

Fine Mapping Benchmark

`https://github.com/gaow/mvarbvs/tree/master/dsc`

May 9, 2018

Benchmark status

Data-set

Genotype

- A GTEx sample region (*FMO2*) of size $N = 698$, $P = 7492$
- A GUEVADIS sample region of size $N = 698$, $P = 1001$
- Parameters to “trim” P , eg from 7492 also to, say, 1001

Phenotype

- The original GTEx Throid and Lung expression for the GTEx sample region
 - should have around 3 eQTLs
- A simple simulated GUEVADIS expression data from DAP-g paper
 - 91 LD blocks

Simulation themes

Univariate

- Simple point mass + `rnorm()` simulation, as in DAP-g paper
- Point mass + mixture of normal, as in ASH paper
 - spiky, near-normal, flat-top, skew, big-normal, bimodal

Multivariate

- Column-wise stacking of univariate simulations
- Point mass + mixture of multivariate normal, as in MASH paper
 - All “canonical” prior covariances
 - Have to provide grid

There are mechanisms to

- Plot save LD heatmap for input data
- Put signals to the most “LD-convoluted” blocks
- Ensure signals are from independent LD blocks
- Permute & break LD structure?

Fine-mapping methods

From Stephens Lab

- varbvs
- susie
- M&M ASH

From the field

- DAP-g
- FINEMAP
- CAVIAR

Each with multiple module “flavors” (parameters)

Single-replicate diagnosis

- When available, compare scattered plots of $\tilde{\beta}$ or $\hat{\beta}$ vs β plots
- Show PIP and log10BF
- ...
- Customized diagnostic plots for SSE methods: `susie` and M&M.

Still working on unifying output from methods and annotate with eg LD info.

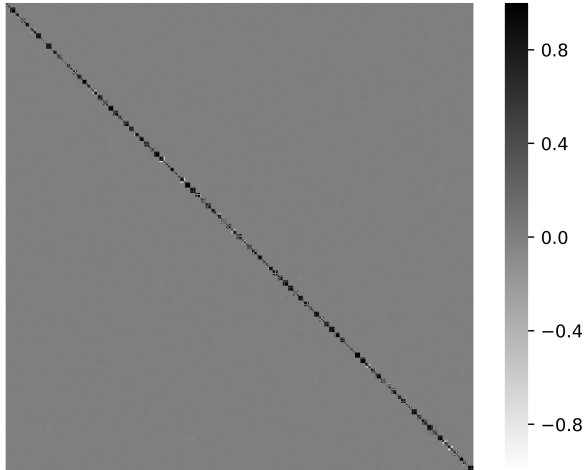
Cross-replicate evaluation

Next to-do:

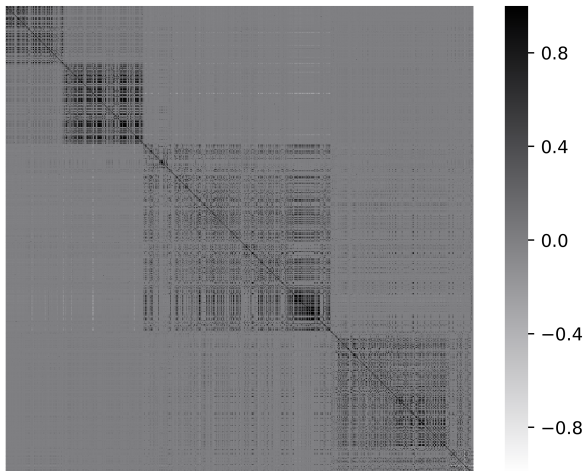
- Power vs false positive: ROC
- Point signal level: PIP / Ifsr
- Set signal level: cluster PIP

LD situation

GUEVADIS sample



Trimmed GTEx sample (to the size of GUEVADIS sample)



