Differential Methylation Analysis

Illumina's MethylationEPIC array interrogates the methylation status of over 850,000 cytosines in the human genome. Because the MethylationEPIC array is closely related to the Infinium HumanMethylation450 BeadChip, the steps presented in this document can be applied to either platform.

This tutorial illustrates how to:

- Import and normalize methylation data
- Annotate samples
- Perform data quality analysis and quality control
- · Detect differentially methylated loci
- Create a marker list
- · Filter loci with the interactive filter
- · Obtain methylation signatures
- Visualize methylation at each locus
- Perform gene set and pathway analysis
- Detect differentially methylated CpG islands
- Optional: Add UCSC CpG island annotations
- Optional: Use MethylationEPIC for CNV analysis
- Optional: Import a Partek Project from Genome Studio

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.

Description of the Data Set

The data set accompanying this document consists of sixteen human samples processed by Illumina MethylationEPIC arrays. The data set is taken from a study of DNA methylation in human B cells and B cells infected with Epstein-Barr virus (EBV).

Infecting B cells with EBV *in vitro* transforms them, making them capable of indefinite growth *in vitro*. These immortalized cell lines are referred to as lymphoblastoid cell lines (LCLs). LCLs behave similarly to activated B cells, making them useful for expanding T cells *in vitro*. Because EBV is a carcinogen and immortalized cell growth is a hallmark of cancer, examining the effects of EBV transformation on B cell DNA methylation might shed light on the roles of DNA methylation in tumor development.

The data files can be downloaded from Gene Expression Omnibus using accession number GSE93373 or by selecting this link - Differential Methylation Analysis data set. To follow this tutorial, download the 32 .idat files (note that two .idat files are generated for each array) and unzip them on your local computer using 7-zip, WinRAR, or a similar program.

Additional Assistance

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

