

# *Regional association study near ZmSWEET4c gene*

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## *Results*

With the control of the population structure and polygenic effects of background QTLs, we conducted the regional association scanning around the ZmSWEET4c gene region. As show in Figure A, two SNPs near the ZmSWEET4c gene were associated with the trait of total kernel weight, an important yield index trait. The most significant SNP could explained about 5% of the total phenotypic variation of the population and about 10% of the heritability of the trait (45% heritability could be accounted by the genome-wide SNPs). As shown in Figure B, non-B73 genotypes improved 4g of the TKW and the magitude of the improvement is 20%.

The Tufte-L<sup>A</sup>T<sub>E</sub>X [ˆtufte\_latex] document classes define a style similar to the style Edward Tufte uses in his books and handouts. Tufte's style is known for its extensive use of sidenotes, tight integration of graphics with text, and well-set typography.

## *Materials and Methods*

### *Regional association study*

A maize diversity panel composed of 282 inbred lines was employed for the regional association study. To conduct the analysis, we obtained Genotype-By-Sequencing (GBS) data from panzea ([www.panzea.org](http://www.panzea.org)) and obtained phenotypic data from Springer et al<sup>[2]</sup>. The SNPs data were filtered with minor allele frequency (MAF) > 0.05 and allele missing rate < 50%. After filtering, a total of 306,190 SNPs were remaining, including 79 SNPs in a 1-Mb region surrounding the ZmSWEET4c gene.

Association study with the mixed-model method was conducted using an R (citation) add-on package "GenABEL" (citation). First of all, a kinship matrix was estimated from the genomic data to control population structure. Secondly, genome-wide polygenic effects were computed with the function "polygenic" for the background control. Finally, the 45 SNPs near zmSWEET4c gene were tested one-by-one as the fixed effect and polygenic effects of genome-wide SNPs were fitted as random effects using the function "mmscore".

## Analysis Pipeline and source codes

Note: currently the pipeline sits in a private repo on github: <https://github.com/yangjl/Misc>. I can share the complete repo upon request. And the SNP data sit on farm in the dir of /group/jrigrp4/AllZeaGBSv2.7impV5.

### A.1 Obtain GBS and phenotypic data for maize diversity panel

```
source("../profiling/4.sweet/4.A.1_GBS_diverse.R")
```

### A.2 Run the following shell codes: convert HapMap to BED5 format

```
# open interactive srun on farm
srun.x11 -p bigmemh --ntasks=8 --nodelist=bigmem4
# run the shell
sh profiling/4.sweet/4.A.2_GBS_hdf2hmp.sh
```

### A.3 Convert hapmap to BED5 format

```
source("../profiling/4.sweet/4.A.3_GBS_bed5format.R")
# Run the following python code
snprfq -p /group/jrigrp4/AllZeaGBSv2.7impV5 -i ZeaGBSv27_Ames282.bed5 \\\
-s 6 -m "0" -a 0 -b 1 -c 2 -o ZeaGBSv27_Ames282.frq
```

### A.4 checking the SNP MAF and missing rate

```
source("../profiling/4.sweet/4.A.4_GBS_maf_mr.R")
```

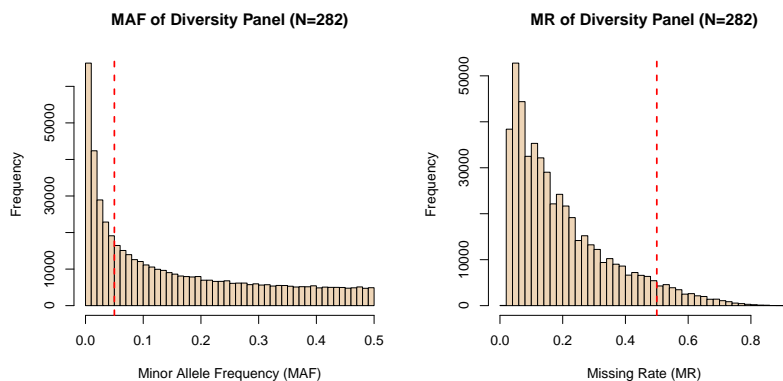


Figure 1: Minor allele frequency (MAF) and missing rate (MR) of the GBS SNPs of the maize diversity panel.

