2011 Informatic Updates for Whole-Genome Microarrays

April 2011 Release

Illumina has released new product files (*.bpm, *.egt, and *.xml) for all currently available human whole-genome genotyping microarrays. These new files provide a number of improvements, including:

- Updated SNP names to the most current rsID or 1000 Genomes ID, where available
- Corrections of errors identified in the October 2010 product file release

What has changed since the release of manifests in October 2010?

The April 2011 manifests include a number of improvements:

- Correction of positional information for CNV probes
- Conversion to the most current SNP name and positional information in cases where discrepancies existed within public databases

Will I need a new version of GenomeStudio® software to read these new manifest files?

GenomeStudio® v1.6 (version 2010.1) or higher is needed to read the new manifest files that include +/-strand information.

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

Researchers should process samples according to standard protocol using the new manifest, cluster, and product descriptor 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 genotypes and SNP metrics using the new files. Alternatively, the project can be continued and completed using the original files. New map positions can then be updated at a later time using a second-stage analysis program. Name and position conversion files (i.e., from the original Build 36 manifest files to the new April 2011 release), and lists of changed and removed loci are available for all currently available arrays upon request from Illumina Technical Support (TechSupport@illumina.com).

How should I use these files if I am finished with my project?

A researcher interested in updating their completed data to the new manifests can use the information in the new manifest files to update chromosome and base pair positions in secondary analysis files (i.e., map files). In addition, a list of changed or removed loci for each currently available whole-genome microarray is available upon request from Illumina Technical Support (TechSupport@illumina.com). Researchers should feel free to use their own professional judgment in accepting these changes, as updates to the reference genome are frequent.

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

Illumina strongly recommends converting to the newest version of these files as soon as possible in any stage of a project. As a reminder, in laboratory instrument management systems (LIMS), the product files are bound to the product that is accessioned into Project Manager. Also, Autocall will use the current file in the LIMS database for all future *.gtc file generation for that BeadChip product. Therefore, researchers should only update Project Manager with the new product files when they are confident that all projects containing the specific BeadChip product can be transitioned to the new versions, and that data previously produced with the older versions can be re-queued for Autocall.

Illumina, Inc. • 9885 Towne Centre Drive, San Diego, CA 92121 USA • 1.800.809.4566 toll-free • 1.858.202.4566 tel • techsupport@illumina.com • illumina.com

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