

Detection of response QTLs with Full Length BRB-seq

Charles Solomon

Introduction

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Materials and methods

Stratified eQTL mapping for Naive and TNFa treated HUVEC

For eQTL mapping, gene expression counts were quantified with featureCounts (Liao, Smyth, and Shi 2019) from reads mapped with STAR v2.7.11a (Dobin et al. 2013). Genes with read count < 10 in more than 5% of the samples were filtered out. This left 11,723 genes as input for eQTL mapping. Sample library sizes were normalized with the “TMM” method from edgeR (Robinson, McCarthy, and Smyth 2010). The samples were then split into Naive and TNFa treated groups and normalized separately by applying inverse normal transformation across the CPM values for each gene.

For sQTL mapping, —————

Genetic variants were sourced from our previous study (Solomon et al. 2022). Briefly, PLINK v2.00a4.3 (Chang et al. 2015) was used to filter out variants that had missing call rates above 1%, Hardy–Weinberg equilibrium (HWE) test P-value $< 1e^{-6}$ and minor allele frequency (MAF) $< 25\%$. The choice of 25% MAF was informed by a previous detailed study to avoid inflated false positives due to our modest sample sizes (Huang et al. 2018). PCA was performed

on the genotype data with PLINK2 and three genotype (PCs) were included as covariates. The genotype data was converted to VCF format with PLINK2 and indexed with bcftools v1.6 (Li 2011) as required by QTLtools (Delaneau et al. 2017).

QTL mapping was conducted with QTLtools with `-grp-best` flag (Delaneau et al. 2017). The option `-window` was set to 1 Mb, `-nominal 1` for nominal pass and `-permute 1000` for a permutation pass. eGenes were defined as genes with permutation adjusted empirical value < 0.05 for the top associated variant.

Condition-specific and response eQTLs

Results

Running Code

When you click the **Render** button a document will be generated that includes both content and the output of embedded code. You can embed code like this:

```
1 + 1
```

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[1] 2
```

You can add options to executable code like this

```
[1] 4
```

The `echo: false` option disables the printing of code (only output is displayed).

References

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