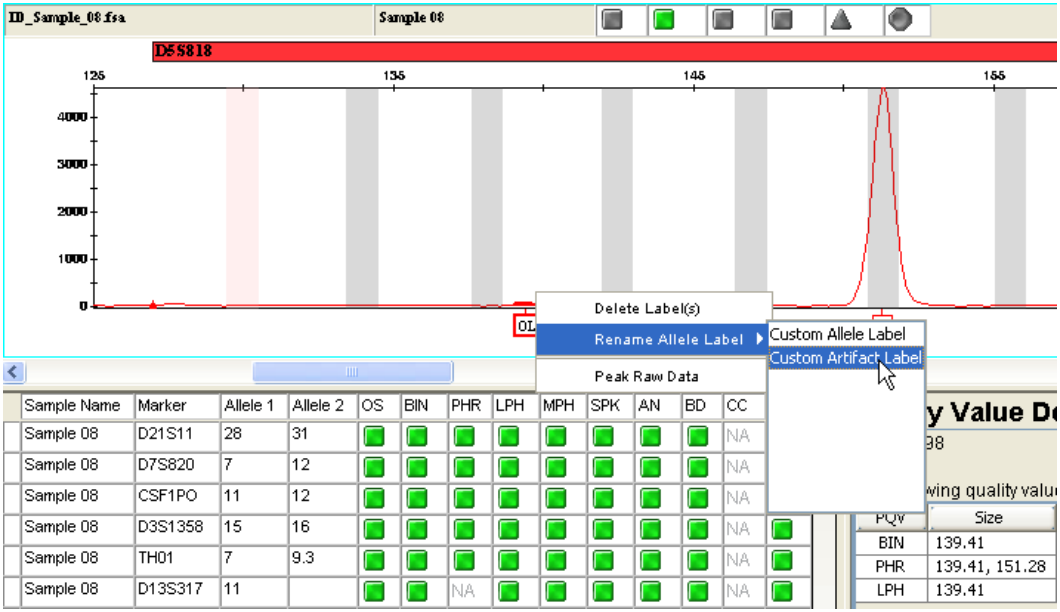
1. Select the **D5S818** row in the Sizing table to deselect the overlaying blue dye peak (the red OL peak should still be highlighted).



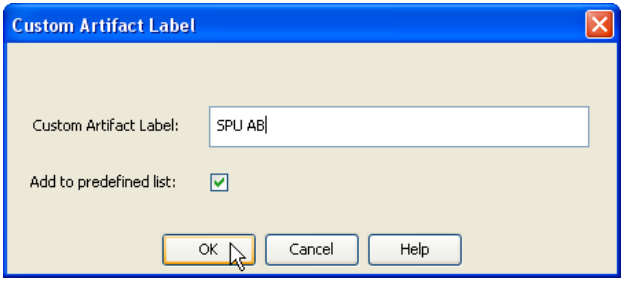
2. Click (Separate Dyes) in the Samples plot toolbar to display each dye color for the sample in separate panes.

3. Click (Genotypes Table) in the Samples plot toolbar to replace the Sizing table with the Genotypes table and QVD pane.

4. Left-click the **OL** peak label in the red dye pane, then right-click the label and select **Rename Allele Label ▸ Custom Artifact Label**.



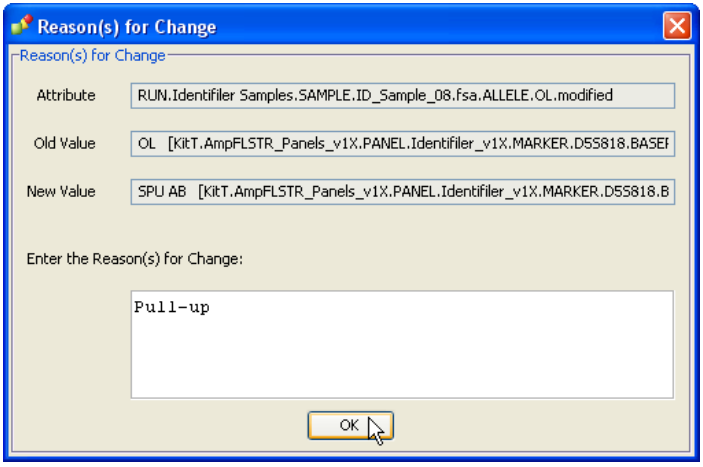
5. Complete the Custom Artifact Label dialog box:
   a. Enter **SPU <your initials>** (for example, **SPU AB**).
   b. Select **Add to predefined list**.



   c. Click **OK**.

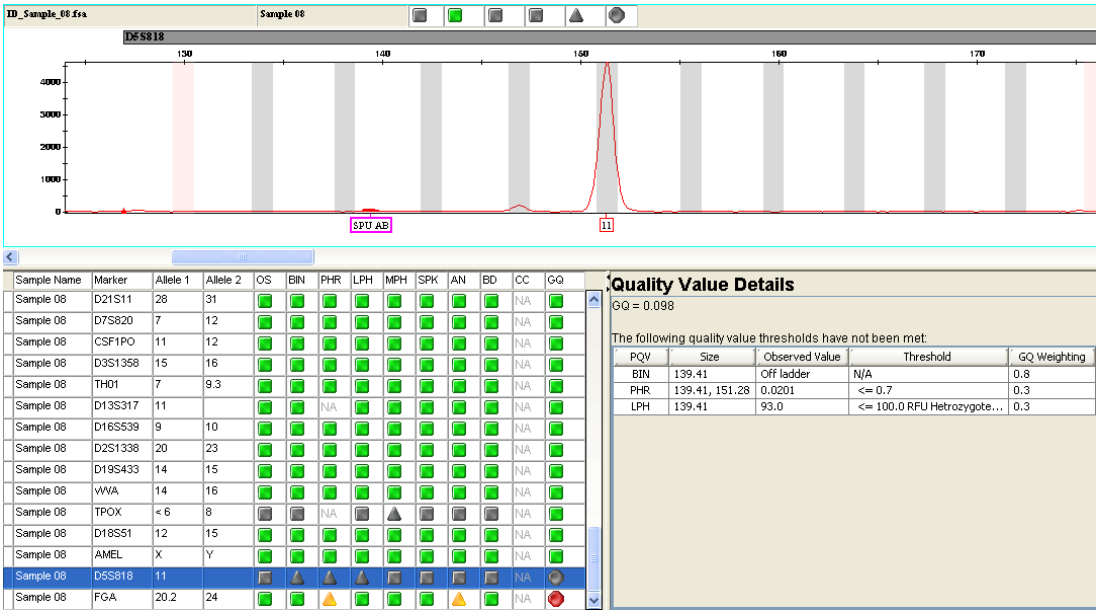
Note: This option allows you to select this custom artifact label from a list of labels in the future.

6. In the Reason(s) For Change dialog box, enter **Pull-up**, then click **OK**. You can also press **Alt+O** to close the dialog box.



Note that after editing:

- The SPU artifact label is displayed with a pink border
- The SPU artifact peak is not added to the Genotypes table since an artifact is not considered a true allele
- All marker-level and sample-level PQVs (except SQ) turn gray and maintain their original shape ( ) to indicate that the marker has been edited
- The marker header color changes to gray (pink if selected)



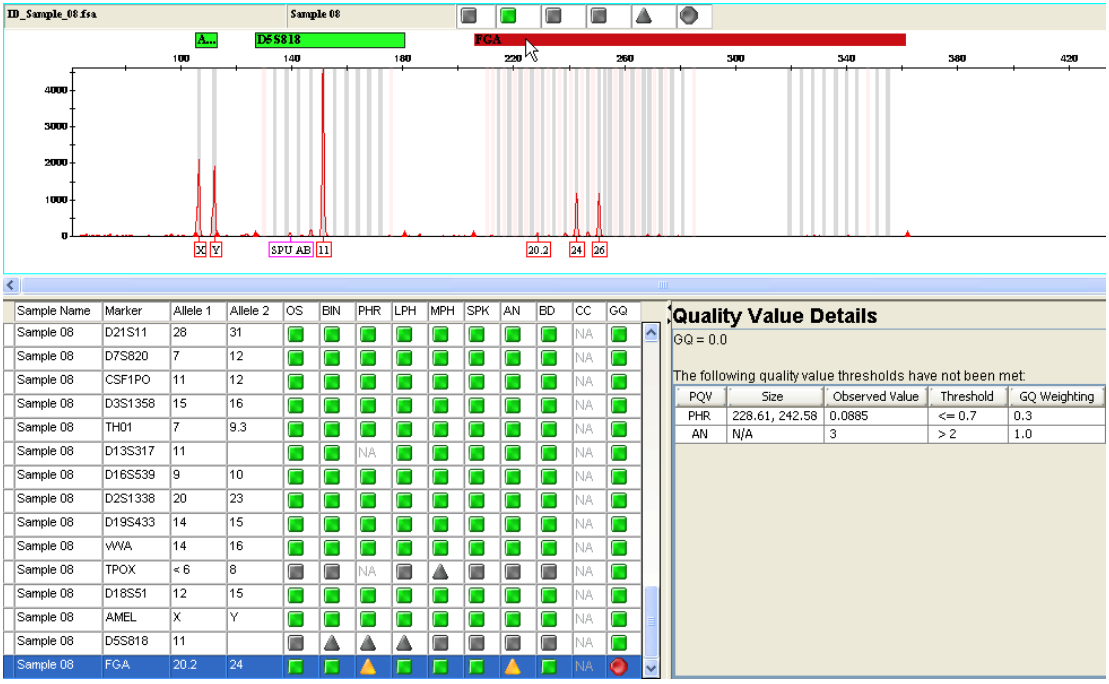
7. Verify the **D5S818** marker row is selected in the Genotypes table, then right-click its GQ PQV.

8. Click **Yes** in the dialog box to override the genotype quality and manually accept the current genotype (11) for the D5S818 marker. The GQ PQV changes to and the marker header turns green.

You have completed reviewing the D5S818 marker. You will now investigate the extra allele present in the FGA marker.

1. Press **Ctrl+Minus Sign (–)** four times to incrementally zoom out on all panes of the Samples plot.

2. Click the red **FGA** marker header in the red dye pane of Sample 08 to display the quality assessment details for this marker.



Note the following:

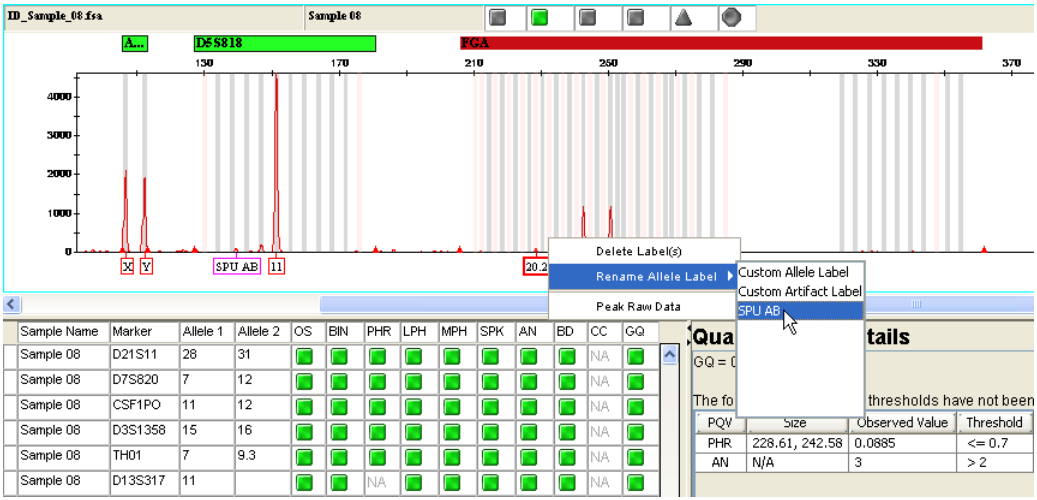
- The software has detected three alleles in the marker range: 20.2, 24 and 26
- The marker-level ⚠ AN PQV is triggered

  Note: A ⚠ AN PQV indicates that the software detects either no alleles or more alleles than the Max Expected Alleles threshold set in the analysis method (in this example, set to two).

- The marker-level ⚠ PHR PQV is triggered

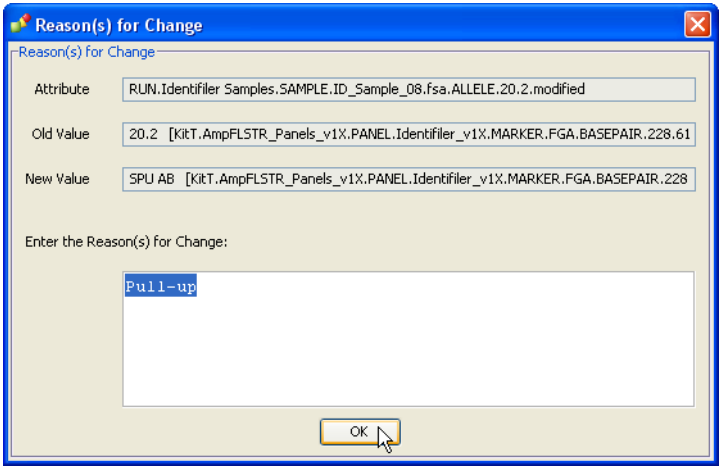
In this example you will assume the extra allele is caused by pull-up as well.

