

CNV Region Report Plug-in v2.0.3 for the GenomeStudio® Genotyping Module

1. General Information

The Illumina GenomeStudio CNV Region Report Plug-in is a software plug-in that works with Illumina's GenomeStudio Genotyping module, v1.0 and above. This plug-in allows you to create three separate reports:

- A **Standard Report** that lists each copy number variation (CNV) and loss of heterozygosity (LOH) region for each sample
- An **Allele-Specific Copy Number Report** that reports copy number informed genotypes such as A- and ABB
- A **PLINK CNV Input Report**, which creates input files for some of the CNV features of the PLINK GWAS and CNV analysis application (<http://pngu.mgh.harvard.edu/~purcell/plink/cnv.shtml>)

This document provides information about downloading, installing, and using the CNV Region Report plug-in with the GenomeStudio Genotyping Module.

2. Version History

Build	Date
v2.0.3, First Release	11/13/09

3. Downloading and Installing the CNV Region Report Plug-in

1. Download and install PLINK from the [Harvard/MGH web site](http://pngu.mgh.harvard.edu/~purcell/plink/).
2. Download the CNV Region Report plug-in from one of three places:
 - The GenomeStudio Portal, in GenomeStudio v2008.1 (Modules v1.0) and later
 - The Software Downloads section of [iCom](http://www.illumina.com)
 - The [illumina•connect](http://www.illumina.com/connect) web page

The setup program places the DLL and associated files into the directory C:\Program Files\Illumina\GenomeStudio\Modules\BSGT\ReportPlugins\CNVRegionReport
3. Run the setup program for the plug-in.
4. Follow the instructions in the installation prompts.

4. Using the CNV Region Report Plug-in

1. In the GenomeStudio Genotyping Module, open a genotyping project.
2. Select **Analysis | Reports | Report Wizard**.
The Report Wizard appears.
3. From the Custom Report dropdown list, choose **CNV Region Report**.

4. In the Options area, adjust the report input parameters (see Section 6 , Adjusting the Parameters).
5. Choose a response to all subsequent report prompts and click **Next**.
6. Click **Finish** to create the report.

A progress bar shows the report creation status. When the report has finished processing, several files are created. These files, and how to use them, are described below.

5. Detailed Usage Notes

1. All reports require that a CNV Analysis algorithm (such as cnvPartition) have already been run on the current project, the project has been saved, and the CNV related columns are populated in the Full Data table.
2. Standard CNV Report
 - a. This report outputs all CNV and LOH regions found in samples selected for the report.
 - b. The output filename is <projectname>_standard_cnv_report.txt.
3. Allele Specific Copy Number Report
 - a. This report estimates the allele specific copy number for each probe entry, e.g. A- or AAB. This is labeled as CN_GTYPE in the output file.
 - b. The CN_GTYPE column is calculated using the CNV Value, the B Allele Freq, the GTYPE, and the theoretical B Allele Frequency normal distributions for each copy number. These theoretical B Allele Frequencies are the same as those used in the cnvPartition algorithm, and described in the [DNA Copy Number Analysis Algorithms Technical Note](#).
 - c. The output filename is <projectname>_allele_specific_cnv_report.txt.
 - d. The output file data can be imported as sub-columns into the GenomeStudio Genotyping Module's Full Data table using the standard "Import columns into the table" toolbar button.
4. PLINK CNV Input Report
 - a. This report creates several output files which can be used to automate the usage of PLINK CNV analysis for the chosen CNV analysis.
 - b. After this report has run, you can run the PLINK CNV features by double-clicking a batch file.
 - c. You are free to edit the script file to customize the analysis done by PLINK.
 - d. Please refer to the PLINK website for a description of the various PLINK input and output files.
 - e. PLINK can provide sample group level information if this is provided in one of the following ways:
 - i. In the Samples table "Sample Properties" dialog, set all Unaffected (Normal) samples to a Sample Group with the name "Normal." Set all Affected samples to a different Sample Group, such as "Cancer." Set all samples with unknown phenotype to Sample Group = "Missing."
 - ii. Import a column named "Affection," which labels samples as 1 for normal, 2 for affected, or 0 for missing.
 - iii. Manually edit the <projectname>.fam file with the affected status.

- iv. Please see the PLINK documentation for more information about Affection status.
- v. The sample group information is used in the *.cnv.summary PLINK output file
- f. Three data files used by PLINK are included with the report installer, and can be customized:
 - cancer_genes.txt – this is an example of a “Gene Subset File”
 - exclude_list.txt – this is an example of a “Region Exclude File”
 - glist-hg18.txt – this is an example of a “Region Intersect File”

See the report output file *.script to observe how these files are used.
- g. All output files are created in a new directory with the name format PLINK_CNV_<timestamp>.
- h. The files created by this report include:
 - <projectname>.bat – batch file to run PLINK
 - <projectname>.sh – Linux shell script to run PLINK
 - <projectname>.script – a text config file for PLINK
 - <projectname>.cnv.map – a map file needed by PLINK
 - <projectname>.cnv – a cnv data file needed by PLINK
 - <projectname>.fam – a family / phenotype data file needed by PLINK

6. Adjusting the Parameters

Perform the following steps to adjust user-configurable parameters of the CNV Region Report plug-in.

1. In the GenomeStudio Genotyping Module, Go to **Analysis | Reports | Report Wizard**.
The Report Wizard appears.
2. From the Custom Report dropdown list, choose **CNV Region Report**.
3. In the options area of the CNV Region Report dialog, modify the parameters as needed.

Parameter Name	Description	Default Value	Mandatory
CNV Analysis File	The path to the CNV Analysis File (<name>.bin)	blank	Yes
Gene Subset File	The path to the PLINK gene subset file (--cnv-subset).	C:\Program Files\Illumina\GenomeStudio\Modules\BSGT\ReportPlugins\CNVRegionReport\PLINK_data\cancer_genes.txt	Yes
PLINK Path	The path to the PLINK executable.	C:\plink\plink-1.05-dos\plink.exe	Yes
Region Exclude File	The path to the PLINK exclude file (--cnv-exclude).	blank	Yes

Parameter Name	Description	Default Value	Mandatory
Region Intersect File	The path to the PLINK intersect file (--cnv-intersect).	C:\Program Files\Illumina\GenomeStudio\Modules\BSGT\ReportPlugins\CNVRegionReport\PLINK_data\glist-hg18.txt	Yes
Run Allele Specific CNV Report	Run the Allele Specific CNV Report.	False	Yes
Run PLINK CNV Input Report	Run the PLINK CNV Input Report	False	Yes
Run Standard CNV Report	Run the Standard CNV Report	True	Yes

7. Technical Support

Direct questions about installing and using the CNV Region Report Plug-in to Illumina Technical Support at techsupport@illumina.com, 1.800.809.4566 (toll-free), or +1.858.202.4566 (outside North America).

Questions about installing and using PLINK should be addressed to the authors of PLINK at <http://pngu.mgh.harvard.edu/~purcell/plink/index.shtml>. Illumina technical support cannot help with support questions related to installing or using PLINK.

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