Haploview Input Report Plug-in v2.0.0 for the GenomeStudio™ Genotyping Module

1. General Information

The Illumina GenomeStudio Haploview Input Report Plug-in is a software plug-in that works with Illumina's GenomeStudio Genotyping Module. This plug-in allows you to create input files for the Haploview application developed by the Broad Institute (http://www.broad.mit.edu/mpg/haploview).

This document provides information about downloading, installing, and using the Haploview Input Report plug-in.

2. Version History

Build	Date
v2.0.0, Initial Release	03/17/09

3. Downloading and Installing the Haploview Input Report Plug-in

1. Download and install Haploview from the Broad web site.

2. Download the Haploview Input Report plug-in from one of three places:

   - The GenomeStudio Portal

   - The software downloads section of iCom

   - The illumina•connect web page

   The setup program places the DLL into the directory C:\Program Files\Illumina\GenomeStudio\Modules\BSGT\ReportPlugins\HaploviewInputReport.

3. Run the setup program for the plug-in.

4. Follow the instructions in the installation prompts.

4. Using the Haploview Input Report Plug-in

1. Open a genotyping project in GenomeStudio.

2. Select **Analysis** | **Reports** | **Report Wizard**.

   The Report Wizard appears.

3. From the Custom Report dropdown list, choose **Haploview Input Report**.

4. In the Options area, adjust the report input parameters (see section 5, 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.

7. When the report has finished processing, two files are created:

    <projectname>.ped, the LINKAGE format input file

    <projectname>.info, the info file

    Note: If the files are too large to open in your default Windows text viewer (e.g., NotePad), you can open them using WordPad.

    The Haploview executable is launched using the two files from the previous step as inputs.

5.   Adjusting the Parameters

Perform the following steps to adjust the user-configurable parameters of the Haploview Input 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 **Haploview Input Report**.

3. In the options area of the Haploview Input Report dialog, modify the parameters as needed.

Parameter Name	Description	Default Value	Mandatory?
CommandLine Parameters	If provided, add these parameters to the command line	-pedfile,[pedfile],-info,[infofile]	Yes
ExePath	Executable program path	C:\Program Files\HaploView\ Haploview.exe	Yes
Chromosome	The chromosome for the region of interest	<blank>	Yes
ChrPosition	The chromosomal position at the center of the region of interest	<blank>	Yes
PedigreeFile	[Optional] The path to the pedigree file	<blank>	No
ProbeName	The name of the probe at the center of the region of interest	<blank>	Yes
RegionWidthInKb	The width of the region of interest (in kilobases)	200	Yes

Note: Either Chromosome and ChrPosition **or** ProbeName should be specified. If both are provided, ProbeName will be used as the center of region created for Haploview.

The parameters ChrPosition and ProbeName maintain a memory of the 5 most recently used input values. The PedigreeFile parameter maintains a memory of only the single most recent pedigree file used. The memory feature is accessible by clicking the value text field for these parameters.

Pedigree File Format, Tab-Delimited

Column Name	Description	Mandatory?
Family ID	Numeric ID representing the family ID	Yes
Sample ID	Sample ID used in the genotyping project	Yes
Father ID	Sample ID for the father; 0 for no father sample available	Yes
Mother ID	Sample ID for the mother; 0 for no mother sample available	Yes
Affected Status	0 = Unknown 1 = Unaffected 2 = Affected	Yes
Additional Columns	Ignored	No

Note: Gender is determined from the Genotyping module project, and thus is not needed in the pedigree file.

6.   Technical Support

Direct questions about installing and using the Haploview Input 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).

Address questions about installing and using Haploview to the authors of Haploview at mailto:haploview@broad.mit.edu.