3. Left-click the **20.2** peak label, then right-click the label and select **Rename Allele Label ▸ SPU <your initials>**.



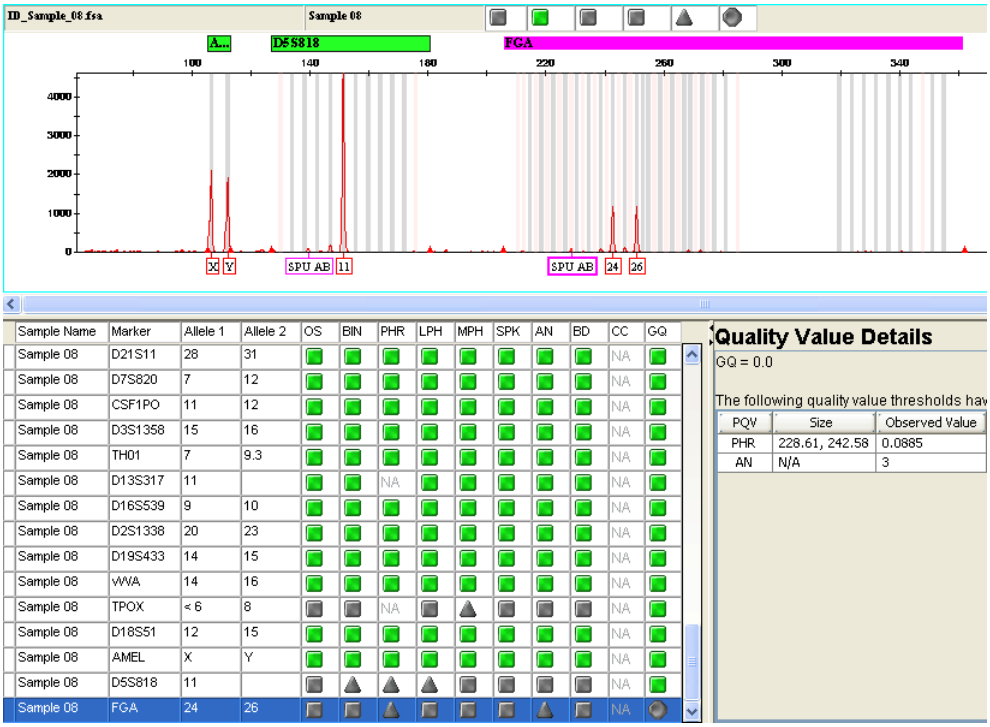
Note: Note that the SPU custom artifact label you created in step 5 on page 125 is now displayed in the list of available labels.

4. In the Reason(s) For Change dialog box, leave the **Pull-up** text as shown (recorded in the audit trail), then click **OK**. You can also press **Alt+O** to close the dialog box.



Note that after editing:

- The SPU artifact label is displayed with a pink border
- The SPU artifact peak is not added to the Genotypes table since an artifact is not considered a true allele
- All PQVs (except SQ) for the FGA marker turn gray and maintain their shape ( ) to indicate that the marker has been edited
- The marker header color changes to gray (pink if selected)



5. Verify the row for the FGA marker is selected in the Genotypes table, then right-click its GQ PQV.

6. Click **Yes** in the dialog box to override the genotype quality and manually accept the current genotype (24, 26) for the FGA marker.

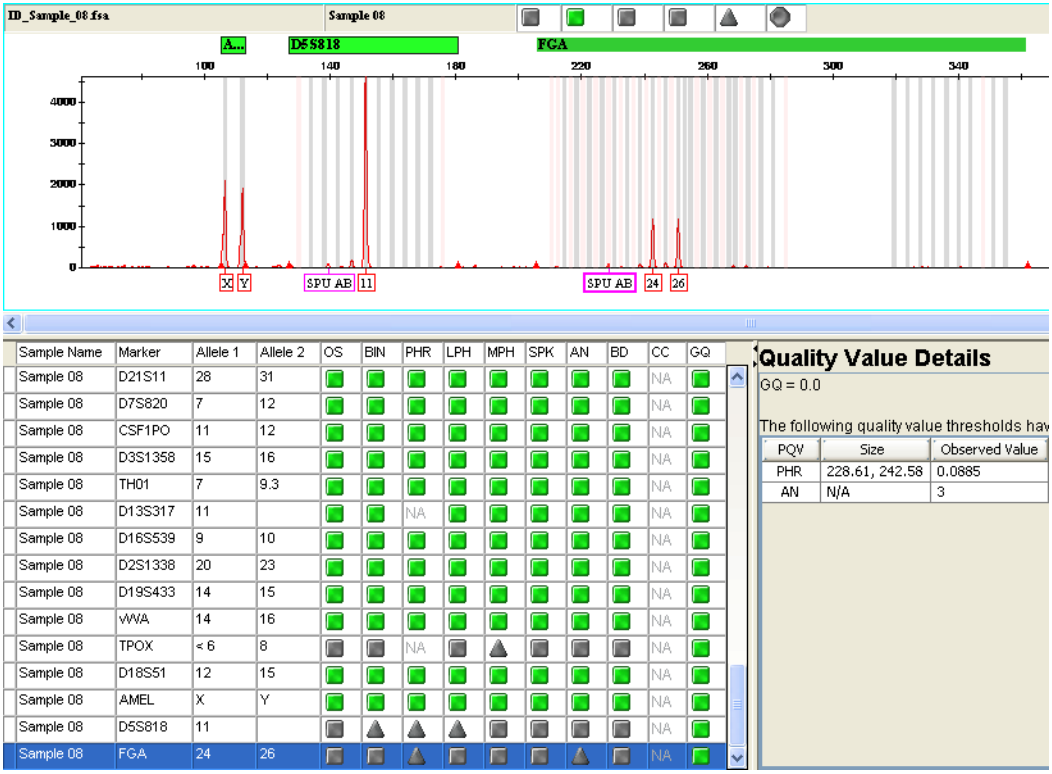
   The next dialog box displays a message indicating that all marker-level GQ PQVs for sample Sample 08 are now green.

7. Click **No** in this Override CGQ dialog box to keep the current CGQ status for this sample.

You will review this sample further in Chapter 6.

Note that after overriding GQ:

- The GQ PQV changes to
- The marker header turns green



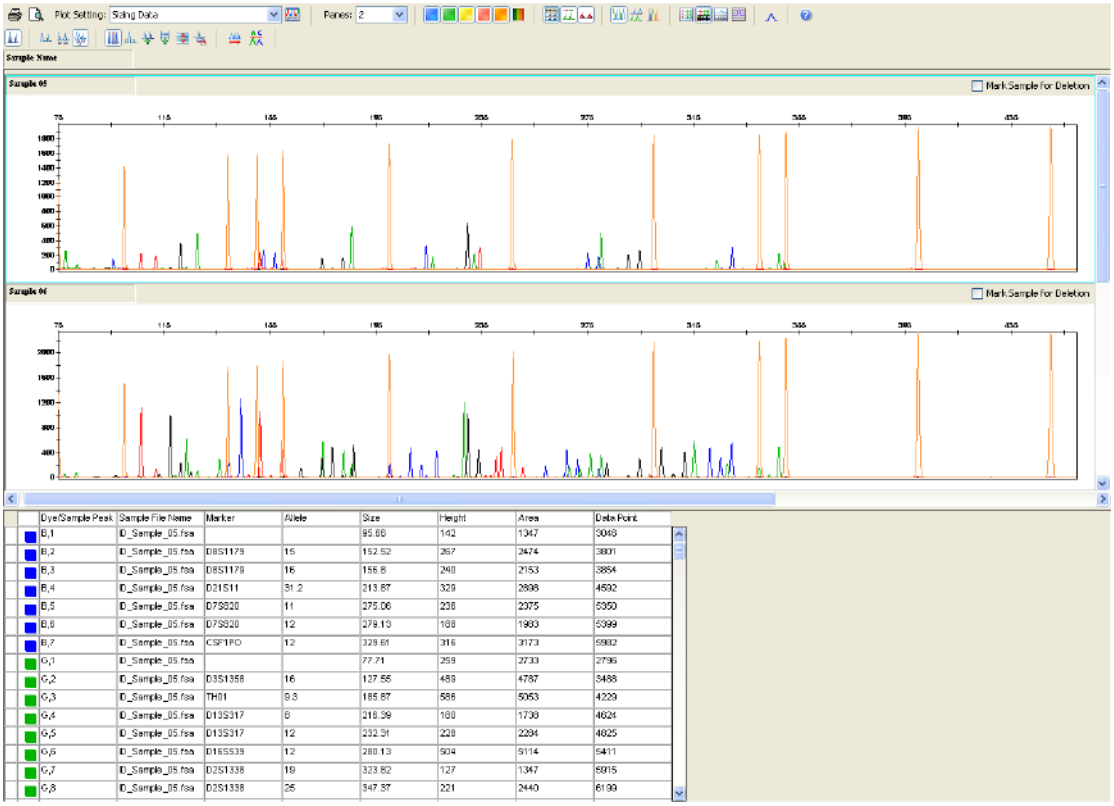
You have now completed reviewing Sample 08.

Step 5: View Additional Plot Settings

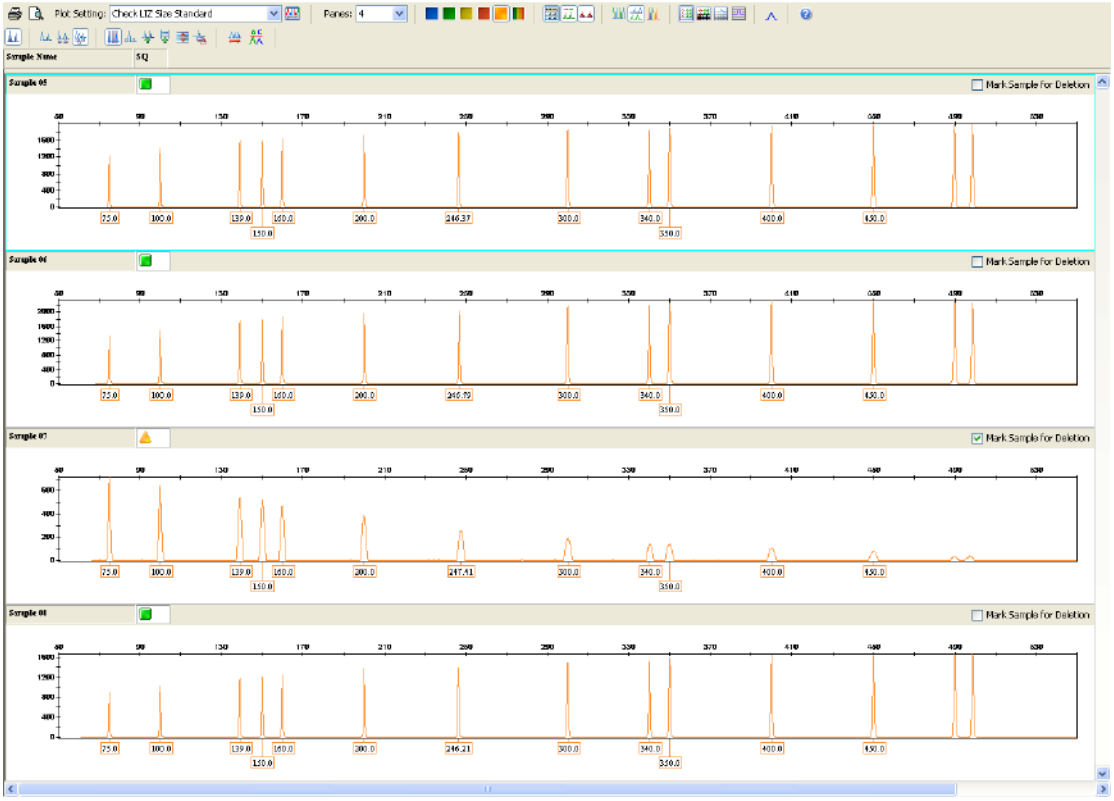
Additional plot settings are supplied with the GeneMapper® *ID-X* Software to assist with performing other functions within the plot window. Follow the procedures on pages 131 through 134 to review the elements of these other plot settings.

