Copy Number Analysis

This tutorial will illustrate:

- Importing Copy Number Data
- Exploring the data with PCA
- Creating Copy Number from Allele Intensities
- Detecting regions with copy number variation
- · Creating a list of regions
- Finding genes with copy number variation
- Optional: Additional options for annotating regions
- Optional: GC wave correction for Affymetrix CEL files
- Optional: Integrating copy number with LOH and AsCN

Note: the workflow described below is enabled in Partek Genomics Suite version 7.0 software. Please fill out the form on Our support page to request this version or use the Help > Check for Updates command to check whether you have the latest released version. The screenshots shown within this tutorial may vary across platforms and across different versions of Partek Genomics Suite.

Introduction to Copy Number Analysis

Copy number analysis asks whether there are regions of the genome with altered abundance. Of particular interest are any genes within those regions and how might a change in gene abundance alter phenotype. Partek Genomics Suite software allows these questions to be answered by analyzing a variety of commercially available assays for copy number analysis. SNP-genotyping arrays with closely spaced genomic markers (Affymetrix and Illumina) and comparative genomic hybridization (CGH) arrays (Agilent, NimbleGen, or custom spotted arrays) can be imported into Partek Genomics Suite and analyzed.

When performing copy number analysis, it is important to remember an inherent limitation of copy number region analysis - the inability to detect copyneutral events caused by copy-number-neutral loss of heterozygosity (LOH) or copy-number-neutral allelic imbalance. This limitation can be addressed by supplementing copy number analysis with SNP genotyping data. Partek Genomics Suite supports both LOH and allele-specific copy number (AsCN) analysis with dedicated workflows. Tutorials on LOH and AsCN analysis are also available.

Introduction to the tutorial data set

The example data set consists of 20 paired samples from an ovarian cancer study in which a fresh-frozen tumor sample and peripheral blood sample were obtained from 10 female patients (Ramakrishna et al. 2010). All 20 samples were analyzed using the Affymetrix Genome Wide Human SNP Array 6.0. To download the data set, select this link - CNV Tutorial Data Set. The data set is also used for the LOH and AsCN tutorials. The spreadsheet used in this tutorial was generated by importing SNP6 CEL files and annotating them with attributes for each sample. The experimental goal is to identify copy number changes present in multiple patient tumors.

References

Ramakrishna M, Williams LH, Boyle SE, Bearfoot JL et al. Identification of candidate growth promoting genes in ovarian cancer through integrated copy number and expression analysis. PLoS One 2010;5(4).

Additional Assistance

If you need additional assistance, please visit our support page to submit a help ticket or find phone numbers for regional support.

