

Using Partek's Report Plug-in for Illumina's GenomeStudio to Export Copy Number Analysis Data

This tutorial describes how to export copy number analysis data using Partek's Report Plug-in for the Illumina® GenomeStudio Genotype Module for use in Partek® Genomic Suite™.

Using the GenomeStudio plug-in allows you to export your data into a project that Partek can open directly. It is the fastest and most consistent way to get fully annotated Illumina genotype and copy number data into Partek.

If you don't have access to GenomeStudio, you can import a Final Report from GenomeStudio. In the main menu, select **File > Import > Illumina Text Data**. This method will not create or link annotations.

Installing the Plug-in

- Download the plug-in from the Illumina's GenomeStudio Portal
- Extract the **PartekReport.Genotype.dll** file from the distributed zip file
- Move the **DLL** file into the Genotype Module's report plug-in folder (Create the folder if it does not exist)
C:\Program Files\Illumina\GenomeStudio\Modules\BSGT\ReportPlugins\PartekReport (Figure 1)
- If you have old versions of the plug-in, please remove them from the GenomeStudio folder hierarchy

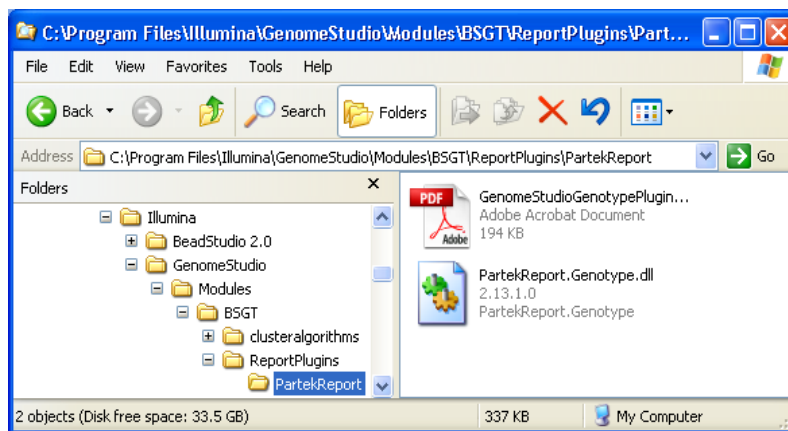


Figure 1: Placing the Partek Report DLL in the appropriate directory

Exporting from GenomeStudio

- Select **Analysis > Reports > Report Wizard** from the main menu
- Select **Custom Report**

- Choose **Partek Report Plug-in** from the list of custom plug-ins (Figure 2)
- Configure export options base on information below