Select each of the following from the Plot Setting drop-down list:

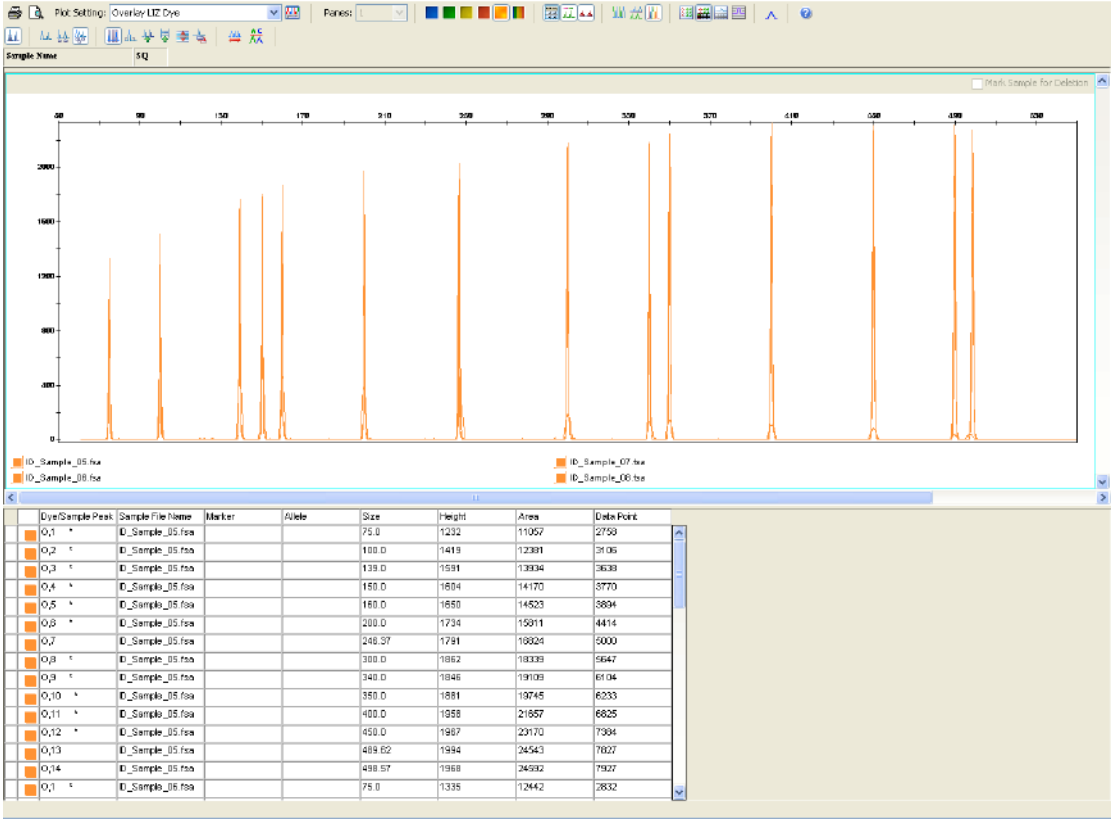
- **Sizing Data** – Displays peak detection and sizing information in a format similar to the GeneScan™ software plots



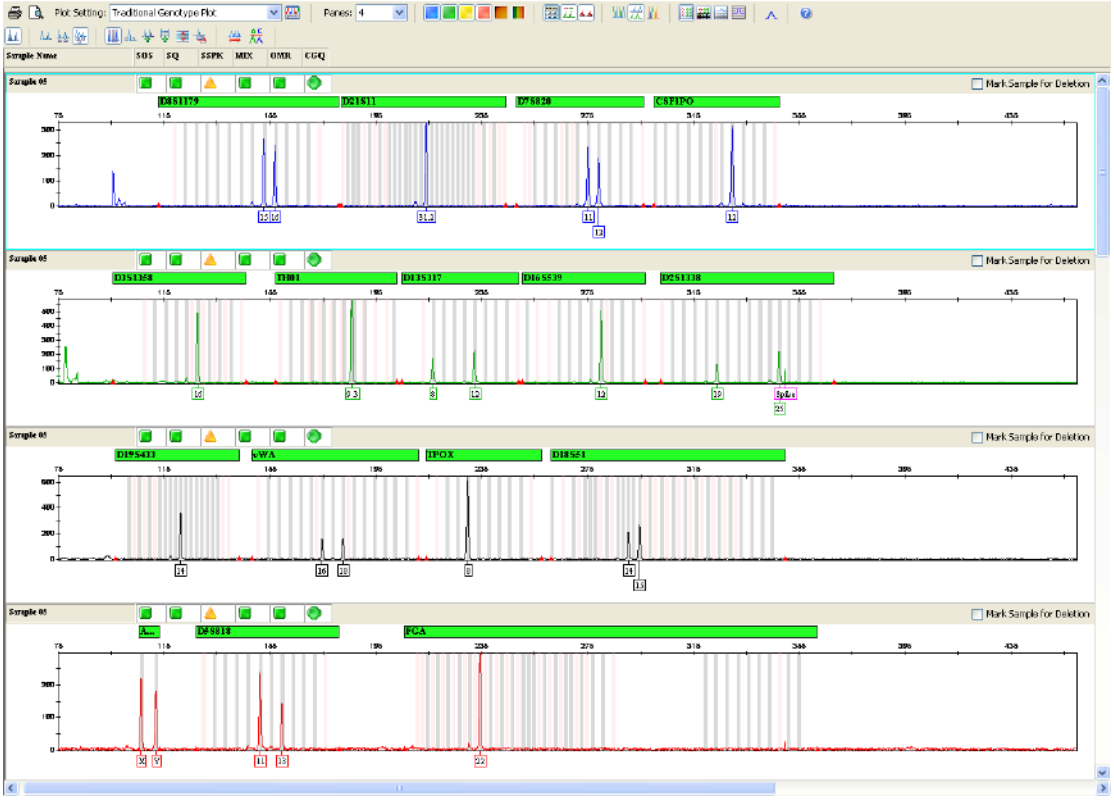
- **Check LIZ Size Standard** – Displays plots in the same format as the Check GS500 Macro in the Genotyper® Software templates



- **Overlay LIZ Dye**– Overlays all selected size standard fragments in one electropherogram pane



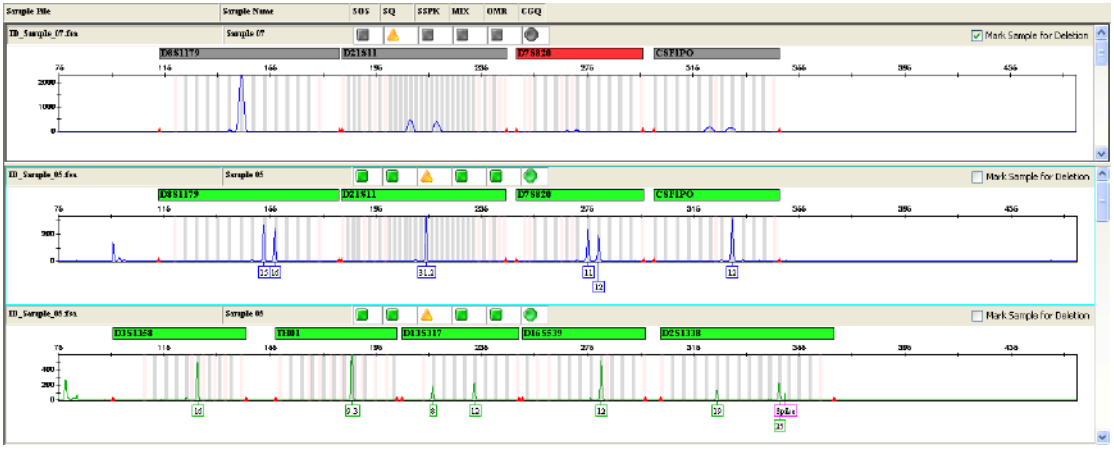
- **Traditional Genotype Plot** – Displays plots in a format similar to the Genotyper® Software



After you have finished reviewing each plot setting, you are now ready to close the Samples plot.

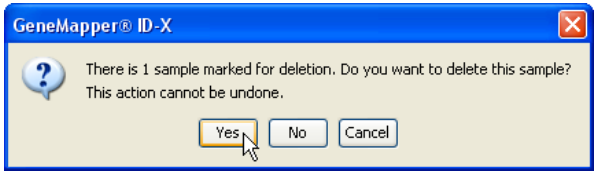
Step 6: Delete Samples from the Project

1. Click (Bring Marked Samples To Top) in the Samples plot toolbar. The sample (Sample 07) you marked for deletion in step 8 on page 110 is displayed in the top pane of the Samples plot.



You can use this feature to verify which samples are marked for deletion before closing the plot window, since closing the plot window automatically deletes these samples from the project.

2. Select **File ▸ Close Plot Window**. A dialog box displays a message indicating that one sample is marked for deletion.

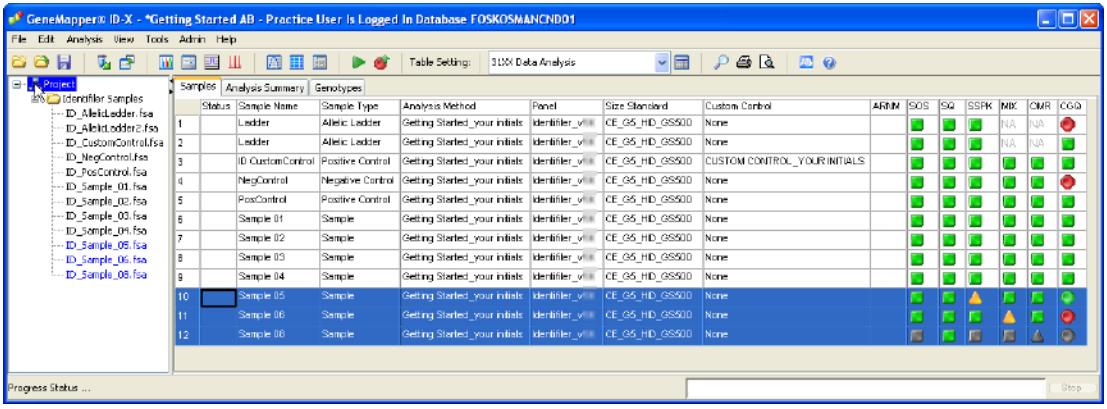


3. Click **Yes** in the dialog box to close the Samples plot and return to the Project window. The software automatically deletes Sample 07 from the Getting Started project.

Note: From step 4 on page 97, the raw data for Sample 05 is still displayed in the Project window.

---

4. Select the **Project** node in the navigation pane to display the remaining samples of the Getting Started project in the Samples table.



Note that Sample 07 is removed from this view. You are now ready to save your edits to the Getting Started project.

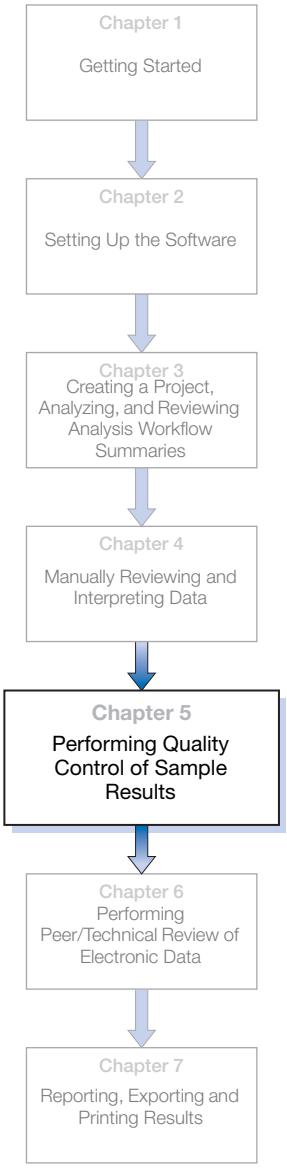
Step 7: Save the Project

Now that you have finished editing the Getting Started project, click (Save Project) to save all project edits (label edits and comments) to the GeneMapper® *ID-X* Software database so they can be viewed the next time the project is opened.

In Chapter 6, you will review your edits to the Getting Started project.

First, in Chapter 5 you will use the Profile Comparison tool to help determine the potential contributors to the negative control with detected peaks and the mixture sample you just reviewed.

5 Performing Quality Control of Sample Results



This chapter covers:

- Overview. 138
- Understanding the Profile Comparison Tool 139
- Step 1: Examine Sample Concordance. 142
- Step 2: Perform Sample Comparison and View Results. . . . 143
- Step 3: Perform Lab Reference Comparison and View Results . 145
- Step 4: Perform Control/QC Comparison and View Results 147
- Step 5: Export Profile Comparison Results 148

Overview

About Quality Control of Sample Results

Use the Profile Comparison tool to perform quality control of sample results in a project. Only samples and controls with ■ or ⚠ SQ are included in comparisons (allelic ladder samples are not included).

The Profile Comparison tool does the following for all samples in a project:

- Groups samples with 100% concordant profiles
- Compares samples in the project against all other samples in the project
- Compares samples in the project against lab reference, custom control, and QC sample profiles stored in the Profile Manager

---

IMPORTANT! Before you use the Profile Comparison tool to evaluate the samples in your project, edit allele labels as needed and ensure that no OL allele labels are present. Samples containing OL-labeled peaks are not considered in comparisons.

