BlueFuse® Multi Annotation Database Release Notes

For use with BlueFuse Multi v4.5

BG_Annotation_Ens74_20230922.db

October 2023

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, and 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 37 for CytoSNP 850K v1.4)
(BG_Annotation_Ens74_20230922.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: a0ac41a2121325abf3d35a9797ab8b9d

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

Changes for BG_Annotation_Ens74_20230922

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

Array Content

The following table describes available probe content for each Infinium BeadChip supported in BG_Annotation_Ens74_20230922.

Array	**Probe Content**
CytoSNP 850K v1.2 iScan and NextSeq550	The annotation database contains 842,908 probes for this array. 2,142 probes were removed due to invalid positions[1].
CytoSNP 850K v1.3 iScan and NextSeq550	The annotation database contains 846,411 probes for this array. 729 probes were removed due to invalid positions[1].
CytoSNP 850K v1.4 iScan and NextSeq550	Support for CytoSNP 850K v1.4 has been added. Probe locations were generated using the B1 manifest file. The annotation database contains 848,207 probes for this array. 695 probes were removed due to invalid positions[1].

[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 37 genome assembly and all annotation tracks are identical to the previous releases (BG_ Annotation_Ens74_20180801.db and BG_ Annotation_Ens74_20220906.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	74
Exons	Exons that are part of the canonical transcript of each Ensembl gene. Includes exon ids and gene names.	Ensembl	74
DGV Gain/Loss	CNV data compiled from multiple studies of normal populations.	DGV	DGV2 2016-05-15 (GRCh 37)
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