### B.1 derive the BLUE values for the phenotypic data and plot the histogram distribution of the traits.

```
source("../profiling/4.sweet/4.B.1_phenotype.R")
```

```
pheno <- read.table("../data/pheno_ames282.txt",
  header = TRUE)
par(mfrow = c(1, 2))
traits <- c("10 kernel weight", "Total kernel weight")
hist(pheno[, 3], breaks = 30, col = "cadetblue",
  main = "10 kernel weight", xlab = "weight (g)")
hist(pheno[, 8], breaks = 30, col = "cadetblue",
  main = "total kernel weight", xlab = "weight (g)")
```

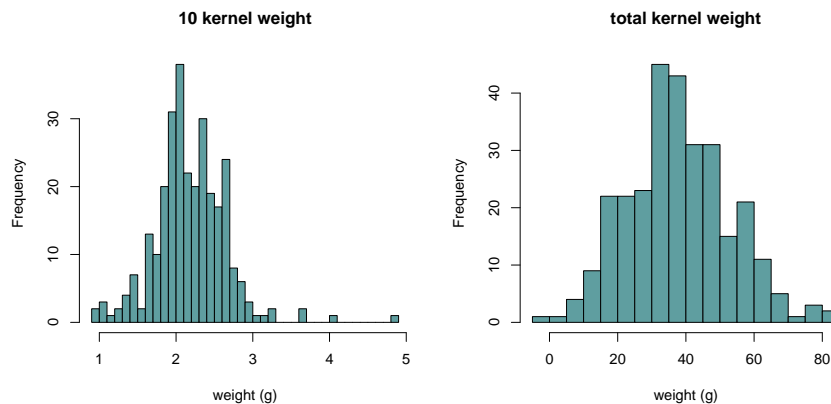


Figure 2: Histogram distribution of the phenotypic traits of 10 kernel weight and total kernel weight of the diversity panel.

## B.2 change the genotype format to GenABEL

```
source("../profiling/4.sweet/4.B.2_GBS_2GenABEL.R")
```

## B.3 and B.4 Regional association study

```
source("../profiling/4.sweet/4.B.3_GenABEL_step1.R")
```

```
source("../profiling/4.sweet/4.B.4_GenABEL_step2.R")
```

```
load("../cache/gwas_res.RData")
```

```
library(GenABEL)
```

```
## Loading required package: MASS
```

```
## Loading required package: GenABEL.data
```

```
par(mfrow = c(1, 2))
```

```
plot(res1, main = "10 kernel weight", pch = 16,
  col = "cadetblue")
```

```
abline(v = 127466000, lwd = 2, col = "red", lty = 2)
```

```
plot(res2, main = "total kernel weight", pch = 16,
  col = "cadetblue")
```

```
abline(v = 127466000, lwd = 2, col = "red", lty = 2)
```

