BCB546-R_Assignment

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part I

Data inspection

Loading the 2 files to be the genotype and pos data frames

```
#Load the Library and import data
library(tidyverse)
```

```
## ── Attaching core tidyverse packages ──────────────────────── tidyverse
2.0.0 ──
## ✓ dplyr     1.1.0     ✓ readr     2.1.4
## ✓ forcats   1.0.0     ✓ stringr   1.5.0
## ✓ ggplot2   3.4.1     ✓ tibble    3.1.8
## ✓ lubridate 1.9.2     ✓ tidyr     1.3.0
## ✓ purrr     1.0.1
## ── Conflicts ────────────────────────────────────────
tidyverse_conflicts() ──
## ✖ dplyr::filter() masks stats::filter()
## ✖ dplyr::lag()    masks stats::lag()
## ℹ Use the ]8;;http://conflicted.r-lib.org/conflicted package]8;; to force
all conflicts to become errors
```

```
genotype <- as.data.frame(read.table("fang_et_al_genotypes.txt",
sep="\t",header=TRUE))
pos <- as.data.frame(read.table("snp_position.txt", sep="\t",header=TRUE))
```

SNP genotypes data

- `fang_et_al_genotypes.txt` is assigned to be the genotype data frame, which is large dimension data set, so use `dim`, `str`, `glimpse` and etc functions to know the number of rows and columns, column names and their variable types.

```
dim(genotype)
```

```
## [1] 2782  986
```

```
sapply(genotype, class)[1:6]
```

```
##   Sample_ID      JG_OTU       Group    abph1.20    abph1.22       ae1.3
## "character" "character" "character" "character" "character" "character"
```

```
#str(genotype)
#glimpse(genotype)
colnames(genotype)[1:6]

## [1] "Sample_ID" "JG_OTU"    "Group"     "abph1.20"  "abph1.22"  "ae1.3"

genotype[1:6,1:6]

##   Sample_ID   JG_OTU Group abph1.20 abph1.22 ae1.3
## 1     SL-15 T-aust-1 TRIPS      ?/?      ?/?   T/T
## 2     SL-16 T-aust-2 TRIPS      ?/?      ?/?   T/T
## 3     SL-11 T-brav-1 TRIPS      ?/?      ?/?   T/T
## 4     SL-12 T-brav-2 TRIPS      ?/?      ?/?   T/T
## 5     SL-18   T-cund TRIPS      ?/?      ?/?   T/T
## 6      SL-2 T-dact-1 TRIPS      ?/?      ?/?   T/T

genotype %>%
  group_by(Group) %>%
  count()

## # A tibble: 16 × 2
## # Groups:   Group [16]
##    Group     n
##    <chr> <int>
##  1 TRIPS    22
##  2 ZDIPL    15
##  3 ZLUXR    17
##  4 ZMHUE    10
##  5 ZMMIL   290
##  6 ZMMLR  1256
##  7 ZMMMR    27
##  8 ZMPBA   900
##  9 ZMPIL    41
## 10 ZMPJA    34
## 11 ZMXCH    75
## 12 ZMXCP    69
## 13 ZMXIL     6
## 14 ZMXNO     7
## 15 ZMXNT     4
## 16 ZPERR     9
```

About `fang_et_al_genotypes.txt` file:

- It has 2782 rows and 986 columns.
- It has 16 different groups.

SNP markers information

- `snp_position.txt` is assigned to be the pos data frame.
- Using the same function to know the data strcuture of the pos data frame.

