

Copy Number Variation (CNV) Analysis with BeadStudio v3.1

CNV Analysis is fully supported with BeadStudio software in combination with all Illumina Infinium® DNA Analysis BeadChips.

INTRODUCTION

Illumina provides many plug-ins designed specifically for copy number variant (CNV) analysis within BeadStudio v3.1. The following list describes several plug-ins and where they can be obtained. All of these applications require BeadStudio v3.1 and internet access. For the latest list of plug-ins, please visit the illumina•Connect website at <http://www.illumina.com/illuminaconnect>.

TABLE 1: CNV ANALYSIS OPTIONS

Plug-in	Function
cnvPartition	Annotate CNV regions and copy number estimate
dChip	Provide copy number estimates and LOH scores
Homozygosity Detector	Detect extended tracts of homozygosity
LOH Score	Provide LOH scores
ChromoZone	Annotate CNV regions

CNVPARTITION

Summary: Developed by Illumina, this algorithm uses a recursive partitioning approach and provides copy number estimates, as well as chromosomal bookmarks.

Availability: This plug-in is currently available for BeadStudio v3.1. It can be downloaded from the BeadStudio portal or from the illumina•Connect plug-ins webpage.

Use: This plug-in is run from the Genotyping Module.

Output: Unlike ChromoZone, cnvPartition is optimized to use intensity information, and thus provides both copy number estimates and bookmarks.

Performance: 25 seconds per sample¹ (Hap550).

DCHIP

Summary: dChip was developed at Harvard (Cheng Li laboratory) and has been used on various types of microarray data². It provides copy number estimates and LOH scores.

Availability: A compatible custom report plug-in is currently available for BeadStudio v3.1. This can be obtained from the illumina•Connect website, or the BeadStudio portal. The setup program will install the plug-in in the correct directory. In addition, dChip must be downloaded from <http://www.hsph.harvard.edu/biostats/complab/dchip/download.htm>.

Use: The custom report plug-in will provide the correct data file for input to dChip. The plug-in is run by selecting **dChipGTInput Reports** from the drop-down list of Custom Reports in **Analysis | Reports | Report Wizard**.

Output: dChip provides both copy number estimates and LOH scores.

Note: Illumina only provides a custom report plug-in that outputs BeadStudio data in a dChip compatible format. Illumina does not currently provide any further support of dChip.

HOMOZYGOSITY DETECTOR

Summary: Developed by Illumina, this algorithm searches for extended regions of homozygosity (LOH) and does not use intensity information.

Availability: This plug-in is currently available for BeadStudio v3.1. It can be downloaded from the BeadStudio portal or from the illumina•Connect plug-ins webpage.

Use: This plug-in is run from the Genotyping Module.

Output: The Homozygosity Detector provides bookmarks.

Performance: 12 seconds per sample¹ (Hap550).

LOH SCORE

Summary: Developed by Illumina, this algorithm calculates values that indicate the likelihood that LOH is present in a region of interest.

Availability: This plug-in is currently available for BeadStudio v3.1. It can be downloaded from the BeadStudio portal or from the illumina•Connect plug-ins webpage.

Use: This plug-in is run through the IGV in the Genotyping Module.

Output: The algorithm provides LOH scores.

Performance: 40 seconds per sample¹ (Hap550).

CHROMOZONE

Summary: Developed by Illumina and released in BeadStudio 3.0, ChromoZone is an auto-bookmarking algorithm for single samples based on the B allele frequency metric only.

Availability: ChromoZone is available as a plug-in to BeadStudio. Users can download this plug-in from a link in the BeadStudio portal, or directly from the illumina•Connect website.

Use: ChromoZone is run as a plug-in from the IGV through the BeadStudio Genotyping Module.

Output: ChromoZone provides bookmarks labeled het split and het sparse.

Performance: 22 seconds per sample¹ (Hap550).

Note: This algorithm does not use intensity information, and therefore does not provide copy number estimates.

PAIRED-SAMPLE BOOKMARKER

This algorithm will no longer be available in BeadStudio v3.1.

COPY NUMBER ESTIMATE

This algorithm will no longer be available in BeadStudio v3.1.

REFERENCES

- (1) Performance measured on an Intel Pentium computer 3.4GHz processor, 2GB RAM, running Windows XP SP2.
- (2) Lin M, Wei LJ, Sellers WR, Lieberfarb M, Wong WH et al. (2004) dChipSNP: significance curve and clustering of SNP-array-based loss-of-heterozygosity data. *Bioinformatics* 20: 1233-1240.

ADDITIONAL INFORMATION

Visit our website or contact us at the address below to learn more about Illumina DNA Analysis BeadChips and software solutions.

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