Simple GUEVADIS simulation

Simple GUEVADIS simulation

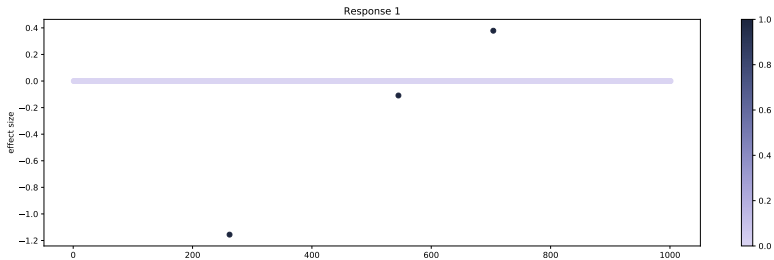


Figure 1: Simple GUEVADIS simulation

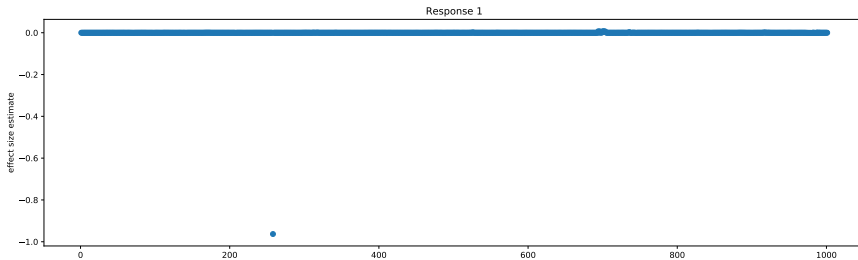


Figure 2: varbvs

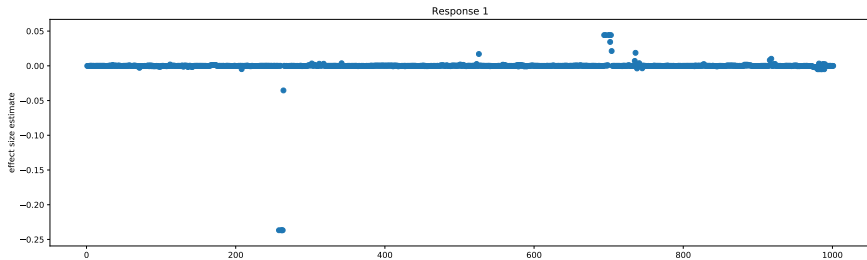
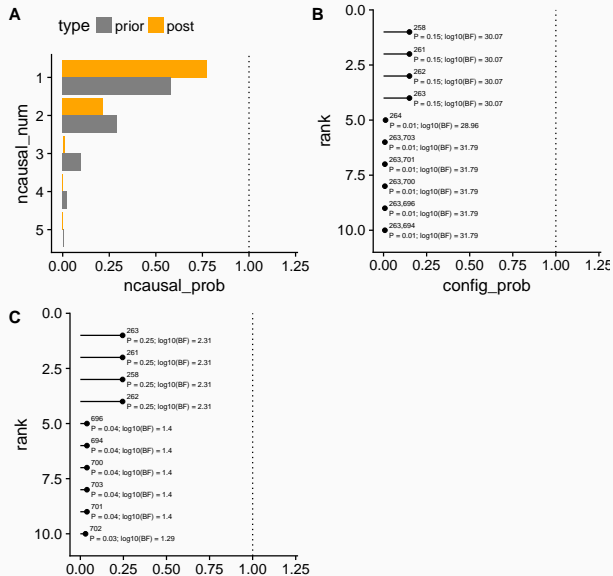
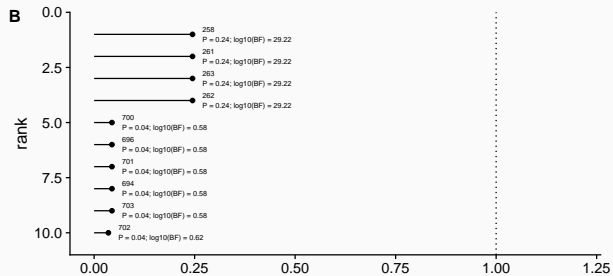
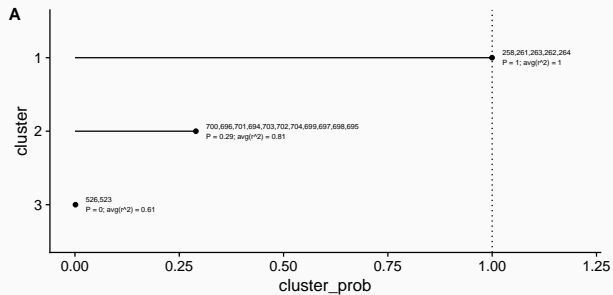
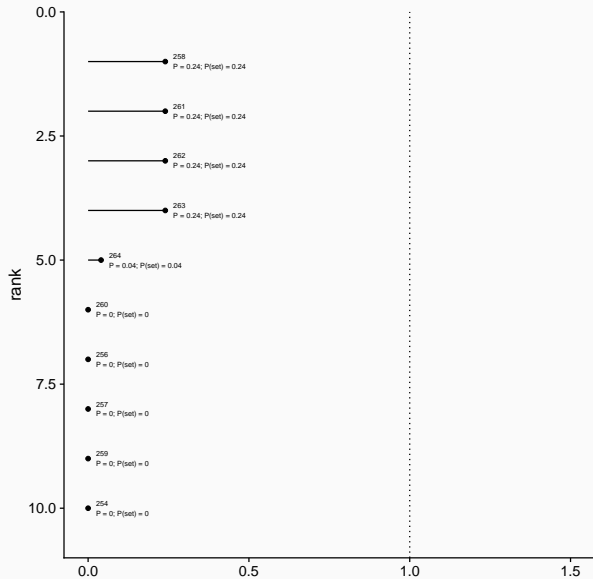


