



cnvPartition CNV Analysis Plug-in v1.0.2 for BeadStudio

1. General Information

The cnvPartition CNV Analysis Plug-in is a software library that works with Illumina's BeadStudio data analysis software. BeadStudio v3.1 or later is required in order to use this plug-in to analyze Illumina array data for the presence of CNVs (copy number variations).

This document provides information about downloading, installing, and using the cnvPartition CNV Analysis Plug-in. For information about how to perform CNV analysis, see [DNA Copy Number Analysis in BeadStudio v3.1](#), #11282031, or the [BeadStudio Genotyping Module v3.2 User Guide](#), #11284301 (Chapter 7, Performing LOH and Copy Number Analysis).

2. Version History

Build	Date
v1.0.2	03/11/08
v1.0.1	01/29/08
v0.9.3	09/04/07
v0.9.2	07/23/07

3. Updates in v1.0.2

- Confidence Threshold parameter—any CNV region with a confidence lower than this value will be removed from the CNV output. The recommended default is 35.
- Probe Gap Size Threshold parameter—a CNV region overlapping with any probe gap larger than this value will not be created. This helps prevent CNV regions from being found across centromeres and other large probe gaps. The recommended default is 1,000,000 base pairs.
- The internal expected Log R Ratio mean and standard deviation values for each copy number value have been changed to better reflect actual Log R Ratio values seen in normal HapMap samples. This results in higher overall accuracy and precision.


4. Downloading and Installing the cnvPartition CNV Analysis Plug-in

1. Download the cnvPartition CNV Analysis Plug-in from one of three places:
 - The BeadStudio Portal, in BeadStudio v3.1 and later
 - The software downloads section of [iCom](#)
 - The [illumina•connect](#) web page

The setup program places the DLL into the directory C:\Program Files\Illumina\BeadStudio 2.0\CNVAlgorithm\cnvPartition.

2. Run the setup program for the plug-in.
3. Follow the instructions in the installation prompts.

5. Using the cnvPartition CNV Analysis Plug-in

1. Open a genotyping project in BeadStudio.
2. Select **Analysis | CNV Analysis**.
The CNV Analysis dialog appears.
3. Select **Create New CNV Analysis | cnvPartition v1.0.2**.
4. In the CNV Analysis Name area, enter a name for this CNV analysis.
5. [\[Optional\]](#) In the Options area, adjust the parameters (see Section 6, Adjusting the Parameters).
6. Click **Calculate New CNV Analysis**.
7. When the analysis is complete, click **OK**.
8. After running the analysis in BeadStudio, you can view the results in many ways:
 - a. In the **Full Data Table**, in the CNV Value and CNV Confidence columns
 Use the column chooser  to display the CNV Value and CNV Confidence columns.
 - b. In the **CNV Region Display** visualization tool
 - c. In the **IGV (Illumina Genome Viewer)** visualization tool
 - d. In the **Bookmark Viewer**

6. Adjusting the Parameters

Perform the following steps to adjust the user-configurable parameters of the cnvPartition CNV Analysis Plug-in.

1. In a BeadStudio genotyping project, Go to **Analysis | CNV Analysis**.
The CNV Analysis dialog appears.
2. In the Options area of the CNV Analysis dialog, modify the parameters as needed.

Parameter Name	Description	Default Value	Mandatory
Confidence Threshold	CNV regions with a confidence below this level are removed from the analysis results.	35	Yes
Include Mitochondrial Chromosomes	Set this flag to True if you want to include mitochondrial chromosomes in this analysis.	False	Yes
Include Sex Chromosomes	Set this flag to True if you want to include X, Y, and XY (pseudoautosomal) chromosomes in this analysis.	False	Yes



Parameter Name	Description	Default Value	Mandatory
Probe Gap Size Threshold	Regions within probe gaps whose size is greater than this number are not considered to be within CNV regions. This helps prevent CNVs from being called across large probe gaps, such as centromeres.	1,000,000 base pairs	Yes

7. Technical Support

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

© 2008 Illumina, Inc. All rights reserved.

Illumina, Solexa, Making Sense Out of Life, Oligator, Sentrix, GoldenGate, DASL, BeadArray, Array of Arrays, Infinium, BeadXpress, VeraCode, IntelliHyb, iSelect, and CPro are registered trademarks or trademarks of Illumina. All other brands and names contained herein are the property of their respective owners.

The cnvPartition CNV Analysis Plug-in v1.0.2 incorporates portions of code from the Cephes Math Library.

Cephes Math Library Release 2.8: June, 2000 Copyright 1984, 1987, 1995, 2000 by Stephen L. Moshier

Contributors: * Sergey Bochkanov (ALGLIB project). Translation from C to pseudocode.

See subroutines comments for additional copyrights.

Redistribution and use in source and binary forms, with or without modification, are permitted provided that the following conditions are met:

-Redistributions of source code must retain the above copyright notice, this list of conditions and the following disclaimer.

-Redistributions in binary form must reproduce the above copyright notice, this list of conditions and the following disclaimer listed in this license in the documentation and/or other materials provided with the distribution.

-Neither the name of the copyright holders nor the names of its contributors may be used to endorse or promote products derived from this software without specific prior written permission.

THIS SOFTWARE IS PROVIDED BY THE COPYRIGHT HOLDERS AND CONTRIBUTORS "AS IS" AND ANY EXPRESS OR IMPLIED WARRANTIES, INCLUDING, BUT NOT LIMITED TO, THE IMPLIED WARRANTIES OF MERCHANTABILITY AND FITNESS FOR A PARTICULAR PURPOSE ARE DISCLAIMED. IN NO EVENT SHALL THE COPYRIGHT OWNER OR CONTRIBUTORS BE LIABLE FOR ANY DIRECT, INDIRECT, INCIDENTAL, SPECIAL, EXEMPLARY, OR CONSEQUENTIAL DAMAGES (INCLUDING, BUT NOT LIMITED TO, PROCUREMENT OF SUBSTITUTE GOODS OR SERVICES; LOSS OF USE, DATA, OR PROFITS; OR BUSINESS INTERRUPTION) HOWEVER CAUSED AND ON ANY THEORY OF LIABILITY, WHETHER IN CONTRACT, STRICT LIABILITY, OR TORT (INCLUDING NEGLIGENCE OR OTHERWISE) ARISING IN ANY WAY OUT OF THE USE OF THIS SOFTWARE, EVEN IF ADVISED OF THE POSSIBILITY OF SUCH DAMAGE.