BlueFuse® Multi Annotation Database Release Notes

For use with BlueFuse Multi v4.4

BG_Annotation_Ens74_20180801.db

Introduction

The information included in the annotation database (DB) is used by BlueFuse Multi during analysis and visualization of experiments.

The annotation database is central to BlueFuse Multi software functionality. It contains information about the human genome to aid in the interpretation of experimental results. The annotation database includes the position of genes, disease regions, and publically available data on CNV frequency shown in the DecisionTrack pane within BlueFuse Multi. It uses information from major public databases such as ENSEMBL for gene annotation, OMIM for inherited diseases, and DGV and Decipher for genetic healthy or disease variants.

Annotation database files are available for download from http://www.cambridgebluegnome.com/account-login (login required). A Color Key explains all the information provided in the Decision Tracks and is available under the Help menu within BlueFuse Multi. The individual sources used are also described below. The release notes for the BlueFuse Multi v4.4 software are also available for download and include information about the new features in the software.

These release notes provide a summary of the updates incorporated into the latest annotation database which is listed on the web site as:

> For VeriSeq™ PGS and constitutional applications including Illumina BeadArrays (genome build 37): BG_Annotation_Ens74_20180801.db

An installation video guide for BlueFuse Multi that includes instructions for importing an annotation database is available here (http://www.screencast.com/t/QbNPBObV). If you have any questions regarding BlueFuse Multi, please contact techsupport@illumina.com.

Database md5 checksum: **28dbedce9028ca4a6baeabaca5fcb747**

To validate your downloaded file, you can optionally use third-party software such as WinMD5Free to confirm the above checksum.

Summary

This annotation database provides support for CytoSNP 850K v1.2 in BlueFuse Multi. Only CytoSNP 850K customers need to update to this annotation database. VeriSeq PGS, Karyomapping and HumanCytoSNP-12 customers do not need to update and should continue using their current annotation database.

Specific Changes

1. CytoSNP 850K v1.2: Support for this new version of the CytoSNP 850K has been added. Probe locations were generated using the B1 manifest file.
2. CytoSNP 850K v1.1: Probe content has been regenerated using the released, C1 version of the manifest file. Overall, 675 probes have been removed because they are either not present in the released manifest or because they have locations which are in conflict between arrays.
3. CytoSNP 850K v1.0: 20 probes have been removed where the locations were in conflict between arrays. 183 probes have been added where the locations are no longer in conflict between arrays.

4. HumanCytoSNP 12v2.1: 105 probes have been removed where the locations were in conflict between arrays. 8 probes have been added where the locations are no longer in conflict between arrays.
5. HumanCytoSNP 12v2.0: 105 probes have been removed where the locations were in conflict between arrays. 21 probes have been added where the locations are no longer in conflict between arrays.
6. HumanKaryomap 12v1: 105 probes have been removed where the locations were in conflict between arrays. 8 probes have been added where the locations are no longer in conflict between arrays.
7. The genome assembly GRCh37 is identical to the previous release.
8. All annotation tracks are identical to the previous release

Note: changes in probe content or locations may change results for experiments reprocessed using this annotation database.

Individual Data Sources

The following details the type and source of data used to create the separate annotation tracks.

Track Name	Track Description	Source	Version
Genes	The set of Ensembl genes which can be displayed in both compressed and expanded track styles. Includes gene names, types and OMIM and HGNC annotation, links to Ensembl genes, OMIM and HGNC.	Ensembl	74
Exons	Exons that are part of the canonical transcript of each Ensembl gene. Includes exon ids and gene names.	Ensembl	74
Disease	These are the regions associated with constitutional disorders which are specifically targeted by additional probes in CytoChip array designs	OMIM through Ensembl	01.2014
DGV Gain/Loss	CNV data compiled from multiple studies of normal populations	DGV	DGV2 2016-05-15 (GRCh 37)
DECIPHER Gain/Loss	Affected CNV data compiled by the Decipher project color coded by their classification.	DECIPHER consortium	12.2009 (transferred from NCBI36)
(Labeled) ISCA Gain/Loss	Annotated CNV regions involved in DD submitted by ISCA consortium members	ISCA and dbVAR	08.2012
ISCA Consensus Gain/Loss	Annotated consensus CNV regions involved in DD reviewed by the ISCA consortium	ISCA	08.2012