In This Chapter

In this chapter, you will use the Getting Started project, saved in Chapter 4, to determine the potential contributors to the Negative Control and mixture samples observed in Chapter 4 by comparing the unknown sample profiles in the project against each other and against the profiles imported into the GeneMapper® *ID-X* Software database in Chapter 2.

Understanding the Profile Comparison Tool

Terms You Need to Know

- **Reference Profile** – The reference profile is the profile against which another profile is compared to determine the % Match. The software performs pairwise comparisons to determine the direction of comparison that yields the higher % Match, then reports only the direction of comparison with the higher % Match.

  A mixed-source sample is used as a reference profile only when it is compared to another mixed-source sample.
- **Comparison Profile** – The comparison profile is compared to the reference profile to determine the % Match result.
- **%-Match** – Calculated using the following formula:

$$\left(\frac{\text{\# reference profile alleles found in comparison profile}}{\text{Total \# reference profile alleles}}\right) \times 100$$

Each sample or group (determined in Sample Concordance tab) is compared to every other individual sample or single-source or mixed-source group in the project. For a pair of samples or groups, two comparisons are performed. Each sample/group is used as a reference, to which the other sample (comparison sample) is compared. The comparison direction that produces the highest % Match is reported.

For example:

– Sample 1 is a single-source sample
– Sample 2 is a mixed-source sample
– Sample 1 contains 10 alleles (all of which are found in Sample 2)
– Sample 2 contains 20 alleles
– When Sample 1 is used as the reference, the % Match = 100%.
– When Sample 2 is used as the reference, the % Match = 50%.

– In this example, the comparison in which Sample 1 is used as the reference is reported.

Note: Results are reported only when the % Match is greater than or equal to the user-defined Percent Match Threshold (range is 50 – 100).

- **Single-source groups** – Samples with no more than three alleles in more than one marker in the Genotypes table, and are 100% concordant (all markers and all alleles match).
- **Mixed-source groups** – Samples with two or more markers with three or more alleles in the Genotypes table, and are 100% concordant.
- **Individual sample**s – Samples that contain unique profiles.
- **Lab Reference and Custom Control Profile** – Profiles imported and stored in the Profile Manager.

Sample Concordance

The software performs a sample concordance check to group samples with identical profiles and minimize the number of comparisons performed on the other tabs of the Profile Comparison tool.

To perform sample concordance, the software:

- Considers all analyzed samples in the open project with a (Pass) or (Check) SQ value, except:
  - Allelic Ladder sample types
  - Samples that contain OL labels
- Compares each sample against every other sample to determine 100% concordance (all markers and all alleles match).
- Groups 100% concordant profiles in one or more single-source or mixed-source groups.

Note: The Sample Concordance tab only displays grouped samples that are 100% concordant. It does not list individual samples. However, individual samples that meet or exceed the user-defined Percent Match Threshold are considered in the Sample Comparison, Lab Reference Comparison, and Control/QC Comparison tabs.

Sample, Lab Reference, Custom Control, and QC Comparison

To perform comparisons, the software:

- For sample comparisons: Performs a pairwise comparison of all individual sample, single-source group, and mixed-source group profiles.
- For lab reference and custom control/QC comparisons: Compares all individual sample, single-source group profiles, and mixed-source group profiles to the lab reference or custom control profiles stored in the Profile Manager (QC comparisons use custom control profiles).
- Calculates the % Match for each comparison.
- Determines the reference-to-comparison % Match to report.

Note: Only the comparison that yields the highest % Match is reported. The comparison that yields the lower % Match is not used or listed in results.

- Displays the % Match results that are greater than or equal to the user-defined Percent Match Threshold.

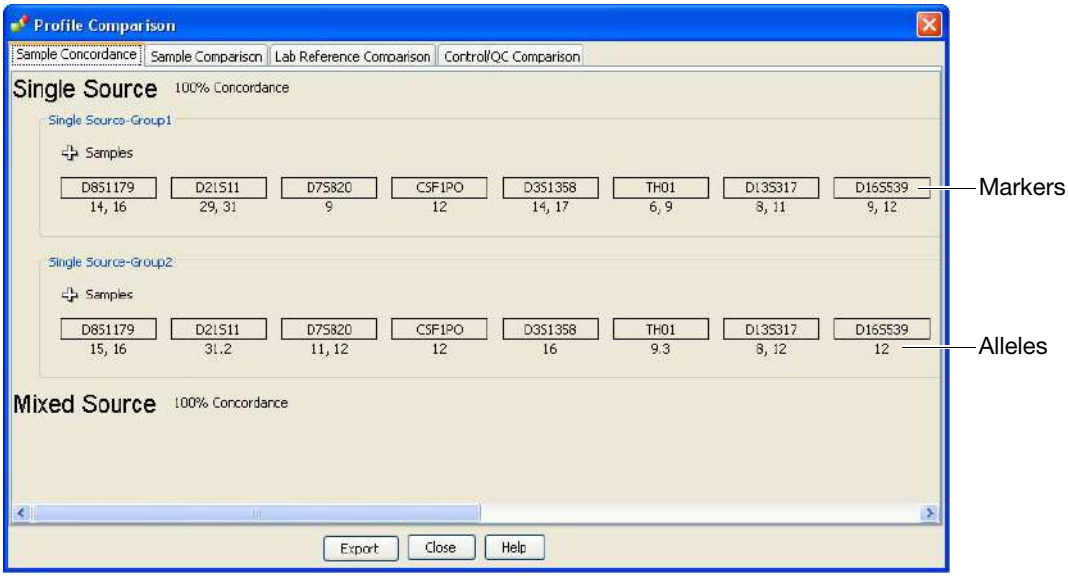
Step 1: Examine Sample Concordance

Overview

In this section, you will use the Profile Comparison tool to check for concordance among all samples in the Getting Started project.

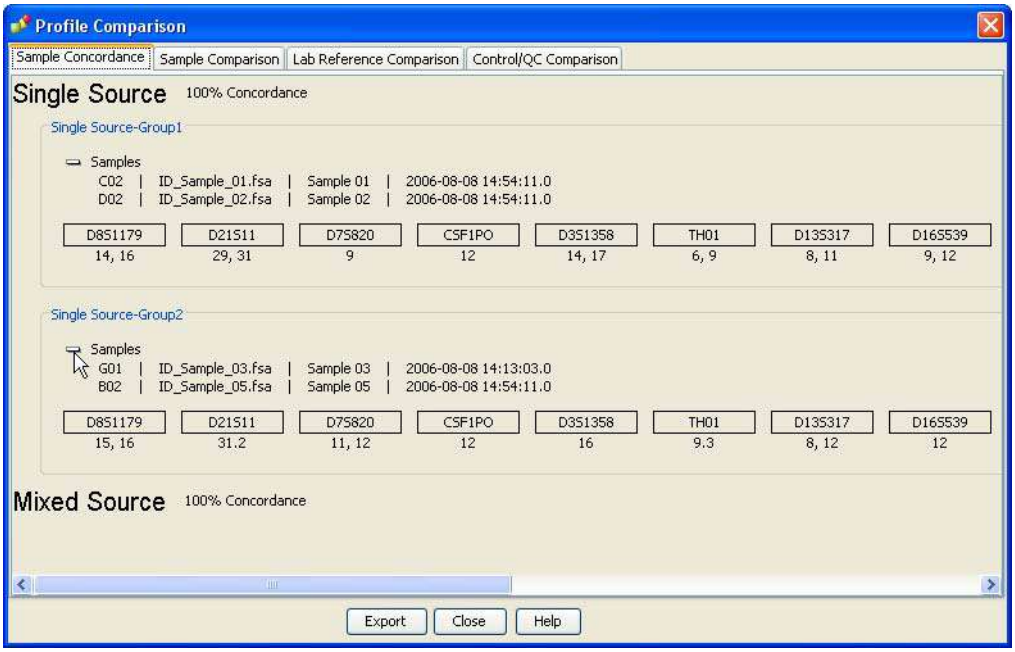
Examining Sample Concordance

1. In the Project window, open the Getting Started project (see step 1 on page 77).
2. Select **Tools ▸ Profile Comparison**. The Profile Comparison tool opens to the Sample Concordance tab.



Two single-source groups (Group 1 and Group 2) are listed on this tab for the Getting Started project.

3. Click ✚ to expand the Samples view for both single-source groups.



In the Getting Started project, ID_Sample_01 and ID_Sample_02 (Single Source-Group 1) are 100% concordant, and ID_Sample_03 and ID_Sample_05 (Single Source-Group 2) are 100% concordant. All other samples in the project have unique profiles and are not shown on this tab.

Step 2: Perform Sample Comparison and View Results

Overview

In this section, you will use the Profile Comparison tool to determine whether any of the sample profiles in the Getting Started project are potential contributors to another sample profile in the project.

Performing Sample Comparison

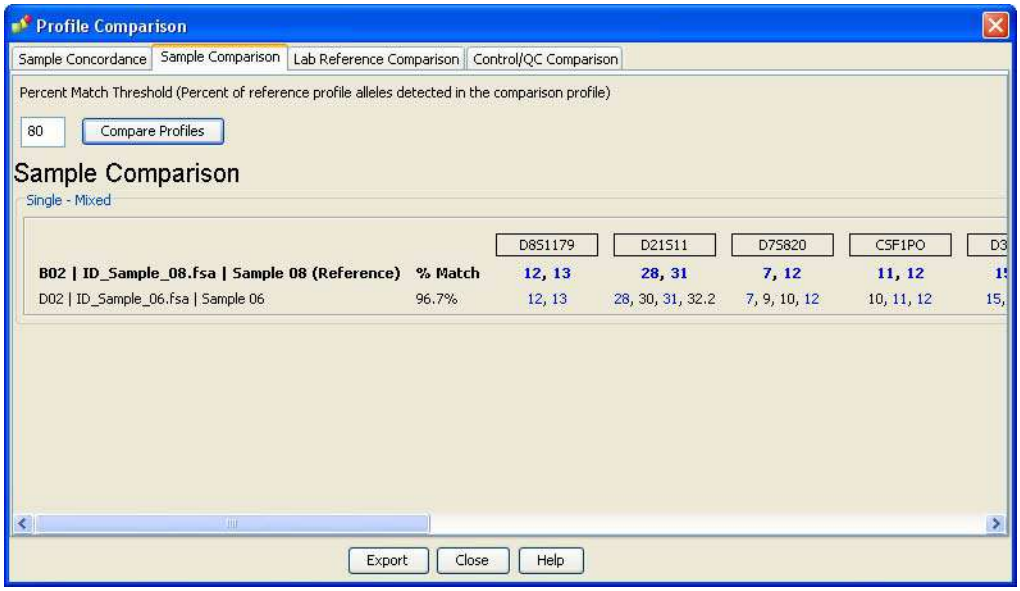
1. Select the **Sample Comparison** tab.
2. Keep the Percent Match Threshold at 80, then click **Compare Profiles**.

Note: The default Percent Match Threshold value is set to 80, with an accepted range of 50–100 percent.

Viewing the Results

Review the sample comparison results.

Note: The matching alleles in the reference sample profile (indicated in **bold**) and the comparison sample profile are displayed in blue.



From Chapter 4, you know that ID_Sample_06 in the Getting Started project is flagged as a potential mixture sample. The sample comparison results indicate that ID_Sample_08 may be contributing to the mixed profile of ID_Sample_06 because ID_Sample_08 (the reference sample) contains 96.7% of the same alleles found in ID_Sample_06 (the comparison sample).

Step 3: Perform Lab Reference Comparison and View Results

Overview

In this section, you will use the Profile Comparison tool to determine whether any of the lab reference sample profiles stored in the Profile Manager are potential contributors to sample profiles in the project.

Note: For information on configuring profile comparison to ignore specified allele labels, refer to the *GeneMapper® ID* Software *Help*.

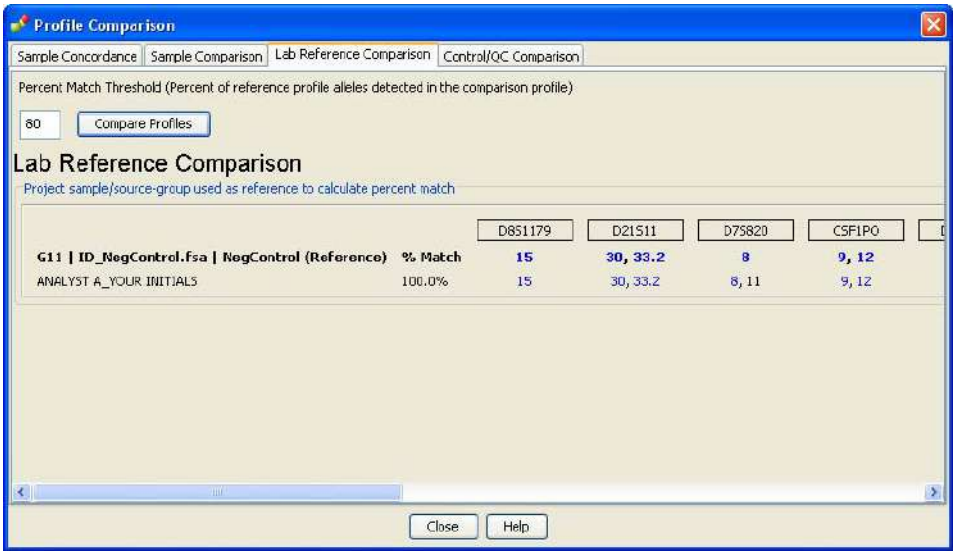
Performing Lab Reference Comparison

1. Select the **Lab Reference Comparison** tab.
2. Keep the Percent Match Threshold at 80, then click **Compare Profiles**.

