

BlueFuse[®] Multi Annotation Database Release Notes

For use with BlueFuse Multi v4.5

BG_Annotation_Ens91_20181015.db

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 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 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:

For Illumina BeadArrays (genome build 38): BG_Annotation_Ens91_20181015.db

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

Database md5 checksum: **f6109f5aba132f32ee0002d71af4fdb8**

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_20181015

This annotation database provides support for build 38 when used with BlueFuse Multi v4.5. This annotation database is only for use with build 38 data generated from Illumina BeadArrays. This annotation database is not compatible with BlueFuse Multi v4.4 and below.

This annotation database supports the CytoSNP-12 v2.1, CytoSNP-850K v1.1 and CytoSNP-850K v1.2. See Array Content for details of available probes for each array. 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 PGS and Karyomapping customers should continue to use their current annotation database.

The available tracks have changed for build 38. See Individual Data Sources for details of included tracks.

Array Content

The following table describes available probe content for each Illumina bead array supported in BG_Annotation_Ens91_20181015

Array	Probe content
CytoSNP-12 v2.1	269 probes were removed due to invalid positions ¹ . 1260 probes were removed due to GC content conflicts between arrays. The annotation database contains 293073 probes for this array.
CytoSNP-850K v1.1 iScan	2342 probes were removed due to invalid positions ¹ . 3975 probes were removed due to GC content conflicts between arrays. The annotation database contains 837073 probes for this array when scanned on an iScan.
CytoSNP-850K v1.1 NextSeq550	2335 probes were removed due to invalid positions ¹ . 3974 probes were removed due to GC content conflicts between arrays. The annotation database contains 837242 probes for this array when scanned on a NextSeq 550.
CytoSNP 850K v1.2 iScan and NextSeq550	2354 probes were removed due to invalid positions ¹ . 4081 probes were removed due to GC content conflicts between arrays. The annotation database contains 840065 probes for this array.

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 PAR1 and PAR2 regions.

Individual Data Sources

The following table details the tracks in the database and the data sources used to construct them.

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