Figure 3: susie







GTEx with MASH “simple het” covariance

Trimmed GTEx, response 1

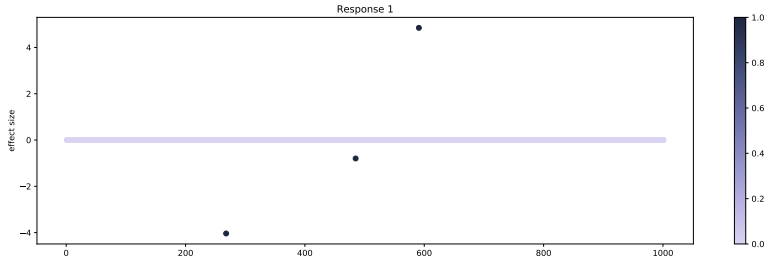


Figure 4: Simulated GTEx tissue 1

Trimmed GTEx, response 2

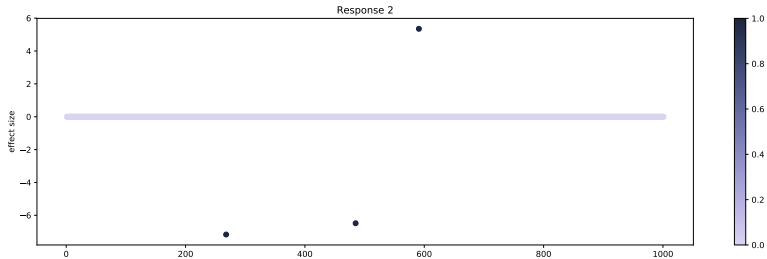


Figure 5: Simulated GTEx tissue 2

varbvs, response 1

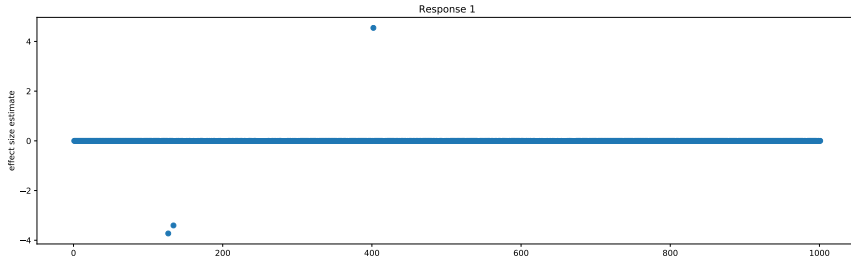


Figure 6: varbvs

varbvs, response 2