Viewing the Results

Review the lab reference comparison results.

From Chapter 4, we know that the ID_Negative Control sample in the Getting Started project contains detected, labeled alleles.



The lab reference comparison results indicate that the Analyst A profile (entered as a lab reference profile in Chapter 2) may be a contributor to the Negative Control sample, because all of the alleles detected in the Negative Control sample profile are found in the Analyst A lab reference profile.

Step 4: Perform Control/QC Comparison and View Results

Overview

In this section, you will use the Profile Comparison tool to help perform a blind QC check by comparing the observed profiles for the custom control and QC samples present in the Getting Started project to the custom control profiles stored in the Profile Manager (QC samples use the Custom Control profile type).

Note: For information on configuring profile comparison to ignore specified allele labels, refer to the *GeneMapper® ID* Software *Help*.

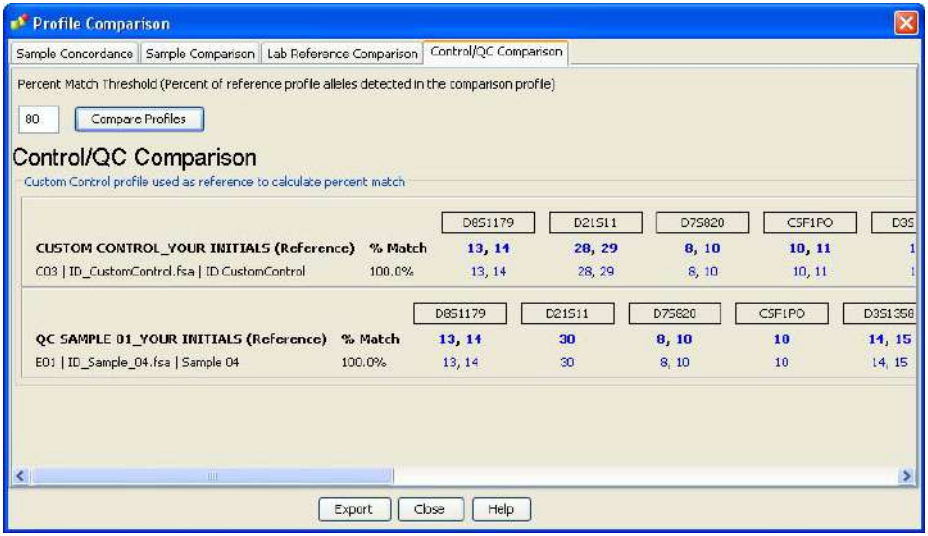
Performing Control/QC Comparison

1. Select the **Control/QC Comparison** tab.
2. Keep the Percent Match Threshold at 80, then click **Compare Profiles**.

Viewing the Results

Review the control/QC comparison results.

In the Getting Started project, ID_Sample_04 is run as a QC sample with profile results expected to match at least one of the custom control profiles stored in the Profile Manager.

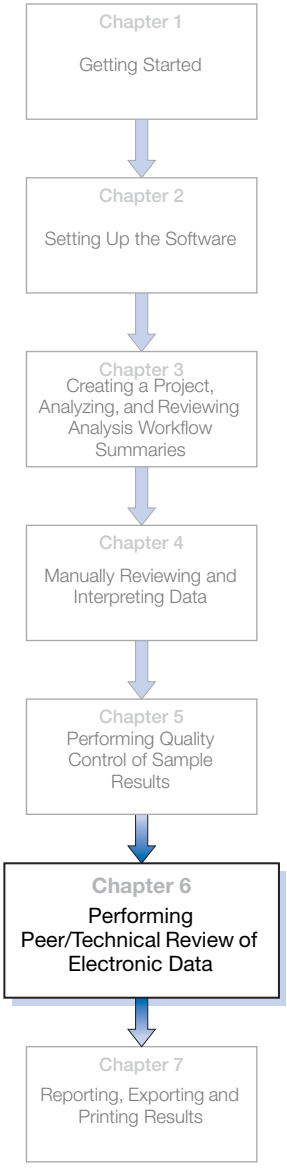


The control/QC comparison results indicate that the QC Sample 01 profile (entered as a custom control profile in the Profile Manager in Chapter 2) matches a stored custom control profile as expected (all alleles in ID_Sample_04 are also found in the QC Sample 01 profile).

Step 5: Export Profile Comparison Results

For information on exporting profile comparison results for further analysis and reporting, refer to the *GeneMapper® ID Software Help*.

6 Performing Peer/Technical Review of Electronic Data



This chapter covers:

- Overview. 150
- Step 1: View Samples with Manual Edits and Overrides. . . 150
- Step 2: View Edits in the Label Edit Viewer 151
- Step 3: View Allele Edits and Comments in the Genotypes Table and Genotypes Plot 157

Overview

About Electronic Peer/Technical Review

The GeneMapper® *ID-X* Software provides several data review tools and features designed to improve the efficiency of electronic peer/technical review of projects. These features may minimize the time spent during manual review and eliminate the need to print electropherograms.

In This Chapter

In this chapter, you will use the Getting Started project results edited in Chapter 4 to learn how to:

- Use table settings to display only those samples containing manual edits or overrides
- Visually confirm manual edits using the Label Edit Viewer
- Export the contents of the Label Edit Viewer
- Use table and plot settings to review manual edits

Step 1: View Samples with Manual Edits and Overrides

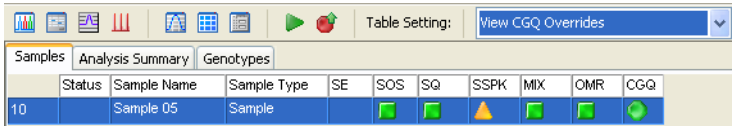
Overview

In this section, you will apply several of the default table settings provided with the software (see Chapter 2) to the Samples table to display only those samples in the Getting Started project that contain manual edits or overrides.

Viewing CGQ Overrides

1. Open the Getting Started project, if not already open (see step 1 on page 77).
2. In the Project window, select **View CGQ Overrides** from the Table Setting drop-down list.

Only those samples in the Getting Started project whose CGQs were manually overridden in Chapter 4 are shown in the Samples table:

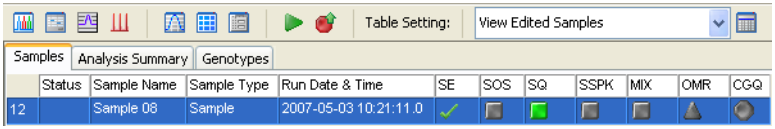


If a sample is displayed in this list, it means during the previous interpretation, the analyst manually accepted the sample profile with or without edits.

Viewing Edited Samples

In the Project window, select **View Edited Samples** from the Table Setting drop-down list.

Only those samples in the Getting Started project that had at least one allele or artifact label edited in Chapter 4 are shown in the Samples table:



Note that there is a ✓ in the Sample Edit (SE) column, and the sample-level PQVs are gray, indicating that peaks within and/or outside of marker ranges have been edited. This table setting allows reviewers to focus on only those samples that have been edited. In an expert system workflow, these may be the only samples that require second review.

Step 2: View Edits in the Label Edit Viewer

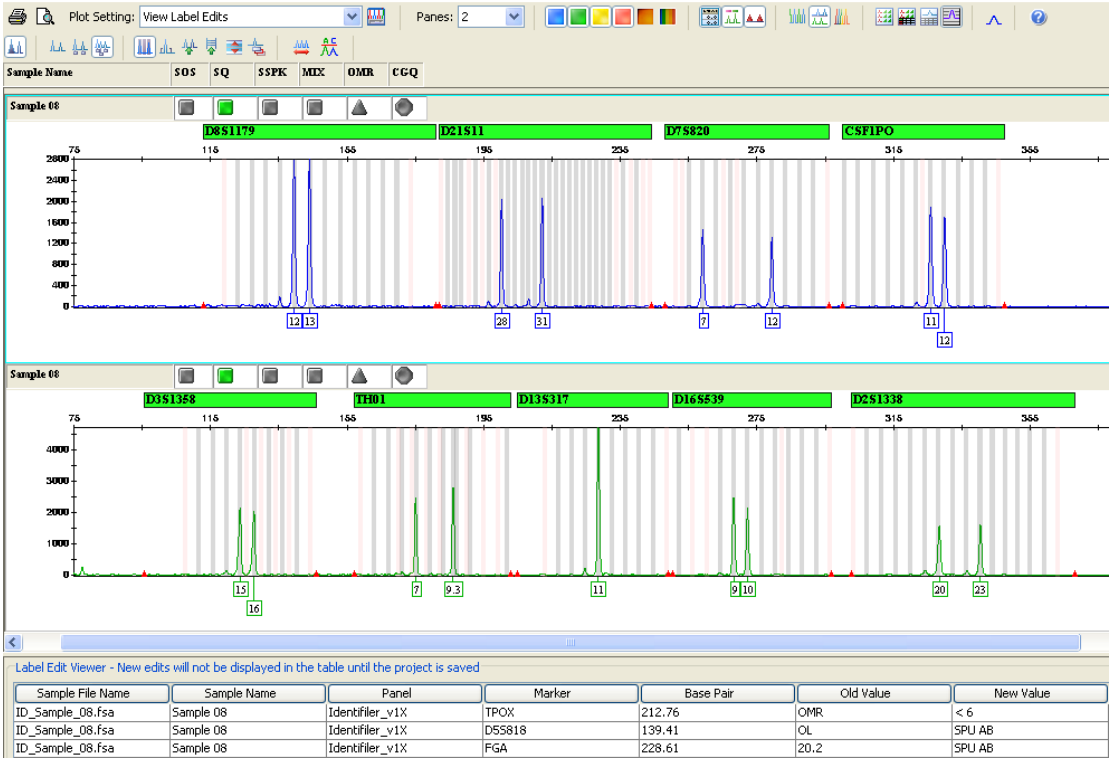
Overview

The Label Edit Viewer contains a list of edits made to the allele and artifact labels displayed in the sample electropherogram plots of the Samples plot. In this section, you will use the Label Edit Viewer to assist in the visual confirmation of manual edits you made to the Getting Started project in Chapter 4.

You can view the Label Edit Viewer from the Project window or the Samples plot.

Viewing Edits from the Samples Plot

1. In the Project window, make sure **View Edited Samples** is selected from the Table Setting drop-down list.
2. Select the edited sample in the filtered Samples table, then click (Display Plots).
3. In the Samples plot, select the **View Label Edits** setting from the Plot Setting drop-down list. The Samples plot view changes.



Sample File Name	Sample Name	Panel	Marker	Base Pair	Old Value	New Value
ID_Sample_08.fsa	Sample 08	Identifiler_v1X	TPOX	212.76	OMR	< 6
ID_Sample_08.fsa	Sample 08	Identifiler_v1X	D5S818	139.41	OL	SPU AB
ID_Sample_08.fsa	Sample 08	Identifiler_v1X	FGA	228.61	20.2	SPU AB

This plot setting displays the sample electropherogram plots above the list of edits shown in the Label Edit Viewer table. In Chapter 4, you made three label edits to Sample 08 in the Getting Started project. The Label Edit Viewer displays detailed information about each of these edits.

Note: The Label Edit Viewer is blank if you have not saved the project.

4. To visually confirm the displayed peak labels for Sample 08 against the label edit entries in the Label Edit Viewer, select the first row in the table. The corresponding edited peak is highlighted in the electropherogram plot.

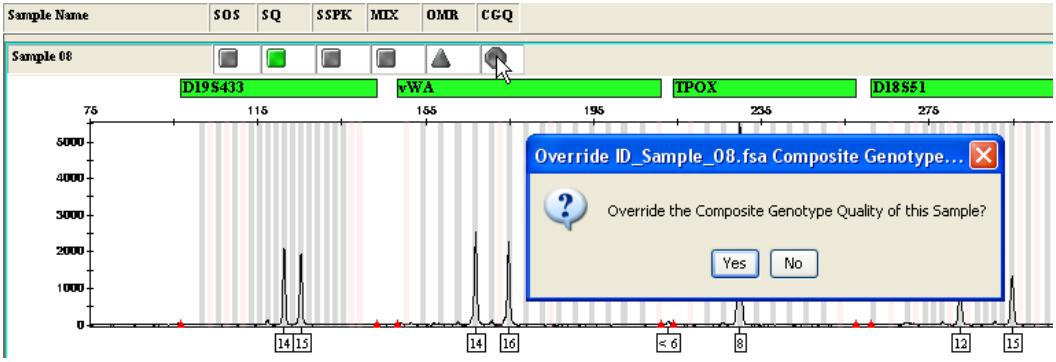
Note: Selecting an edited peak in the electropherogram plot will also highlight the corresponding edit in the Label Edit Viewer.

