Universal CNV Adapter Plug-in v2.0 for GenomeStudio™ Software

1. Introduction

The Universal CNV Adapter Plug-in is a software library that works with Illumina's GenomeStudio data analysis software, and with separately-installed, Illumina- or third-party-provided executable programs for CNV (copy number variation) analysis. The Universal CNV Adapter Plug-in allows you to use the GenomeStudio software CNV Analysis API to create input files for and read output files from a local copy of the external CNV analysis executable application.

2. Installing the Universal CNV Adapter Plug-in

Perform the following steps to install the Universal CNV Adapter Plug-in.

1. Obtain and install the third-party CNV analysis application.
2. Download the Universal CNV Adapter Plug-in setup program, SetupUniversalCNVAdapterPlugin_2.0.0.msi, from the illumina•connect web page.
3. Run the Universal CNV Adapter Plug-in setup program.
4. Adjust the configuration file (C:\Program Files\Illumina\GenomeStudio\CNVAlgorithm\UniversalCNVAdapter\UniversalCNVAdapterPlugin.dll.config) as required to interface with the third-party program. See the Section 4 of this document for additional details about the configuration file.

3. Running the Universal CNV Adapter Plug-in

Perform the following steps to run the Universal CNV Adapter Plug-in.

1. Open a genotyping project in GenomeStudio.
2. Run a CNV Analysis by selecting **Analysis** | **CNV Analysis** from the main menu.
3. In the Create New CNV Analysis dropdown menu, select the algorithm you want to use (e.g., QuantiSNP 1.0).
4. After running a CNV analysis in GenomeStudio, you can view the results three ways:
   - In the Full Data Table columns **CNV Value** and **CNV Confidence**
   - In the **CNV Region Display** visualization tool
   - In the IGV (Illumina Genome Viewer), by selecting **View** | **CNV Analysis as Bookmarks**

4. Configuring the Universal CNV Adapter Plug-in

Because the plug-in's communication with the executable program and processing of input and output data are controlled by a modifiable configuration file, the plug-in works with multiple third-party CNV analysis programs. The default config file is located at C:\Program Files\Illumina\GenomeStudio\CNVAlgorithm\UniversalCNVAdapter\UniversalCNVAdapterPlugin.dll.config. The table below describes each of the configuration settings.

You can modify or overwrite the default config file to work with different executable programs. Only CNV analysis programs that process a single sample at a time are supported. Also, only programs that work with the input and output data specified below are supported.

Parameter Name	Description	Default Value	Mandatory
AlgorithmAuthor	The author of the external algorithm	<blank>	Yes
AlgorithmName	The name of the external algorithm	<blank>	Yes
AlgorithmVersion	The version of the external algorithm	<blank>	Yes
CommandLine Params	If provided, add these parameters to the command line. Text in brackets is treated as follows: [outputfile] – replace with name of output file, which is sampleid+Input FilesExtension+OutputFileExtension+ OutputFilesAdditionalExtension [sampleid] – replace with the sample id [gender] – replace with sample gender. If gender is not male or female, default to female. [inputfile] – replace with the name of the input file, sampleid+InputFilesExtension	[outputfile], [sampleid], [gender],10,2000000, 4, 1, 0, 0, gc_data_dir, [inputfile], 300	Depends on the program
DeleteTempFiles	Delete temporary files. Temporary files include the input and output files.	True	Yes
ExePath	Executable program path	<blank>	Yes
InputFileColumn Names	The comma-delimited list of input file column names. These are defined by the CNV application called by GenomeStudio.	Name, Chromosome, Position, Log R Ratio, B Allele Frequency	Yes
InputFileColumns	The comma-delimited list of input file column values. These follow a controlled vocabulary. Possible choices are: • SNPName • Chromosome • Position • LogRRatio • BAlleleFreq • XRaw • YRaw • XNorm • YNorm • Call	SNPName, Chromosome, Position, LogRRatio, BAlleleFreq	Yes

Parameter Name	Description	Default Value	Mandatory
InputFiles Extension	If provided, the extension for input files.	*.txt	No
OutputFile Columns	The comma-delimited list of output file column values. These follow a controlled vocabulary. Unused columns in the output file should be accounted for by single commas. Possible choices are: • SampleID • Start • Stop • Value • Confidence	SampleID, Chromosome, Start, Stop, Value, Confidence	Yes
OutputFile HeaderSkipLines	The number of output file header lines to skip when parsing the output data.	1	Yes
OutputFiles	If provided, store input and output files in this directory. If left blank, the following directory is used: C:\Documents and Settings\ <username>\Local Settings\ Application Data\ Illumina\Universal CNVAdapter.	<blank>	No
OutputFiles Additional Extension	If provided, this is the additional extension the CNV program appends to the output files before the OutputFiles Extension.	_output	Depends on the program
OutputFiles Extension	If provided, final extension for output files.	.txt	Depends on the program
RedirectOutput	Redirect output to the log file.	False	Yes
ScoreThreshold	CNV regions with a score below the threshold are ignored.	50.0	Yes

5. Updates in v2.0

Updated Universal CNV Adapter Plug-in for compatibility with Illumina's GenomeStudio software

6. Contacting Technical Support

Direct questions about installing and using the Universal CNV Adapter Plug-in to Illumina Technical Support at techsupport@illumina.com or 1-800-809-4566.

Please note that Illumina cannot answer questions about particular algorithms, such as how an algorithm works or how to choose input parameters. Direct questions about third-party CNV analysis algorithms to the technical support group of the organization that provided the algorithm.