

MiSeq FGx Forensic Genomics System Software Release Notes

MiSeq FGx Control Software 1.1.0

ForenSeq Universal Analysis Software 1.1

For MiSeq FGx Forensic Genomics System Software

July 6, 2015

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Introduction

These release notes describe notes on the software and known issues of software components of the MiSeq FGx Forensic Genomics Software Package for the v1.1 update. Only changes from the v1.0 release are described in this section.

The detail level of the release notes is meant to describe the bug fixes and features added from a cursory level. Additional detail on the software is provided in the *ForenSeq Universal Analysis Software Guide (PN 15053876)*. The software package includes:

- MiSeq FGx Control Software version 1.1.0
- BaseSpace Broker version 2.4.0.29
- MiSeq Reporter version 2.5.1.3
- Real-Time Analysis (RTA) version 1.18.54
- Recipes version 3.0.10
- Genomes version 1.1.0
- ForenSeq Universal Analysis Software Service Broker 1.0.15063.0844

The ForenSeq Universal Analysis Software includes:

- ForenSeq Universal Analysis Software version 1.1.15139
 - Management Services 1.1.15139
 - Analysis Services 1.1.15139
 - Web Services 1.1.15117
 - Analysis Worker 1.0.15119

I. MiSeq FGx Control Software v1.1.0

NEW FEATURES:

- Titles have been added to the Sequencing page to group the metrics and clarify the read indicators are indicative of Q-scores.

KNOWN ISSUES:

- None

OPTIMIZATIONS:

- The support URL on the About screen has been updated to <http://support.illumina.com/sequencing.html>

II. ForenSeq Universal Analysis Software v1.1.15139

NEW FEATURES:

- The gender for a sample result can be re-assigned by the user.
- Isometric alleles (two or more typed alleles with the same length) is visually highlighted with a gray box around the typed allele names on the Sample Details page.
- The language throughout the application is updated from bio-geographical to biogeographical to represent current convention.
- The sample level reports have the loci ordered according to chromosomal position instead of alphabetically.
- Validation for the calculation of population statistics is based on the loci used in the calculation instead of all loci. For example, having an STR locus with more typed alleles than the locus copy number will not create an error for the calculation of population statistics against only SNP population groups.
- Negative amplification controls and reagent blanks are not assessed for a gender call and are assigned an inconclusive gender.
- Samples listed under the Negative Control Metrics section of the application are linked to the sample results by clicking on the sample name.
- Samples listed under the Positive Control Metrics section of the application are linked to the sample results by clicking on the sample name.
- rs201326893_Y152OCH displays as Y152OCH on the locus box with both names present on the Locus Detail view.
- rs312262906_N29insA displays as N29insA on the locus box with both names present on the Locus Detail view.
- Titles have been added to the Quality Metrics page to group the metrics and clarify the read indicators are indicative of Q scores.
- On the Sample Representation plot of the Quality Metrics page, each sample name or bar links to the Sample Details page for the respective sample.
- Population statistics calculations are updated to account for loci flagged to use the 2p rule. This caused updates in the following locations:
 - Loci called as a homozygote have a toggle to apply the 2p rule for the sample
 - The population group settings page has a configuration setting to specify which calculation to use in the application of the 2p rule (2p or 2p-p2)
- Updates to the analysis or sample states can only happen one state at a time. For example, a review state of Primary Review can't be updated to Review Complete without first updating to Technical Review.
- When defining a new locus threshold template, the user can no longer add (Default) to the end of the template name.

- Usernames can accept capital letters for authentication and account creation. All system usernames remain in lowercase characters.
- Run quality information was added to the dashboard view for runs.
 - A Q is visible with the run information and colored according to the run metrics against suggested specifications.
 - A bar was added to the run information to graphically depict the progress of the run (cycles completed).
 - Hovering over the Q will provide a tooltip with detailed information of the run metrics.
- The ability to view the analysis processing queue is added and is accessible from the header. Currently processing analyses, analyses that are queued, and the three most recently completed analyses are presented.
- Sample level reports (i.e. Sample Details, Sample Summary) can be created from the Project Landing Page.
 - Multiple sample reports can be selected for generation at the same time.
 - When a report is requested, but not yet completed, genotype updates are disallowed until the report is created.
- Sample and project names support international characters.
- The Sample Description is visible during run creation.
- An About screen is added to display the current versions of the system, web module, management module, analysis module, and installed analysis workflows.
- A maintenance function is added to permit using or not using specific loci in the calculation of population statistics.
- The iconography of the maintenance page is updated such that each function has a unique icon.
- Reporting of dates throughout the system is updated to be formatted consistently.
- The lengths of names and descriptions are updated throughout the system for consistent maximum lengths (i.e. 55 characters for names, 100 characters for descriptions).
- Interlocus balance is not assessed for negative amplification controls and reagent blanks.
- The displayed error message for a failed population statistics calculation is updated to be more descriptive.
- The displayed error message for a failed phenotype estimation calculation is updated to be more descriptive.
- Attempted activities after a session timeout does not complete the action, but redirect the user to the login screen to authenticate.
- The Locus Thresholds page in the maintenance section is now named the Analysis Values page.
- The header of the Define Content page is renamed. The header changed from Locus Panel to Define Content.

- The analysis module version is reported on the analysis details page.
- The analysis module version is reported on the Settings worksheet of sample level reports.

DEFECT REPAIRS:

- Alleles are now always reported with a comma delimiter instead of a slash.
- Allele ordering for the Positive Control Metrics section of the application is from smallest to largest.
- Allele ordering in all generated reports is from smallest to largest.
- Updating the genotype of a positive control or negative control sample now updates the coloring of the P or N without a refresh of the application.
- The ordering of samples in the application no longer changes when assigning samples to a different project.
- The Create Report button of the Project Landing Page is now only active when there are results to report.
- The analysis log is now moved inside of each analysis folder to prevent issues when the same run is being processed with separate analyses at the same time.
- The analysis state names are updated to Primary Review -> Technical Review -> Review Complete
- The sample state names are updated to Primary Review -> Technical Review -> Review Complete
- Updating a SNP locus to be called or uncalled now updates the scatter plot of intensity vs. length
- The displayed source attribution threshold for population statistics calculations now uses the user supplied value defined with the population group settings.
- The Completed date for a run is now displayed for all runs that have finished sequencing or are errored.
- The window for the definition of a new population group will not close if a click is received outside of the modal.
- The legend entry of the Sample Read Count Threshold for the Sample Representation plot of the Quality Metrics display correctly corresponds to the color of the plot.
- When creating a new analysis and balance was the only value adjusted, the override tag is now added to the analysis settings name for the completed analysis.
- The intra-locus balance value was updated to only accept values greater than 0% and less than 100%.
- The SNP locus box for the Common SNPs of the Phenotype Estimation page is re-organized to account for the long rsIDs.

- An issue was corrected where some alleles of a positive control sample were unable to be toggled between typed and untyped.
- The name for sample details reports includes the name Sample Details instead of Sample Genotype in the file name.
- The state of an analysis are now correct regardless of system or service shutdowns.
- The error message for an unrecognized username was updated to be more descriptive, instead of internal server error.
- The disabled color of the P, N, and Q letters in the task panel and on the analysis details page are the same color.
- The sort arrow for intensity of the locus details display renders correctly.
- The Single Source Sample indicator for STRs now has correct capitalization.
- % Total Intensity for SNPs is now reported to the tenths place.

KNOWN DEFECTS:

- None

FORENSIC GENOMICS ANALYSIS MODULE

- The analysis error as a result of a file concatenation step no longer prevents the completion of the analysis.