BlueFuse® Multi Annotation Database Release Notes

For use with BlueFuse Multi v4.5

BG_Annotation_Ens91_20220906.db

September 2022

Introduction

The information included in the annotation database 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, exons, and publicly available data on copy number variation (CNV) frequency shown in the DecisionTrack pane within BlueFuse Multi. It uses information from major public databases such as Ensembl for gene annotation, Online Mendelian Inheritance in Man (OMIM) for inherited diseases, Database of Genomic Variants (DGV) and dbVAR for genetic healthy or disease variants.

To download annotation database files, go to https://support.illumina.com/array/array_software/bluefuse-multi-software/downloads.html and select BlueFuse Multi Annotation Database. 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 described below. The release notes for the BlueFuse Multi v4.5 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:

BlueFuse Multi Annotation Database (build 38 for CytoSNP 850K v1.3)
(BG_Annotation_Ens91_20220906.db)

Please refer the BlueFuse Multi v4.5 User Guide for annotation database installation instructions. If you have any questions regarding BlueFuse Multi, please contact techsupport@illumina.com.

Database md5 checksum: 639d0cb56f740d07766ab9fdf32ae854

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

Changes for BG_Annotation_Ens91_20220906

This annotation database provides support for build 38 data generated from supported Infinium™ BeadChips when used with BlueFuse Multi v4.5 This annotation database is not compatible with BlueFuse Multi v4.4 and below. It supports CytoSNP 850K v1.2 and CytoSNP 850K v1.3 Infinium BeadChips. See Array Content for details of available probes for each array. Genotype call (GTC) files generated using the build 38 versions of the manifest files must be imported with an Ens91 annotation database. GTC files generated using the build 37 versions of the manifest files must be imported with an Ens74 annotation database. VeriSeq Preimplantation Genetic Screening (PGS) and Karyomapping customers should continue to use BlueFuse Multi

Annotation Database (BG_Annotation_Ens74_20160909.db). See Individual Data Sources for details of included tracks for build 38.

Array Content

The following table describes available probe content for each Infinium BeadChip supported in BG_Annotation_Ens91_20220906.

Array	Probe Content
CytoSNP 850K v1.2 iScan and NextSeq550	The annotation database contains 839,273 probes for this array. Probe content has been regenerated using the released, D2 version of the manifest file. 2,343 probes were removed due to invalid positions[1]. 4,110 probes were removed due to guanine-cytosine (GC) content conflicts between arrays.
CytoSNP 850K v1.3 iScan and NextSeq550	Support for CytoSNP 850K v1.3 has been added. Probe locations were generated using the B2 manifest file. The annotation database contains 842,559 probes for this array. 442 probes were removed due to invalid positions[1]. 4,139 probes were removed due to GC content conflicts between arrays.

[1] Invalid probe positions include probes on the mitochondrial chromosome or other non-autosomal, non-allosomal chromosomes. This also includes probes marked as homologous between X and Y which are outside of the pseudoautosomal (PAR1 and PAR2) regions.

Individual Data Sources

The following table details the tracks in the database and the data sources used to construct them. The build 38 genome assembly and all annotation tracks are identical to the previous release (BG_ Annotation_Ens91_20181915.db).

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	91
Exons	Exons that are part of the canonical transcript of each Ensembl gene. Includes exon ids and gene names.	Ensembl	91
DGV Gain/Loss	CNV data compiled from multiple studies of normal populations.	DGV	DGV Variants GRCh38 Release Date: 2016-05-15
dbVAR Gain/Loss	Structural variation data compiled from multiple studies and annotated with clinical significance. Includes data originally maintained by the ISCA consortium. The following studies are used to generate this track: estd232, estd228, nstd103, nstd102, nstd101, estd208, nstd93, nstd89, nstd83, nstd75, estd186, nstd59, nstd52, nstd51, nstd45, nstd42, nstd40, nstd37, nstd36, nstd33, nstd28.	dbVAR	Access date: 2018-01-25