5. Repeat step 4 above for each entry in the Label Edit Viewer.

6. After you confirm all label edits, manually accept the sample profile by overriding the CGQ PQV:

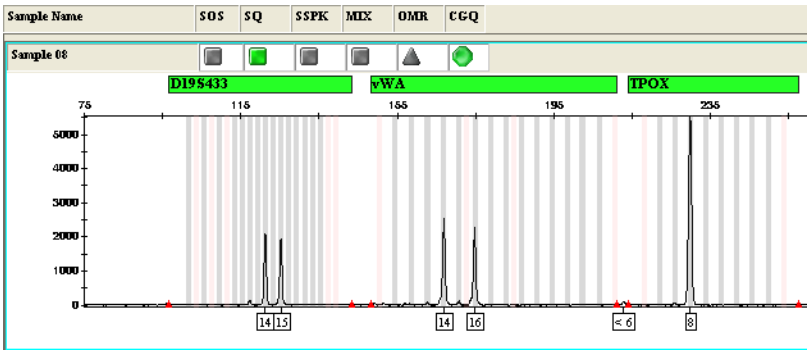
   a. Right-click the CGQ in the Samples plot header.

   Note: A CGQ indicates that one or more peaks within a marker have been edited.



   b. Click **Yes** in the message dialog to override the sample CGQ. The CGQ PQV changes to (Manually Overridden).

   Note: Overriding CGQ allows the reviewer to manually accept the entire profile for a particular sample and also provides evidence that the sample was visually inspected by the reviewer.



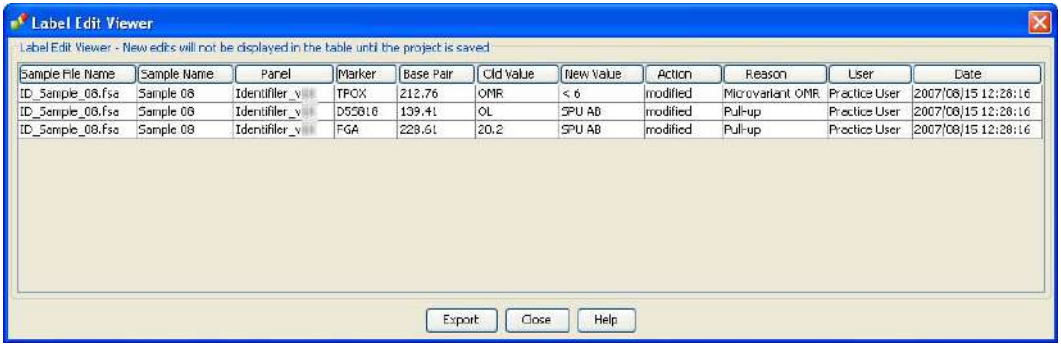
7. Select **File ▸ Close Plot Window** to return to the Project window.

8. Click (Save Project) to save your changes to the Getting Started project.

Viewing Edits from the Project Window

1. In the Project window, make sure **View Edited Samples** is selected from the Table Setting drop-down list.

2. Select the edited sample in the filtered Samples table, then click (Label Edit Viewer). The Label Edit Viewer opens in a separate window.



3. Proceed to the next section if you wish to export the contents of the Label Edit Viewer. Otherwise, click **Close** in the Label Edit Viewer to return to the filtered Samples table.

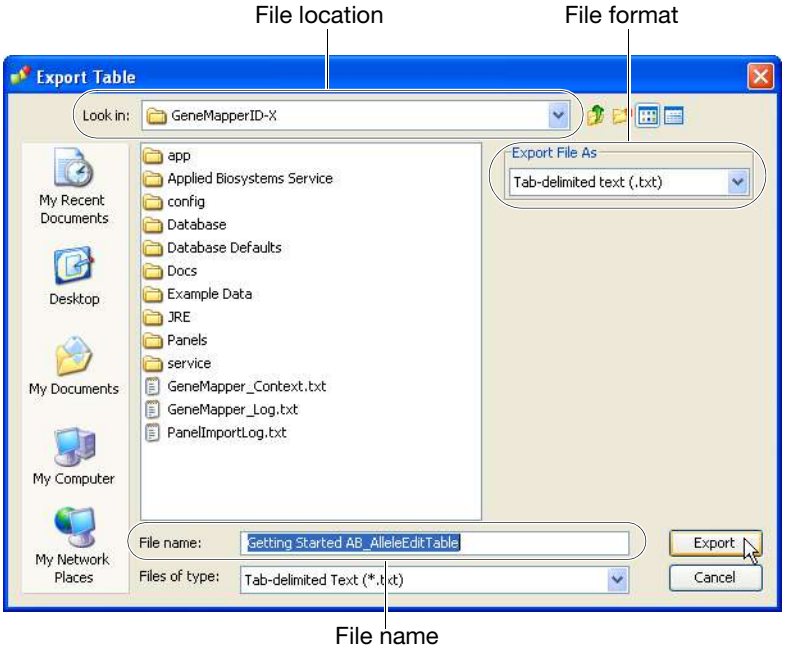
Exporting the Label Edit Viewer from the Project Window

Note: You can also export the contents of the Label Edit Viewer from the Samples plot. See "Exporting from the Samples Plot" on page 172.

1. Display the Label Edit Viewer from the Project window (see step 2 above).

2. In the Label Edit Viewer, click **Export**.

3. In the Export Table dialog, specify a location, name and format (.txt or .csv) for the exported file, then click **Export**.



Note: The Files of type selection filters the list of file names displayed in the navigation pane, it does not determine the format of the exported file.

4. Click **Close** in the Label Edit Viewer to return to the filtered Samples table.

5. Open the exported file with a spreadsheet software that supports tab-delimited or comma-separated text, such as Microsoft® Excel®.

6. Print the exported file as needed.

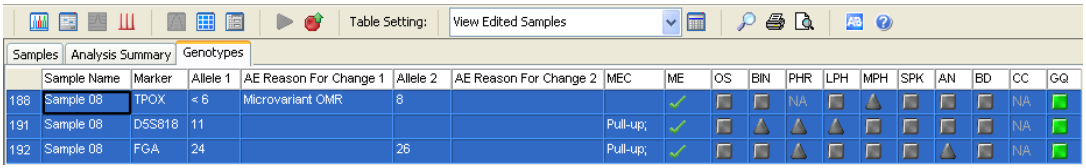
Step 3: View Allele Edits and Comments in the Genotypes Table and Genotypes Plot

Overview

In this section, you will use the Genotypes table and Genotypes plot to view the allele edits and comments for the Getting Started project.

Viewing Allele Calls in the Genotypes Table

1. In the Project window, select the **Project** node in the navigation pane, then select the **Genotypes** tab.
2. Verify that the **View Edited Samples** setting is selected from the Table Setting drop-down list. Only the markers that were edited in the selected sample (in this example, Sample 08) are displayed in the Genotypes table.



	Sample Name	Marker	Allele 1	AE Reason For Change 1	Allele 2	AE Reason For Change 2	MEC	ME	OS	BIN	PHR	LPH	MPH	SPK	AN	BD	CC	GQ
188	Sample 08	TPOX	< 6	Microvariant OMR	8			✓			NA						NA	
191	Sample 08	D5S818	11				Pull-up;	✓									NA	
192	Sample 08	FGA	24		26		Pull-up;	✓									NA	

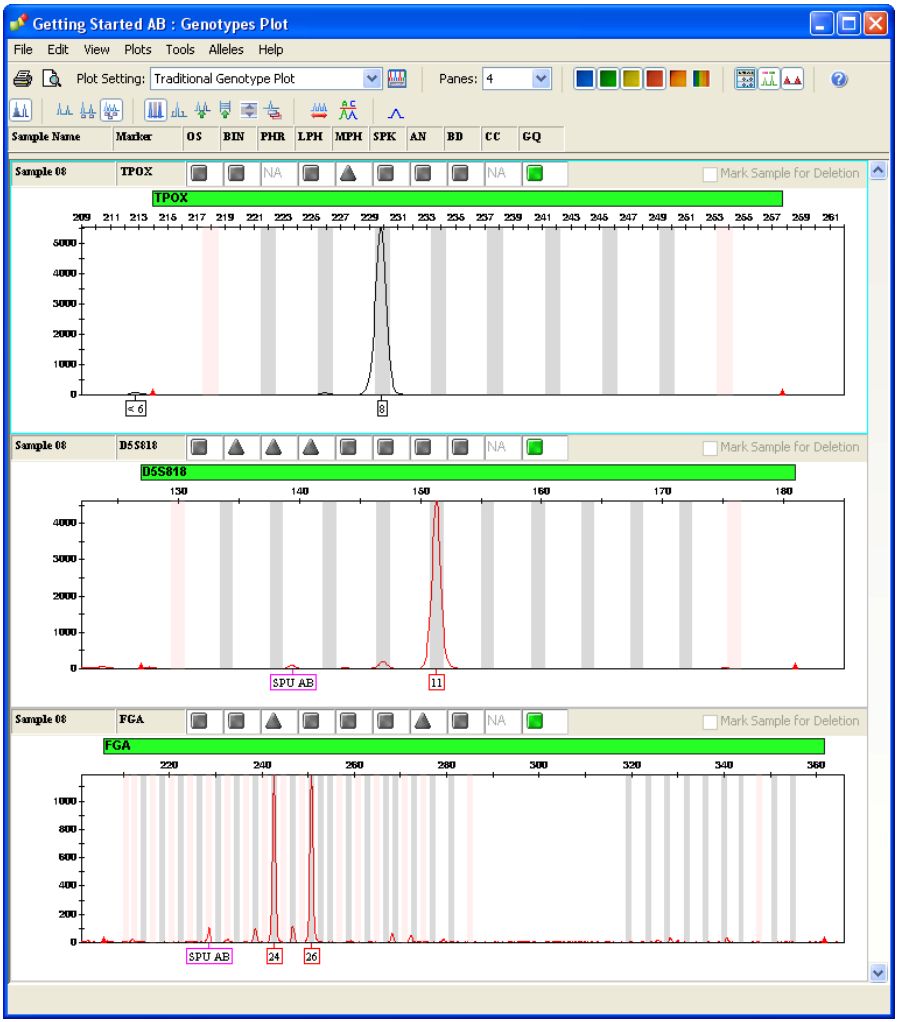
3. Review the entries in the following table columns:
   - **Allele Edit (AE) Reason For Change**: Displays the last reason for change entered for an edit that yields an allele label
   - **Marker Edit Comment (MEC)**: Displays the reason for change entered for an edit that yields an artifact label or when alleles are deleted
   - **Marker Edit (ME) flag**: Displays ✓ (true) if allele or artifact labels are edited within a marker size range

Note: You can export the contents of the Genotypes table from the Project window. This procedure is outlined in "Step 4: Export Table Data" on page 169.

Viewing Allele Calls in the Genotypes Plot

1. In the Project window, Shift-click to select all rows in the Genotypes table (if not already selected), then click (Display Plots). The Genotypes plot window opens.

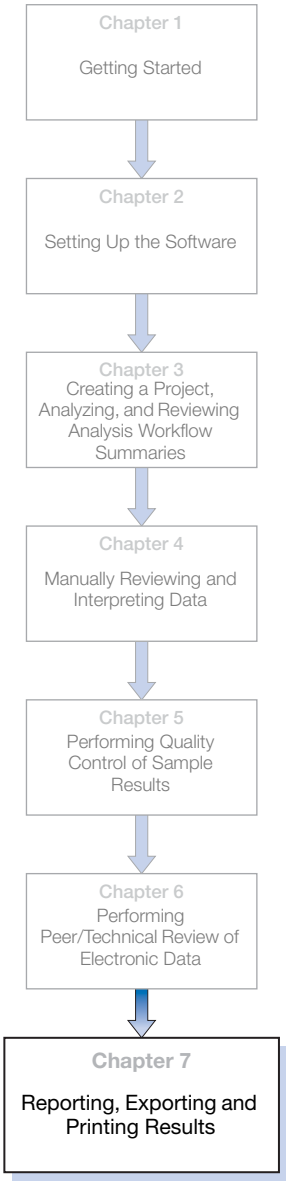
2. In the Genotypes plot, select the **Traditional Genotype Plot** setting from the Plot Setting drop-down list. The Genotypes plot view changes.



This plot setting displays one marker per pane for each of the markers selected in the Genotypes table. For the Getting Started project, only the markers for Sample 08 are displayed in the Genotypes plot.