- Replacing the `unknown` and `multiple` in Position column to be `NA` and to know how many numbers of SNP markers, and their maximum and minimum position value in each of chromosome.

```
dim(pos)

## [1] 983  15

sapply(pos, class)[1:6]

##         SNP_ID  cdv_marker_id     Chromosome       Position        alt_pos
##    "character"      "integer"    "character"    "character"    "character"
## mult_positions
##    "character"

str(pos)

## 'data.frame':    983 obs. of  15 variables:
##  $ SNP_ID              : chr  "abph1.20" "abph1.22" "ae1.3" "ae1.4" ...
##  $ cdv_marker_id       : int  5976 5978 6605 6606 6607 5982 3463 3466 5983
5985 ...
##  $ Chromosome          : chr  "2" "2" "5" "5" ...
##  $ Position            : chr  "27403404" "27403892" "167889790"
"167889682" ...
##  $ alt_pos             : chr  "" "" "" "" ...
##  $ mult_positions      : chr  "" "" "" "" ...
##  $ amplicon            : chr  "abph1" "abph1" "ae1" "ae1" ...
##  $ cdv_map_feature.name: chr  "AB042260" "AB042260" "ae1" "ae1" ...
##  $ gene                : chr  "abph1" "abph1" "ae1" "ae1" ...
##  $ candidate.random    : chr  "candidate" "candidate" "candidate"
"candidate" ...
##  $ Genaissance_daa_id  : int  8393 8394 8395 8396 8397 8398 8399 8400 8401
8402 ...
##  $ Sequenom_daa_id     : int  10474 10475 10477 10478 10479 10481 10482
10483 10486 10487 ...
##  $ count_amplicons     : int  1 0 1 0 0 1 1 0 1 0 ...
##  $ count_cmf           : int  1 0 1 0 0 1 0 0 1 0 ...
##  $ count_gene          : int  1 0 1 0 0 1 1 0 1 0 ...

glimpse(pos)

## Rows: 983
## Columns: 15
## $ SNP_ID              <chr> "abph1.20", "abph1.22", "ae1.3", "ae1.4",
"ae1.5"…
## $ cdv_marker_id       <int> 5976, 5978, 6605, 6606, 6607, 5982, 3463,
3466, 5…
## $ Chromosome          <chr> "2", "2", "5", "5", "5", "1", "3", "3", "4",
"4",…
## $ Position            <chr> "27403404", "27403892", "167889790",
"167889682",…
## $ alt_pos             <chr> "", "", "", "", "", "", "", "", "", "", "",
```

```
"", "…
## $ mult_positions       <chr> "", "", "", "", "", "", "", "", "", "", "",
"", "…
## $ amplicon             <chr> "abph1", "abph1", "ae1", "ae1", "ae1", "an1",
"ba…
## $ cdv_map_feature.name <chr> "AB042260", "AB042260", "ae1", "ae1", "ae1",
"an1…
## $ gene                 <chr> "abph1", "abph1", "ae1", "ae1", "ae1", "an1",
"ba…
## $ candidate.random     <chr> "candidate", "candidate", "candidate",
"candidate…
## $ Genaissance_daa_id   <int> 8393, 8394, 8395, 8396, 8397, 8398, 8399,
8400, 8…
## $ Sequenom_daa_id      <int> 10474, 10475, 10477, 10478, 10479, 10481,
10482, …
## $ count_amplicons      <int> 1, 0, 1, 0, 0, 1, 1, 0, 1, 0, 0, 1, 0, 1, 1,
0, 1…
## $ count_cmf            <int> 1, 0, 1, 0, 0, 1, 0, 0, 1, 0, 0, 1, 0, 1, 1,
0, 1…
## $ count_gene           <int> 1, 0, 1, 0, 0, 1, 1, 0, 1, 0, 0, 1, 0, 1, 1,
0, 1…
```

```
colnames(pos)[1:6]
```

```
## [1] "SNP_ID"         "cdv_marker_id"  "Chromosome"     "Position"
## [5] "alt_pos"        "mult_positions"
```

```
pos[1:6,1:6]
```

```
##     SNP_ID cdv_marker_id Chromosome  Position alt_pos mult_positions
## 1 abph1.20          5976          2  27403404
## 2 abph1.22          5978          2  27403892
## 3    ae1.3          6605          5 167889790
## 4    ae1.4          6606          5 167889682
## 5    ae1.5          6607          5 167889821
## 6    an1.4          5982          1 240498509
```

```
pos[pos == "unknown"] <- NA
pos[pos == "multiple"] <- NA
pos %>%
  group_by(Chromosome) %>%
  summarise(Max=max(Position, na.rm = T), Min=min(Position, na.rm = T),
Number=length(Position))
```

```
## # A tibble: 11 × 4
##    Chromosome Max        Min         Number
##    <chr>      <chr>      <chr>        <int>
##  1 1          "95897171" "10069039"     155
##  2 10         "96216463" "10432605"      53
##  3 2          "69623323" "10429605"     127
##  4 3          "95541392" "106631676"    107
```

```
##  5 4          "78946482" "103665461"     91
##  6 5          "945545"   "100227859"    122
##  7 6          "98507715" "113705211"     76
##  8 7          "43948320" "104898448"     97
##  9 8          "83913342" "115257234"     62
## 10 9          "94285743" "104237516"     60
## 11 <NA>       ""         ""              33
```

About `snp_position.txt` file:

- It has 983 rows and 15 column.

Data processing

Subset the pos data frame to keep the SNP_ID, Chr, and Pos columns for the merging purpose

- Adjust the variable type of `chromosome` and `Position` to be numeric and remove the `unknown` and `multiple` which are regarded as NAs out to be posred dataframe.

- This following code is performing several operations on the data frame pos using the dplyr package and assigning the resulting data frame to a new object called posred. The str() function is then used to display the structure of posred.

Let's break down the individual operations:

`dplyr::select(SNP_ID, Chromosome, Position)`: This selects only the columns named SNP_ID, Chromosome, and Position from the pos data frame.

`mutate(Chromosome=as.numeric(Chromosome), Position=as.numeric(Position))`: This converts the Chromosome and Position columns to numeric data types using the as.numeric() function. The resulting numeric values are assigned back to the same columns.

`filter_all(all_vars(. != "NA"))`: This filters out any rows where any column has a missing value ("NA"). The filter_all() function applies the same condition to all columns, and all_vars(. != "NA") specifies the condition that all values in each column must not be equal to "NA". The . represents the current column being evaluated.