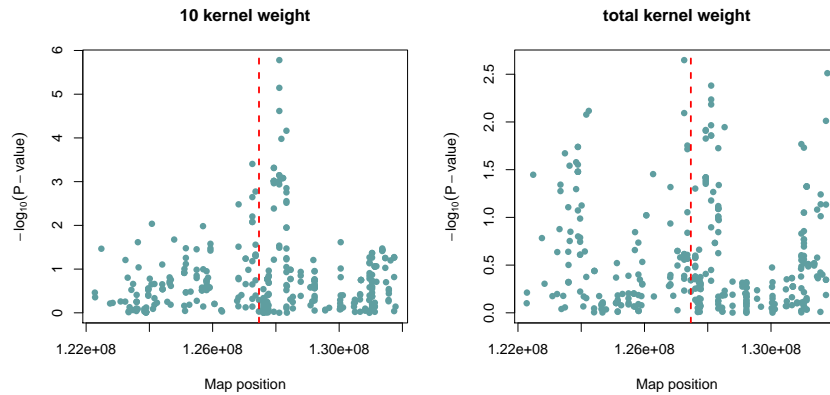


Figure 3: Regional GWAS with the simplest linear model.

```
# load('../cache/gwas_res.RData')
par(mfrow = c(1, 2))
plot(res1.eg, main = "10 kernel weight", pch = 16,
     col = "cadetblue")
abline(v = 127466000, lwd = 2, col = "red", lty = 2)
plot(res2.eg, main = "total kernel weight", pch = 16,
     col = "cadetblue")
abline(v = 127466000, lwd = 2, col = "red", lty = 2)
```

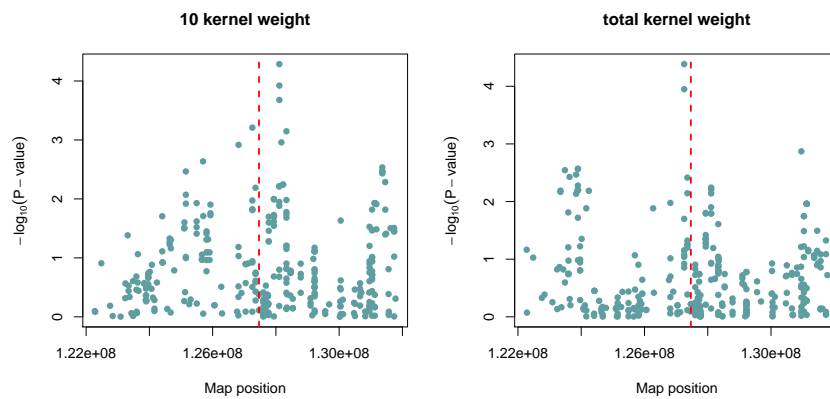


Figure 4: Regional GWAS with kinship matrix calculated from genome-wide marker to control the population structure.

```
# load('../cache/gwas_res.RData')
par(mfrow = c(1, 2))
plot(res1.mm, main = "10 kernel weight", pch = 16,
     col = "cadetblue")
abline(v = 127466000, lwd = 2, col = "red", lty = 2)
plot(res2.mm, main = "total kernel weight", pch = 16,
     col = "cadetblue")
abline(v = 127466000, lwd = 2, col = "red", lty = 2)
```

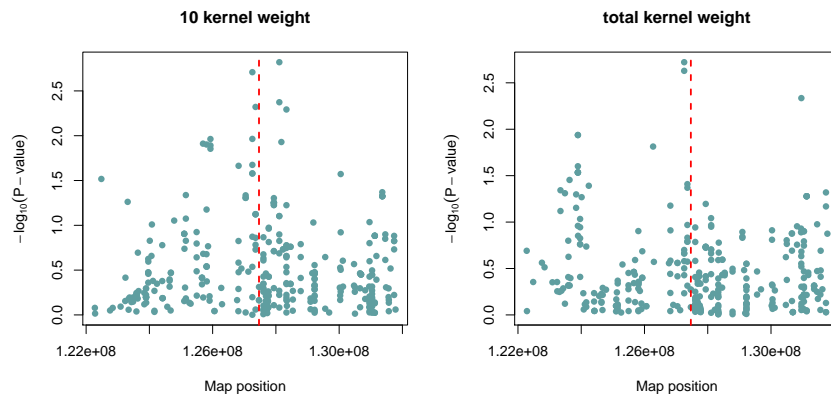


Figure 5: Regional GWAS with kinship matrix calculated from genome-wide marker to control the population structure and with the background QTL control.