3. Select **File ▸ Close Plot Window** to return to the Project window.

7 Reporting, Exporting and Printing Results



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- Overview 160
- Step 1: Create a Custom Report Setting 160
- Step 2: Generate the Report 164
- Step 3: Export the Report 166
- Step 4: Export Table Data 169
- Step 5: Print Results 171
- Other Export Options 172

Overview

In this chapter, you will learn how to generate custom reports, and export reports and data tables. You will practice these tasks using the Getting Started project you created using the procedures in Chapter 3 of this guide.

Note: For information on exporting data in a format suitable for CODIS, see the *GeneMapper® ID-X Software Help*.

Step 1: Create a Custom Report Setting

Overview

GeneMapper® *ID-X* Software can generate table-formatted reports from any combination of the columns in the Samples and Genotypes tables. You can configure the report with custom columns, save the report to the project, print it, or export it as tab-delimited or comma-separated text. You can also save the report settings to generate the same report for other projects.

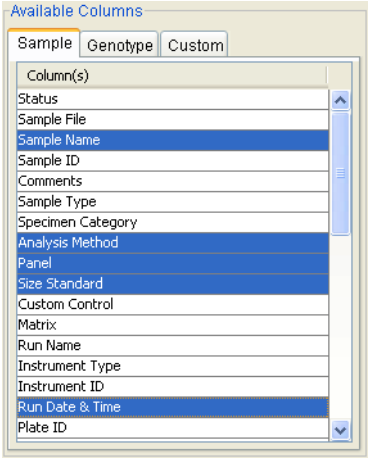
Creating a Custom Report Setting

In this section, you will use the Report Settings Editor to create a new report setting specifically designed for the Getting Started project.

1. Launch the software and log in as the Practice User, if not already logged in (see "Starting the Software and Logging In" on page 22).
2. Open the Getting Started project, if not already open (see step 1 on page 77).
3. In the Project window, click (GeneMapper ID-X Manager).
4. In the GeneMapper ID-X Manager, select the Report Settings tab, then click **New**. The Report Settings Editor window opens.

5. Complete the top portion of the Report Settings Editor:
   a. Enter **Getting Started Report <your initials>** as the report Name.
   b. Enter a description for the new report setting.
   c. Verify that the Practice Security Group is selected from the drop-down list.

6. Select the Samples table columns to display.
   a. In the Sample tab, Ctrl-click to select the following columns from the Available Columns table.

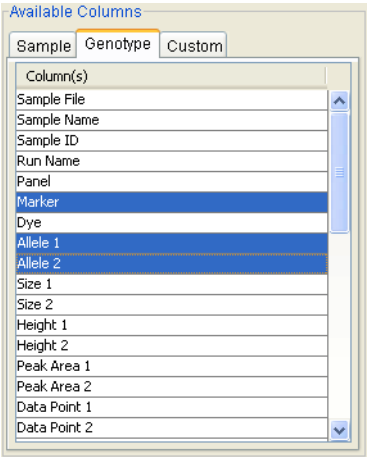


   b. Click to add these columns to the Selected Columns table.

Note: Columns you add to the Selected Columns table are shown in the Preview Table (at the bottom of the Report Settings Editor).

7. Select the Genotypes table columns to display.

   a. In the Genotype tab, Ctrl-click to select the following columns from the Available Columns table.

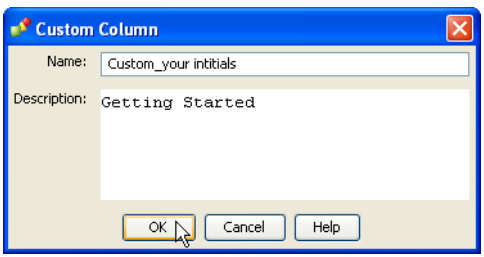


   b. Click to add these columns to the Selected Columns table.

8. Add a custom column to the report. The custom column will appear as a column of blank, editable cells in the report table.

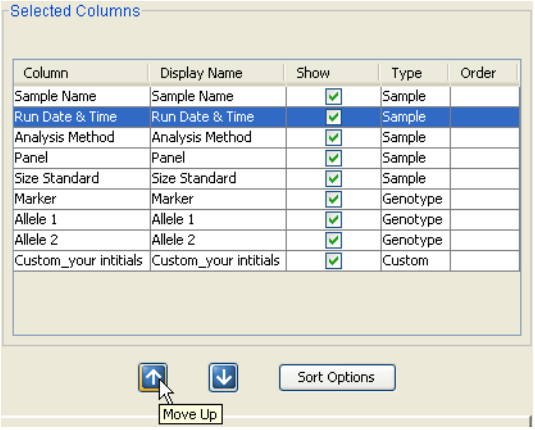
   a. Select the Custom tab, then click **Create.**

   b. Enter the following information, then click **OK.**

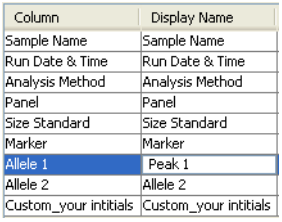


   c. In the Custom tab, select the **Custom <your initials>** column from the Available Columns table, then click to add this new column to the Selected Columns table.

9. Adjust the order that the columns appear in the report.
   a. In the Selected Columns table, select the **Run Date & Time** column.
   b. Click (Move Up) repeatedly until this column is positioned underneath the Sample Name column.



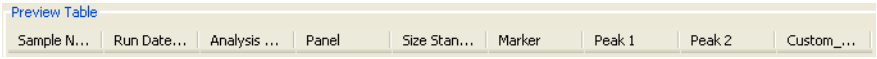
10. Adjust the column header names to be displayed in the report.
    a. In the Display Name column, click the **Allele 1** field, then delete this text and enter **Peak 1**.



    b. Replace Allele 2 with **Peak 2**.

11. In the Number of Alleles field, verify the number of alleles to report is set to **2.**

12. In the Preview Table, review the column display changes you just made to the Getting Started Report setting.



13. Click **OK** to save your settings and close the Report Settings Editor.

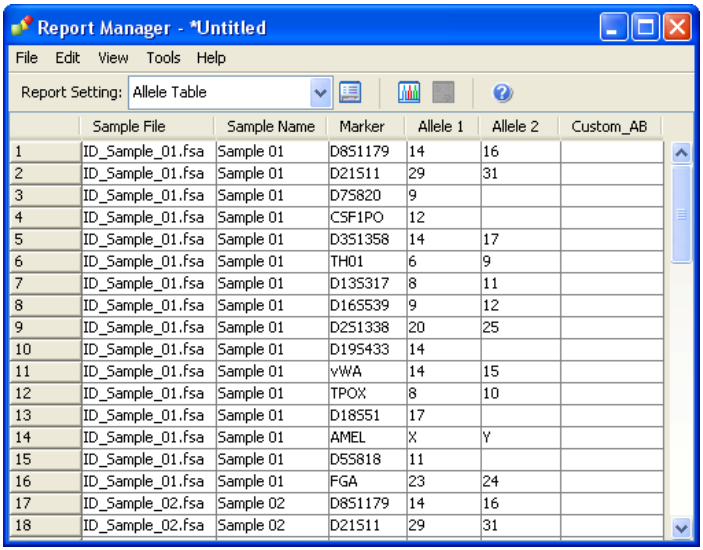
14. Click **Done** to close the GeneMapper ID-X Manager.

Step 2: Generate the Report

1. In the Project window, make sure the Samples tab is selected, then select **View Unedited Samples** from the Table Setting drop-down list.

1. Shift-click to select the rows **Sample 01** through **Sample 04** in the Samples table to report.

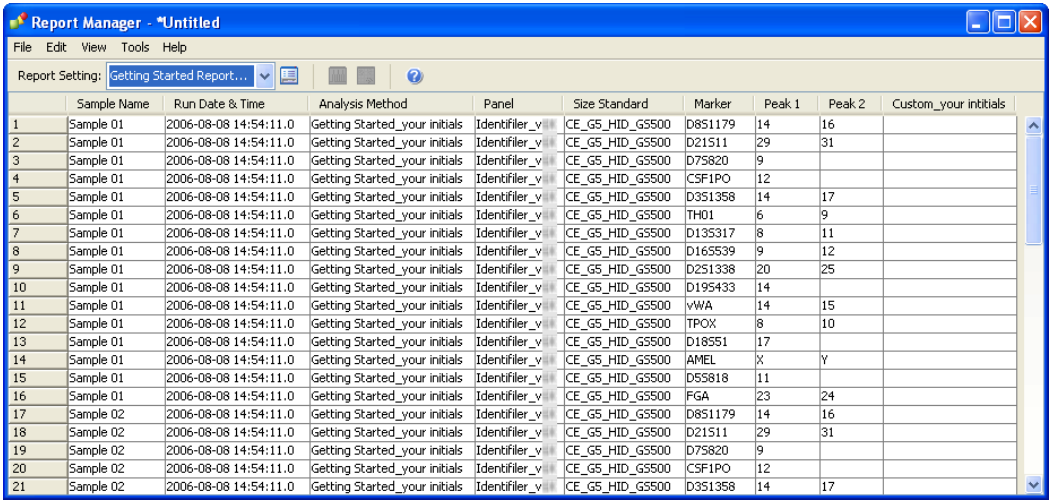
2. Click (Report Manager). A report is displayed in the Report Manager using the first report setting in the drop-down list.



	Sample File	Sample Name	Marker	Allele 1	Allele 2	Custom_AB
1	ID_Sample_01.fsa	Sample 01	D8S1179	14	16	
2	ID_Sample_01.fsa	Sample 01	D21S11	29	31	
3	ID_Sample_01.fsa	Sample 01	D7S820	9		
4	ID_Sample_01.fsa	Sample 01	CSF1PO	12		
5	ID_Sample_01.fsa	Sample 01	D3S1358	14	17	
6	ID_Sample_01.fsa	Sample 01	TH01	6	9	
7	ID_Sample_01.fsa	Sample 01	D13S317	8	11	
8	ID_Sample_01.fsa	Sample 01	D16S539	9	12	
9	ID_Sample_01.fsa	Sample 01	D2S1338	20	25	
10	ID_Sample_01.fsa	Sample 01	D19S433	14		
11	ID_Sample_01.fsa	Sample 01	vWA	14	15	
12	ID_Sample_01.fsa	Sample 01	TPOX	8	10	
13	ID_Sample_01.fsa	Sample 01	D18S51	17		
14	ID_Sample_01.fsa	Sample 01	AMEL	X	Y	
15	ID_Sample_01.fsa	Sample 01	D5S818	11		
16	ID_Sample_01.fsa	Sample 01	FGA	23	24	
17	ID_Sample_02.fsa	Sample 02	D8S1179	14	16	
18	ID_Sample_02.fsa	Sample 02	D21S11	29	31	

3. Select the **Getting Started Report <your initials>** setting you created from the Report Setting drop-down list.

4. Review the report. Note the following:
   - The report contains the columns specified in the report setting, including the Custom column
   - The Allele 1 and Allele 2 columns are reported as Peak 1 and Peak 2



	Sample Name	Run Date & Time	Analysis Method	Panel	Size Standard	Marker	Peak 1	Peak 2	Custom_your intitials
1	Sample 01	2006-08-08 14:54:11.0	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	D8S1179	14	16	
2	Sample 01	2006-08-08 14:54:11.0	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	D21S11	29	31	
3	Sample 01	2006-08-08 14:54:11.0	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	D7S820	9		
4	Sample 01	2006-08-08 14:54:11.0	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	CSF1PO	12		
5	Sample 01	2006-08-08 14:54:11.0	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	D3S1358	14	17	
6	Sample 01	2006-08-08 14:54:11.0	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	TH01	6	9	
7	Sample 01	2006-08-08 14:54:11.0	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	D13S317	8	11	
8	Sample 01	2006-08-08 14:54:11.0	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	D16S539	9	12	
9	Sample 01	2006-08-08 14:54:11.0	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	D2S1338	20	25	
10	Sample 01	2006-08-08 14:54:11.0	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	D19S433	14		
11	Sample 01	2006-08-08 14:54:11.0	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	vWA	14	15	
12	Sample 01	2006-08-08 14:54:11.0	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	TPOX	8	10	
13	Sample 01	2006-08-08 14:54:11.0	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	D18S51	17		
14	Sample 01	2006-08-08 14:54:11.0	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	AMEL	X	Y	
15	Sample 01	2006-08-08 14:54:11.0	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	D5S818	11		
16	Sample 01	2006-08-08 14:54:11.0	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	FGA	23	24	
17	Sample 02	2006-08-08 14:54:11.0	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	D8S1179	14	16	
18	Sample 02	2006-08-08 14:54:11.0	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	D21S11	29	31	
19	Sample 02	2006-08-08 14:54:11.0	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	D7S820	9		
20	Sample 02	2006-08-08 14:54:11.0	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	CSF1PO	12		
21	Sample 02	2006-08-08 14:54:11.0	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	D3S1358	14	17	