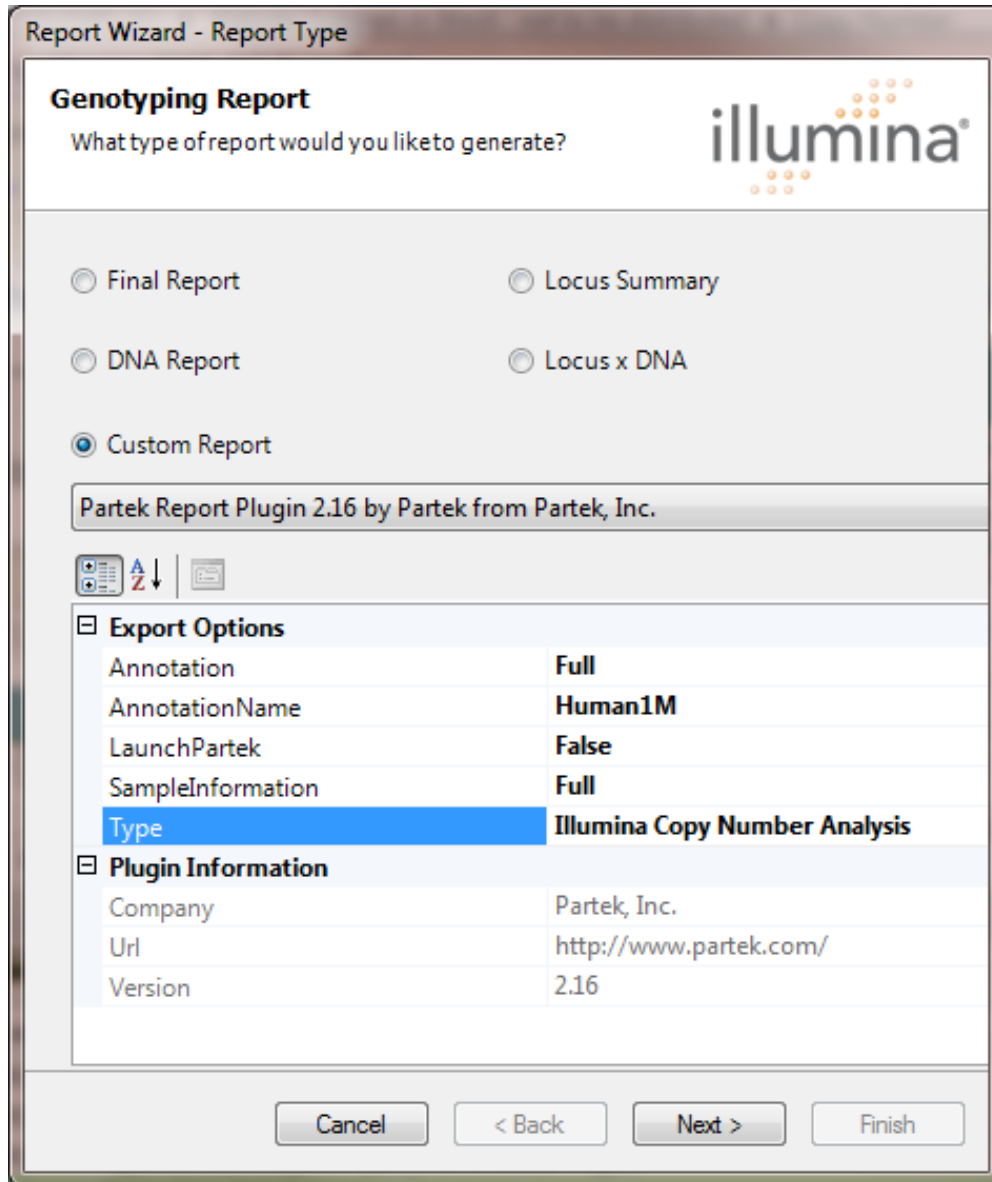


Figure 2: Configuring the Report Wizard

Export Options

Annotation	The level of detail you would like your exported annotation to have. Minimal will contain the required information to do data analysis, and Full will contain everything from GenomeStudio.
AnnotationName	This should be equivalent to the name of the bpm file you imported the data with; usually it is the same as the assay. Example values are Human1M or HumanHap550Duo. If you don't know this information, you should just use something unique to your data set.
LaunchPartek	After the plug-in has completed exporting, you can launch your data in Partek afterwards. If Partek is not already running it will be started. The exported data is then loaded and the appropriate Genotype workflow items are checked off.
SampleInformation	The level of detail you would like your exported sample information to have. If you have entered data from your experiment into the samples table, you should select Full.

Table 1: Export options for the genotyping report

There will be three result spreadsheets output in the project containing genotype calls, log R ratio which is copy number in log scale, and B Allele Frequency.

LogR ratio spreadsheet will be used for copy number analysis on copy number workflow, genotype spreadsheet will be used for genotype analysis like LOH, association workflow.

Illumina Copy Number Analysis <i>Use Illumina's Log R Ratio for copy number estimation.</i>	Genotype Log R Ratio B Allele Frequency
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Table 2: Output types

Advanced Options

These are not necessary for Partek workflows. For more information about these values, please consult GenomeStudio documentation.

