

BlueFuse Multi v4.5

Software Release Notes

Client v4.5.32178

December 2018

Introduction

These release notes detail the key changes to the BlueFuse Multi client software since v4.4. The main new features in this release are:

1. GRCh38 support for Illumina Bead Arrays
2. A number of usability enhancements to the Karyomapping module

In addition, there are a number of smaller features and bug fixes. To enable use of the GRCh38 Bead array support in this release, users should also import the latest annotation database. To download, go to <https://support.illumina.com/> and search for 'BlueFuse'.

For more information on BlueFuse Multi v4.5, see the BlueFuse Multi User Guide which is accessible from the Help menu within the user interface, and also available to download from Illumina Support. This guide has been updated with the new features available in v4.5.

Note: there is no update to the BlueFuse Server product required for this release; the existing v1.7 is compatible with BlueFuse Multi v4.5.

I. Karyomapping features

BlueFuse Multi v4.5 implements a number of usability improvements requested by Karyomapping users:

1. Display Informative SNPs

Previous versions of the software did not display informative parental SNPs in the Haploblock chart. These are now displayed next to the chromosome ideogram when a case has no embryos; this allows an analyst to visualize how informative SNPs are distributed across the region/s of interest. Previously only a count of the statistics was provided in the report.

2. Whole Genome Haplotype View

Previous versions of the software did not allow users to scroll across chromosome boundaries in the Haploblock chart. This functionality can now be enabled by checking the 'Whole Genome Haplotype View' option in the 'Tools | Configuration | Karyomapping | Chart Configuration' dialog.

Note: if this option is enabled, the 'Show SNPs in Haploblock View' option is not available.

3. Independent Flanking Region Sizes

Previous versions of the software only allowed equally sized flanking regions. In certain cases, analysts may want to set independently sized flanking regions (e.g., 2.0 Mb on the 3' end but 3.5 Mb on the 5' end). This can now be achieved in the Case wizard by deselecting the default flanking region size option.

II. New features for Cytogenetics users

1. GRCh38 Support for Bead Arrays

BlueFuse Multi v4.5 now allows Bead Arrays to be processed using a GRCh38 annotation database. However, users cannot mix GRCh37 and 38 experiments in the same database. To support customers who wish to migrate existing GRCh37 decision tracks, .bed file export functionality has been included to allow track data to be processed through utilities such as UCSC Lift Genome Annotations. Customers wishing to use this functionality are advised to download the GRCh38 database and review the Tech Note on using this feature.

The Disease track has been removed from the GRCh38 annotation database and this may result in differences to the automated region assessment for Bead Arrays when compared with the same experiment processed with a GRCh37 annotation database. A small percentage of previously classified 'Pathogenic?' regions may become 'Unknown?', and similarly a small percentage of previously classified 'Unknown?' regions may be classified as 'Benign?'

2. ISCN Report Update

Regions in the ISCN section of the report now use the ISCN 2016 notation.

III. New features for PGS users

There are no new features for PGS users in this release and PGS only customers can continue to use v4.4.31851.

IV. Other general features

1. Removal of the requirement to register a new BlueFuse Multi installation

Users are no longer required to submit a request for a registration key for each new BlueFuse Multi installation; the software is fully operational after a new install.

V. Notable defect repairs

- A substantial lag time which occurred when initiating Duo/Trio analysis has been reduced.
- Clarified error messages that appear when a user using BlueFuse Multi in Classic mode attempts to access a database where the BlueFuse Server already has write permission.
- Log R and BAF deviations, and the Call Rate I BeadArray QC measures were previously labeled as mean values They have been correctly relabeled as median values
- Occasionally significant variation in Bead Array LogR Dev metric has been observed after the rescanning of an array. BlueFuse Multi now removes probes with a zero raw intensity from this calculation and this issue should no longer occur.

VI. Known issues in this release

- After editing the algorithm settings for a bead array, an option to 're-run Bluefuse' appears in the Algorithm side panel
- Decision track ordering/display can be affected by the import of a new annotation database into an existing experiment database; the issue is exacerbated if tracks have been imported or customized. Users can work around this issue by creating a new database and importing the annotation database into this.

Upgrading to This Version

Users of v3.x BlueFuse Multi must upgrade to v4.4 prior to upgrading to this release. For assistance with upgrading to this version of BlueFuse Multi, please contact Technical Support techsupport@illumina.com