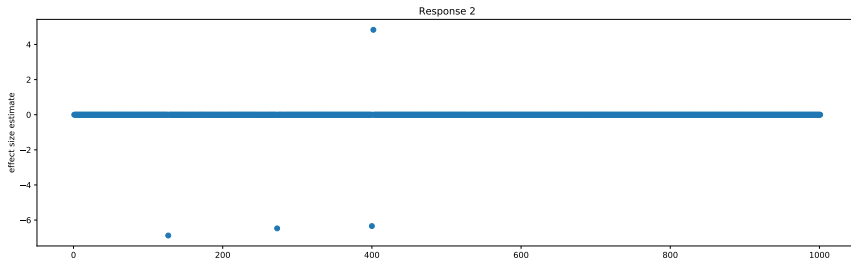


Figure 7: varbvs

susie, response 1

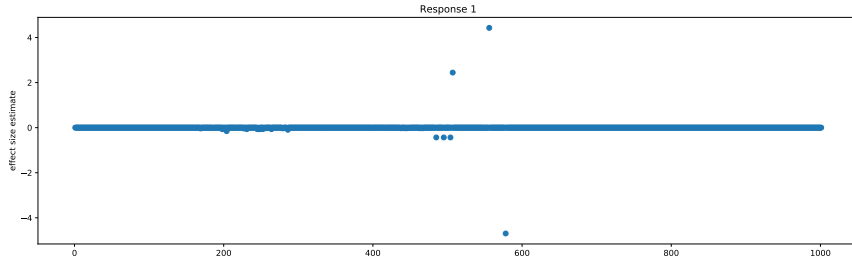


Figure 8: susie

susie, response 2

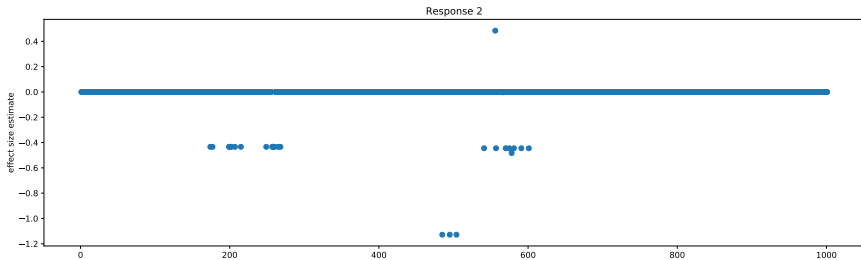


Figure 9: susie

M&M, response 1

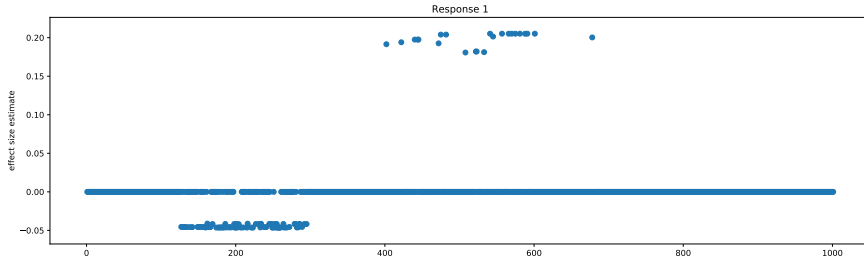


Figure 10: M&M

M&M, response 2

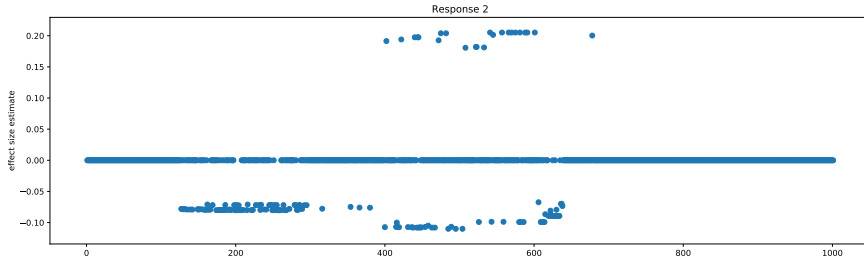


Figure 11: M&M