5. To edit a cell in the Custom column, double-click the cell, type the desired text, then press **Enter**.

	Sample Name	Run Date & Time	Analysis Method	Panel	Size Standard	Marker	Peak 1	Peak 2	Custom_your initials
1	Sample 01	2006-08-08 14:54:11.0	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	D8S1179	14	16	enter text here
2	Sample 01	2006-08-08 14:54:11.0	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	D21S11	29	31	
3	Sample 01	2006-08-08 14:54:11.0	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	D7S820	9		
4	Sample 01	2006-08-08 14:54:11.0	Getting Started_your initials	Identifiler_v	CE_G5_HID_GS500	CSF1PO	12		

6. Leave the Report Manager window open for the next steps (see “Step 3: Export the Report” on page 166).

Step 3: Export the Report

Overview

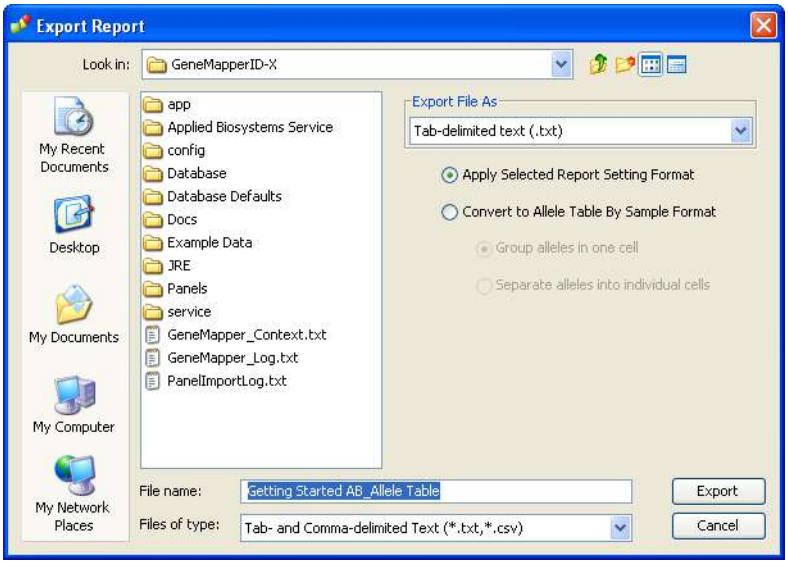
You can export the report generated in the Report Manager as a formatted table. The exported table format depends on the report setting selected when generating the report. By default, the export table format reflects the display in the Report Manager window. However, you can also export in allele table format to list markers in columns or rows, and to group the called alleles for a marker into one or more cells, depending on the number of alleles selected to be displayed.

Exporting the Report

1. In the Report Manager, select **Allele Table** from the Report Setting drop-down list.

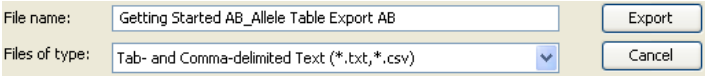
   Note: You can use this report setting to export genotype data in a traditional allele table format (see step 6 on page 167).

2. Select **File ▸ Export** to open the Export Report dialog box:



3. Navigate to the location in which to save the file.

4. Enter **Export <your initials>** as a suffix to the default Getting Started <your initials>_Allele Table export File name.

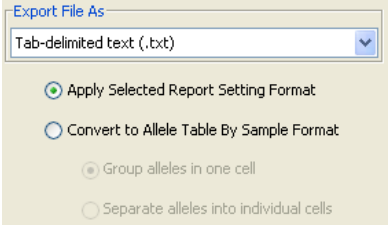


Note: The Files of type selection filters the list of file names displayed in the navigation pane, it does not determine the format of the exported file.

5. Verify that **Tab-delimited text (*.txt)** is selected as the export file format.

Note: This selection separates the alleles with tab-stops, and allows you to open the exported file using a spreadsheet application (see step 11 on page 168).

6. Select a format option:



- **Apply Selected Report Setting Format** (default) – Exports the report as it is displayed in the Report Manager (one marker per row)

	1	2	3	4	5
1	Sample File	Sample Name	Marker	Allele 1	Allele 2
2	ID_Sample_01.fsa	Sample 01	D8S1179	14	16
3	ID_Sample_01.fsa	Sample 01	D21S11	29	31
4	ID_Sample_01.fsa	Sample 01	D7S820	9	
5	ID_Sample_01.fsa	Sample 01	CSF1PO	12	

- **Convert to Allele Table by Sample Format** – Generates a list of samples (one sample per row) with column(s) for each marker that lists the called alleles for the marker

Note: Markers without called alleles are also listed.

For heterozygote alleles, you can also choose to:

- **Group alleles in one cell** – Generates a single column for each marker and places all alleles for the marker into one cell (that is, a traditional allele table)

	1	2	3	4	5
1	Sample File	Sample Name	D8S1179	D21S11	D7S820
2	ID_Sample_01.fsa	Sample 01	14, 16	29, 31	9,
3	ID_Sample_02.fsa	Sample 02	14, 16	29, 31	9,
4	ID_Sample_03.fsa	Sample 03	15, 16	31.2,	11, 12
5	ID_Sample_04.fsa	Sample 04	13, 14	30,	8, 10

- **Separate alleles into individual cells** – Generates multiple columns per marker and places only one allele per cell

	1	2	3	4	5	6
1	Sample File	Sample Name	D8S1179 1	D8S1179 2	D21S11 1	D21S11 2
2	ID_Sample_01.fsa	Sample 01	14	16	29	31
3	ID_Sample_02.fsa	Sample 02	14	16	29	31
4	ID_Sample_03.fsa	Sample 03	15	16	31.2	
5	ID_Sample_04.fsa	Sample 04	13	14	30	

7. Click **Export**.

Note: If you are exporting an allele table, a dialog box displays the names of the files created. Click **OK**.

---

8. Select **File ▸ Exit** to close the Report Manager.
9. Click **Yes** in the Save Report dialog, then enter **Getting Started_Allele Table Report <your initials>** in the Save dialog.
10. Click **OK** to save the Allele Table Report to the Getting Started project.
11. Navigate to and open the exported **Getting Started_Allele Table Export** file in a spreadsheet software (such as Microsoft® Excel®) or Microsoft® Notepad.

Step 4: Export Table Data

IMPORTANT! When exporting or copying and pasting PQV flags, include the ME column (from the Genotypes table) and the SE column (from the Samples table) to indicate whether allele labels were edited, which may affect individual PQV values.

Exporting Individual Tables

When you export data from the Samples or Genotypes table individually, the following are exported in a single table:

- All samples in the project (not selected samples)
- Only the displayed columns from the individual table that you are viewing

To export individual tables:

1. In the Project window, select **View ▸ Samples**, then select the Samples tab to export the Samples table or the Genotypes tab to export the Genotypes table.
2. Select **View Unedited Samples** from the Table Setting drop-down list.
3. Shift-click a column header in the Samples tab (or Genotypes tab) to sort the table and determine the sample order.
4. Click (Export Table).
5. In the dialog box, navigate to the location to save the exported table file.
6. Select the export file format:
   - **Tab-delimited text (*.txt)**
   - **Comma-delimited values (*.csv)**

7. Enter a name for the exported file.

   Note: The Files of type selection filters the list of file names displayed in the navigation pane, it does not determine the format of the exported file.

8. Click **Export Table**.

Exporting Combined Tables

When you export data from the Samples table and Genotypes table together, the following are exported in a combined table:

- All samples in the project (not selected samples)
- Only the displayed columns from the Samples table and Genotypes table

To export combined tables:

1. In the Project window, select **View Unedited Samples** from the Table Setting drop-down list.
2. Select **File ▸ Export Combined Table**.
3. In the dialog box, navigate to the location to save the exported table file.
4. Select the export file format (***.txt** or ***.csv**).
5. Select a Merge option:
   - **Allele table by sample**

     IMPORTANT! This option does not allow you to control the order of columns in the exported file. Columns are exported in the order in which they appear in the Samples and Genotypes tables. To control the order of columns in the exported file, export from the Report Manager. See "Step 3: Export the Report" on page 166.

   - **One line per sample**
   - **One line per marker** (default)

6. Enter a name for the exported file.
7. Click **Export**.

Note: If you are exporting an allele table, a dialog box displays the names of the files created. Click **OK**.

Copying and Pasting Table Data

You can copy and paste the content of most tables into a spreadsheet or text file.

1. In the desired table of the GeneMapper® *ID-X* Software, select the cells to copy.
2. To copy the data:
   a. **Without column headers** – Press **Ctrl+C**.
   b. **With column headers** – Press **Ctrl+Shift+C**.
3. In the desired location (spreadsheet or text file), select **Edit ▸ Paste**.

Step 5: Print Results

Use the print preview function in the GeneMapper® *ID-X* Software to examine data items (reports, tables, plots, sample information, raw data, and EPT data) on screen before they print.

When you are satisfied with the results, select **File ▸ Print**:

Window/Tab	Access From Project Window By Selecting
Project window – Samples tab	**View ▸ Samples**
Project window – Genotypes tab	**View ▸ Genotypes**
Project window – Info tab	**View ▸ Sample Info**
Project window – Raw Data tab	**View ▸ Raw Data**
Project window – EPT Data tab	**View ▸ EPT Data**

Window/Tab	Access From Project Window By Selecting
Samples plot	The Samples tab, then **View ▸ Display Plots**
Genotypes plot	The Genotypes tab, then **View ▸ Display Plots**
Report Manager	**Tools ▸ Report Manager**
Size Match Editor	**Tools ▸ Size Match Editor**

Printing to PDF

To print to PDF, you must install a PDF print driver.

Other Export Options

Exporting from the Samples Plot

You can export results from the following tables by selecting **File ▸ Export Table** in the Samples plot:

Table	Access From Samples Plot By Selecting
Genotypes table	**Plots ▸ Tables ▸ Genotypes Table**
Label Edit Viewer	**Plots ▸ Tables ▸ Label Edit Viewer**
Sizing table	**Plots ▸ Tables ▸ Sizing Table**

Exporting from the GeneMapper ID-X Manager

You can export the following data objects stored in the GeneMapper® *ID-X* Software database by clicking **Export...** in the GeneMapper ID-X Manager dialog:

- Projects
- Analysis Methods
- Table Settings
- Plot Settings
- Matrices

  Note: You can export a matrix file for use in the Data Collection Software on the 310 Genetic Analyzer.

- Size Standards

- Report Settings

Exporting from the Panel Manager

You can export the following data objects stored in the GeneMapper® *ID-X* Software database by selecting **File ▸ Export...** in the Panel Manager window:

- Panels
- Bin Set
- All Kits
- Marker Stutter

Exporting from the Profile Manager

You can export the following data objects stored in the GeneMapper® *ID-X* Software database by clicking **Export** in the Profile Manager dialog:

- Lab Reference profiles
- Custom Control profiles

Note: All of the profiles stored in the Profile Manager will be exported together in a single file.

Exporting using the command line interface

You can export the following data objects stored in the GeneMapper® *ID-X* Software database using the command line interface described in the *GeneMapper® ID-X Software Administrator's Guide*:

- Projects
- Analysis Methods
- Size Standards

Documentation and support

Related documentation

Document	Part number	Description
GeneMapper® ID-X Software Version 1.5 Installation Guide	100031706	Explains how to install the GeneMapper *ID-X* software.
GeneMapper® ID-X Software Version 1.5 Getting Started Guide — Basic Features	100031701	Explains how to set up the software, set up a project, and analyze the example data provided with the GeneMapper *ID-X* Software.
GeneMapper® ID-X Software Version 1.5 Quick Reference Card — Basic Features	100031702	Provides an easy-to-follow workflow for using basic system features.
GeneMapper® ID-X Software Version 1.5 Getting Started Guide — Mixture Analysis Tool	100031704	Explains how to set up the software for mixture analysis and analyze the example data provided with the GeneMapper *ID-X* Software.
GeneMapper® ID-X Software Version 1.5 Quick Reference Card — Mixture Analysis Tool	100031705	Provides an easy-to-follow workflow for using the mixture analysis tool.
GeneMapper® ID-X Software Version 1.5 Reference Guide	100031707	Describes process quality values (PQVs) and algorithms.
GeneMapper® ID-X Software Version 1.5 Administrator's Guide	100031703	Describes how to configure the administrative features of the software, maintain the GeneMapper® *ID-X* Software and database dashboard software, and use the command-line interface.
GeneMapper® ID-X Software Version 1.5 Integration User Bulletin	100031709	Describes command line interface commands that can be scripted and used to automate GeneMapper® *ID-X* Software operations.

How to obtain support

For HID support:

- In the United States and Canada – send an email to HIDTechSupport@lifetech.com, or call 888-821-4443 option 1.
- Outside the United States and Canada – contact your local support office.

For the latest services and support information for all locations, go to:

www.lifetechnologies.com/support

At the web site, you can:

- Access worldwide telephone and fax numbers to contact Technical Support and Sales facilities.
- Search through frequently asked questions (FAQs).
- Submit a question directly to Technical Support.
- Search for user documents, Material Safety Data Sheets (MSDSs), vector maps and sequences, application notes, formulations, handbooks, certificates of analysis, citations, and other product support documents.
- Download .pdf documents.
- Obtain information about customer training.

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