# Copy Number Variation (CNV) Analysis with BeadStudio

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. The following list describes several plug-ins and where they can be obtained. All of these applications require BeadStudio 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 |
| QuantiSNP | Provide copy number estimates with confidence scores |
| 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. 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 sample[1] (Hap550).

## QuantiSNP

**Summary:** Developed at Oxford University, QuantiSNP has been designed and optimized for Illumina SNP genotyping and CNV data. Based on a Hidden-Markov Model[2], it provides copy number estimates and bookmarks with confidence scores. QuantiSNP is optimal for finding small-sized CNV regions.

**Availability:** A plug-in is required to call this algorithm in BeadStudio. This plug-in is available for download from the illumina•Connect website or the BeadStudio portal. The QuantiSNP Windows executable is also required and can be obtained by contacting Dr. Ioannis Ragoussis at (ioannisr@well.ox.ac.uk) or from the Oxford University website at http://www.well.ox.ac.uk/QuantiSNP/.

**Use:** The BeadStudio plug-in outputs the correct data input for QuantiSNP. The plug-in is run by selecting QuantiSNP CNV Analysis from the drop-down list of CNV Analysis algorithms in Analysis | CNV Analysis. QuantiSNP results can then be input into BeadStudio in the form of a CNV Analysis. Bookmarks can also be created within the Illumina Genome Viewer (IGV) based on the CNV Analysis.

**Output:** QuantiSNP provides copy number estimates with confidence scores (p-values).

**Performance:** Nine minutes per sample using 10 optimization steps[1] (Hap550), 40–60 minutes for Human1M-Duo.

## dChip

**Summary:** dChip was developed at Harvard (Cheng Li laboratory) and has been used on various types of microarray data[3]. It provides copy number estimates and LOH scores.

**Availability:** A compatible custom report plug-in is currently available for BeadStudio. 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.

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 sample[1] (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. 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 sample[1] (Hap550).

## ChromoZone

**Summary:** Developed by Illumina and first released in BeadStudio v3.0, 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 is no longer be available in BeadStudio.

## Copy Number Estimate

This algorithm is no longer be available in BeadStudio.

## References


1. Performance measured on an Intel Pentium computer 3.4GHz processor, 2GB RAM, running Windows XP SP2.
2. Colella S, Yau C, Taylor JM, Mirza G, Butler H et al. (2007) QuantiSNP: an Objective Bayes Hidden-Markov Model to detect and accurately map copy number variation using SNP genotyping data. Nucleic Acids Res 35: 2013-2025.
3. 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.


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