# Informatic Updates for Whole-Genome Microarrays

Illumina has released new manifest files for all currently available human whole-genome genotyping microarrays. These new files provide several improvements requested by researchers, including:

- Updated strand and allele information
- Corrected annotation of PAR regions

# Which products have been updated?

The affected products include: the HumanOmniExpress, HumanOmni2.5-8, HumanOmni5, HumanOmniZhonghua, and HumanOmniExpress\_FFPE BeadChips.

#### Are updates available for the semi-custom chip manifests?

There are no available updates for the semi-custom chip manifests, but Illumina suggests the following resolution: Review the parent chip manifest (e.g. HumanOmni5 for HumanOmni5+ data) and correct any observed differences in the semi-custom data with a downstream analysis program, such as PLINK (pngu.mgh.harvard.edu/~purcell/plink).

#### How do I use these manifest revisions in laboratory information management systems (LIMS)?

In order to use a new manifest in LIMS, first load the manifest, associate it with the correct BeadChip in LIMS, and then select the correct cluster file. Note that the cluster file revision letter need not match the revision letter of the manifest.

#### How do I find which SNPs have been updated?

A list of updated markers for each BeadChip will be available for download with the product support files on Mylllumina (my.illumina.com).

# Do the changes impact genotypes?

Yes, markers that change strand designation will have changes to their genotypes; however, homozygous or heterozygous designations will not change. Similarly, altered PAR region designation will impact whether a genotype is considered AA or A- (for example).

# Do the changes impact CNV analysis?

Yes, changes to the PAR designation will affect CNV analysis of the X and Y chromosomes.

# What genome build do these new manifests use?

The new manifests use the GRCh37/hg19 assembly, which was released in February 2009 and can be visualized using the UCSC Genome Browser (www.genome.ucsc.edu/cgi-bin/hgGateway).

# Will other new files be available in addition to the updated product files?

Illumina will provide the following files for download from Mylllumina: .csv manifest, gene annotation files, .bed file, demo dataset, and MAF files. The final genotype report files will be available on the FTP site (www.illumina.com/forms/ftp.ilmn) in the Whole Genome Genotyping directory under Current Products (username: guest; password: illumina).

# Do I need a new version of GenomeStudio® to read these new manifest files?

While there is no required update, Illumina strongly recommends using GenomeStudio 2011.1 for all products.

#### How should I use these files if I am starting a new project?

Researchers should process samples according to standard protocol using the new product files.

### How should I use these files if I am in the middle of a project?

There are two options for this situation. Illumina recommends that all data be reloaded into GenomeStudio to re-call all genotypes and SNP metrics using the newest revisions of the product files. Alternatively, the project can be completed using the original files. The new map positions can be updated at a later time using a downstream analysis program.

#### How should I use these new manifests if I am finished with my project?

Completed data can be updated to the new manifests by updating the new chromosome and base pair positions in secondary analysis (i.e., map files).

#### How should I use these files if I am a LIMS customer?

Note that in LIMS, the product files (.bpm, .egt, and .xml) are bound to the product in Project Manager. AutoCall will use the current file in the LIMS database for all future \*.gtc file generation for a given BeadChip product. Therefore, researchers should only update Project Manager with the new product files when all projects containing the specific BeadChip product can be transitioned to the new versions. Data previously generated with the older versions should be re-queued for AutoCall.

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