DNA: Sample Analysis and Electronic Data

Data analysis

Data analysis is accomplished using GeneMapper® ID-X.

- Create a project in GeneMapper® ID-X.
- All projects should be analyzed using the following parameters:

	Identifiler Plus	Yfiler
Analysis Method	*Identifiler Plus Analysis Method ID-X*	*Yfiler Analysis Method*
Panel	*Identifiler_Plus_Panels_v1X-dup*	*Yfiler_v2*
Size Standard	*Identifiler Plus Size Standard*	*Yfiler Size Standard*
Peak Amplitude Threshold	50 RFU	75 RFU
Analysis Range	Full Range	Full Range

  - Identifiler® Plus and Yfiler® stutter percentage settings can be found in the *DNA: Profile Quality* section of the DNA Manual.
  - All other analysis settings can be found online or in Applied Biosystems GeneMapper® ID-X Software Guides and *User Bulletins*.
- All projects should include at least one allelic ladder, one positive control, one negative control, and all applicable case samples.
- All true off-ladder (OL) alleles and artifactual peaks should be edited electronically.
- Review electropherogram data for all formamide blanks.
  - Evaluate for artifacts, spikes, and/or injection abnormalities (e.g., carryover).
  - Electropherograms should be printed and included in the GM ID-X project when carryover is observed.
- Select one formamide blank sample to incorporate into the GM ID-X project.
- Use the Profile Comparison tool to evaluate the run.

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About the Profile Comparison tool

The Profile Comparison tool within GeneMapper® ID-X software is used to evaluate the run by automatically performing multiple quality checks. Profiles within a project are compared to one another and to designated controls. Profile Comparison results can be used to evaluate quality, detect possible contamination and sample-switch events, and identify sample associations by allowing the analyst to:

- evaluate reagent blanks, negative controls, and formamide blanks as a group
- compare samples in a project to one another using a defined match percent threshold to evaluate sample concordance and allele matching
- compare samples in a project to laboratory reference profiles
- evaluate concordance of all designated controls

Using the Profile Comparison tool

This is the procedure for using the Profile Comparison tool.

Step	Action
1	Edit allele labels as needed so that no OL (off ladder) allele labels are present in the project. Samples containing OL peaks are not considered in comparisons.
2	In the project window toolbar, select Tools and go to Profile Comparison.
3	Click the **Sample Concordance** tab. This tab displays groups of samples with 100% concordance. Use the mouse to hover over each group to expand the profiles listed and review the results. Evaluate the single-source group containing "blank" samples (reagent blanks, formamide blanks, and negative controls) for expected results. Evaluate the results of each additional Single Source or Mixed Source group.
4	Click the **Sample Comparison** tab and verify that the Percent Match Threshold is set to 80%. Click Compare Profiles and review the results of pairwise Single-Single, Single-Mixed, and Mixed-Mixed sample comparisons within the project.
5	Click the **Lab Reference** tab and verify that the Percent Match Threshold is set to 80%. Click Compare Profiles and review the results of comparisons between samples within the project and known laboratory reference samples.
6	If needed, the analyst may adjust the Percent Match Threshold to adjust the stringency and re-evaluate the results.

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Evaluating Profile Comparison results

To aid in the detection of possible contamination or sample-switch events, the analyst should identify and evaluate:

- blank samples observed in groups that contain detected alleles
- evidence samples expected to contain no DNA results or very partial DNA results (based on quantitation values) within groups that contain detected alleles
- samples from different or unrelated cases existing within the same group
- associations between samples (other than QC samples) and known laboratory reference samples

The analyst should use caution when evaluating the significance of profiles with high percentages of allele matches. The following are general considerations when evaluating Profile Comparison results:

- When partial profiles are compared to other single-source or mixture profiles, high percentage allele matches may be observed. In general, the fewer alleles in the partial profile, the higher the tendency to generate up to 100% allele matches to other profiles or to laboratory reference samples.
- As the number of contributors to a mixture profile increases, more alleles will be detected. Therefore, there may be a greater tendency to generate high percentage allele matches to other profiles within the project or to laboratory reference samples.
- The analyst may adjust the Percent Match Threshold, as necessary when performing Profile Comparison, to adjust the stringency of the comparison.
- When Profile Comparison results indicate that possible contamination or sample switching has occurred, the analyst should evaluate the electropherograms to aid in the assessment.

Electronic data access and security

Authorized access to electronic DNA quantitation files and run folder data on the 3130 Genetic Analyzers is limited to those who have pre-programmed access cards for the Biology Unit.

GeneMapper® ID-X data analysis software is password-protected and limited to DNA analysts and the Biology supervisor. All electronic data files associated with a DNA request are stored on a DVD as evidence.

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Electronic data storage

The DVD containing the electronic data is stored as evidence. The data should consist of

- any photographs taken during examination
- quantitation file(s)
- run folder(s)
- GeneMapper® ID-X project(s)