

# 6.881 Lab 7

## Intermediate Phenotypes and eQTLs

April 5, 2016

### Part 1

Run the PEER method on the datasets presented (expression.csv and genotype.csv) as described in Stegle et. al. [1] to generate Fig 2b. in the paper.

### Part 2

In step 3 of the PEER method, the maximum number unobserved factors to model was required as an input parameter. How does this choice affect identifying the PEER factors?

### Part 3

- Simulate eQTLs (could be done by eqtlbma <https://github.com/timflutre/eqtlbma/wiki>):

You could use the rnorm function of R to do simulation

Or here is some example script by using eqtlbma to simulate eQTL (you could change the option to reduce the time cost, run Rscript eqtlbma/eqtlbma-1.3.1-deploy/bin/tutorial\_eqtlbma.R to check their help information and options):

```
Rscript eqtlbma/eqtlbma-1.3.1-deploy/bin/tutorial_eqtlbma.R -pkg eqtlbma/eqtlbma-1.3.1/
-nsbgrps 1 -ninds 100 -ngenes 100 -nchrs 22 -asg 10 -anchor TSS -cr5 10000 -cr3 10000 -
ncores 1
```

- Rerun PEER on these simulated genotype and expression matrices

- call eQTLs before and after PEER correction (eQTL calling: MatrixEQTL [http://www.bios.unc.edu/research/genomic\\_software/Matrix\\_eQTL/](http://www.bios.unc.edu/research/genomic_software/Matrix_eQTL/) or your own linear function).
- Check the correlation (box plot) for the set  $s$  of SNP-gene pairs before and after applying PEER. Using these plots to measure the effect of correct factor number or other settings in PEER for the eQTL calling.

## 1 References

[1] Stegle, O., Parts, L., Piipari, M., Winn, J., & Durbin, R. (2012). Using probabilistic estimation of expression residuals (PEER) to obtain increased power and interpretability of gene expression analyses. *Nature Protocols*, 7(3), 5007. <http://doi.org/10.1038/nprot.2011.457>

MIT OpenCourseWare  
<https://ocw.mit.edu>

6.881 Computational Personal Genomics: Making Sense of Complete Genomes  
Spring 2016

For information about citing these materials or our Terms of Use, visit: <https://ocw.mit.edu/terms>.