X & Y	X & Y
Theta & R	Theta R
X Raw & Y Raw	X Raw & Y Raw
Theta Raw & R Raw	Theta Raw R Raw

Table 3: Advanced options

- On the next panel, *Browse* to the final file location you would like. It is **strongly recommended** to put the exported files in their own folder. This allows you to move the folder instead of all the files individually.
- Select **Finish** to start the export process

The result of the export process is a project file (.ppj) which you can open directly in Partek. The project file will open the data files and associate the annotation information to those data files. Annotation information describes the SNP markers which you've just created data for. The plug-in also creates a manifest file describing the files and date of creation.

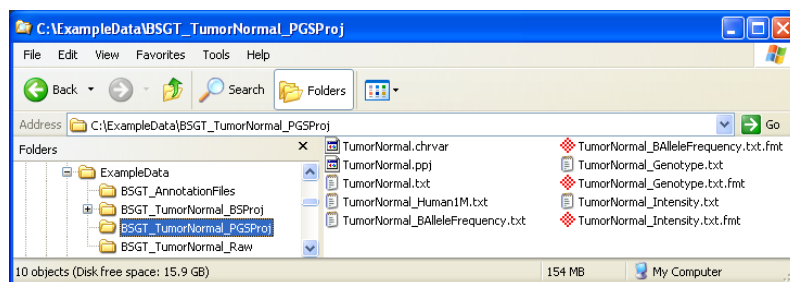


Figure 3: Exported Partek Copy Number Analysis Files from the GenomeStudio project

Opening the Project in Partek® Genomic Suite™

If you selected the **LaunchPartek** option on the report dialog (Figure 2), this step has already been completed.

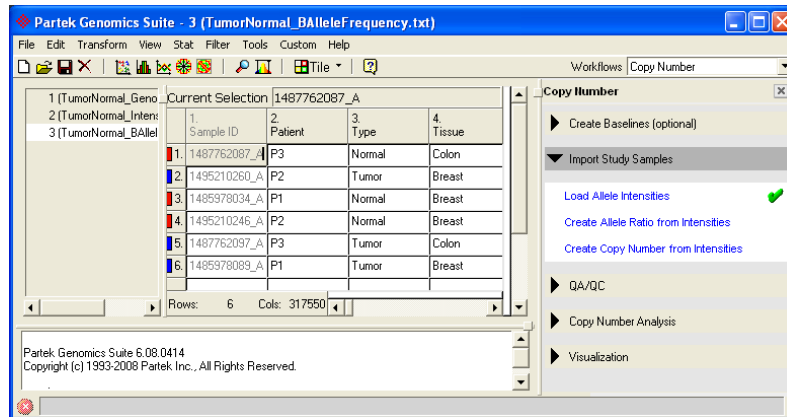


Figure 4: New project loaded in Partek

- Open the *Copy Number* workflow in the upper right hand corner of Partek (Figure 4)
- In the *Import* section, select **Import Samples**
- Select **Load Project** as the source of the data
- The data spreadsheets will be loaded

After loading the project, you can proceed with the **QA/QC** section.

Frequently Asked Questions

Q: Why doesn't the Partek plug-in appear in my list of custom plug-ins?

A: You have placed the DLL file in the incorrect location.

GenomeStudio opens pop-up dialogs if a plug-in is faulty, so if you receive no errors then it just did not find the file.

Q: After I open my project in Partek, my annotations are missing or the annotations are incorrect. Is the annotation corrupted?

A: If you use the same name as a previously created annotation, Partek will not overwrite existing annotation references. You will need to manually change the annotation links in your file, or update the File Manager so all files use your new annotation. For more information, please see the **Associating a Spreadsheet with an Annotation File** document on our tutorials website.

End of Tutorial

This is the end of Illumina data import tutorial. If you need additional assistance with this data set, you can call our technical support staff at +1-314-878-2329 or email support@partek.com.