Overall, this code is creating a new data frame posred that contains only the selected columns from pos, with Chromosome and Position converted to numeric data types, and any rows containing missing values are removed. The `str()` function is then used to display the structure of the resulting data frame.

```
posred <- pos %>%
  dplyr::select(SNP_ID, Chromosome, Position) %>%
  mutate(Chromosome=as.numeric(Chromosome),
         Position=as.numeric(Position))%>%
  filter_all(all_vars(. != "NA"))
str(posred)
```

```
## 'data.frame':    939 obs. of  3 variables:
##  $ SNP_ID    : chr  "abph1.20" "abph1.22" "ae1.3" "ae1.4" ...
```

```
##  $ Chromosome: num  2 2 5 5 5 1 3 3 4 4 ...
##  $ Position  : num  2.74e+07 2.74e+07 1.68e+08 1.68e+08 1.68e+08 ...
```

Subset the genotype data into maize and teosinte datasets

- Subset the maize and teosinte genotypes by the `Group` column. Either `[ which()]` or `%>% filter()` works out.

```
maize <- genotype[which(genotype$Group=="ZMMIL" | genotype$Group =="ZMMLR" |
genotype$Group == "ZMMMR"),]
teosinte <-genotype[which(genotype$Group=="ZMPBA" | genotype$Group =="ZMPIL"
| genotype$Group == "ZMPJA"),]
```

- Another method:

```
maize <- genotype %>% filter(Group=="ZMMIL" | Group =="ZMMLR" | Group ==
"ZMMMR")
teosinte <-genotype %>% filter(Group=="ZMPBA" | Group =="ZMPIL" | Group ==
"ZMPJA")
```

Formatting the maize genotype with SNP information by merging the posred and maize data

- Transform the `maize` data for merging with `posred` by `SNP_ID` column, and descend the Chromosome and Position.

```
maize <- maize[,c(-2,-3)]
maize[1:6,1:6] ## have a look

##      Sample_ID abph1.20 abph1.22 ae1.3 ae1.4 ae1.5
## 1210 ZDP_0752a      C/G      A/A   T/T   G/G   C/C
## 1211 ZDP_0793a      C/G      A/A   T/T   G/G   C/T
## 1212 ZDP_0612a      C/C      A/A   T/T   G/G   C/C
## 1213 ZDP_0602a      C/G      A/A   G/T   A/G   C/T
## 1214 ZDP_0581a      C/C      A/A   T/T   G/G   C/T
## 1215 ZDP_0552a      C/G      A/A   T/T   G/G   C/T

maize <- t(maize)
maize <- cbind(rownames(maize),maize)
rownames(maize) <- NULL
colnames(maize) <- maize[1,]
maize <- maize[-1,]
maize <- as.data.frame(maize)
colnames(maize)[1] <- "SNP_ID"
maizewp <- merge(posred, maize, by = "SNP_ID")
maizewp <- maizewp %>% arrange(Chromosome,Position)
## maize genotypes with SNP position information
```

- This above code is performing several operations on the maize data frame and the posred data frame, and assigning the resulting data frame to a new object maizewp.

Here's a breakdown of the individual operations:

- `maize <- maize[,c(-2,-3)]`: This selects all columns of the maize data frame except for the second and third columns, and reassigns the resulting data frame to maize.

- `maize <- t(maize)`: This transposes the maize data frame.
- `maize <- cbind(rownames(maize),maize)`: This adds a new column to the maize data frame containing the row names of the original data frame.
- `rownames(maize) <- NULL`: This removes the row names from the maize data frame.
- `colnames(maize) <- maize[1,]`: This sets the column names of the maize data frame to the values in the first row.
- `maize <- maize[-1,]`: This removes the first row of the maize data frame.
- `maize <- as.data.frame(maize)`: This converts maize to a data frame.
- `colnames(maize)[1] <- "SNP_ID"`: This renames the first column of maize to "SNP_ID".
- `maizewp <- merge(posred, maize, by = "SNP_ID")`: This merges the posred data frame with the modified maize data frame using the common column "SNP_ID". The resulting data frame is assigned to maizewp.
- `maizewp <- maizewp %>% arrange(Chromosome,Position)`: This arranges the rows of maizewp in ascending order of Chromosome and Position using the arrange() function from the dplyr package.

Overall, this code is modifying the maize data frame by removing certain columns, transposing the data frame, and adding and removing row and column names. It then merges the modified maize data frame with the posred data frame and sorts the resulting data frame by Chromosome and Position. The resulting data frame is assigned to maizewp.

Formatting the teosinte genotype with SNP information by merging the posred and teosinte data

- The same methods as with maize for merging data frame.

```
teosinte <- teosinte[,c(-2,-3)]
teosinte <- t(teosinte)
teosinte <- cbind(rownames(teosinte),teosinte)
rownames(teosinte) <- NULL
colnames(teosinte) <- teosinte[1,]
teosinte <- teosinte[-1,]
teosinte <- as.data.frame(teosinte)
colnames(teosinte)[1] <- "SNP_ID"
teosintewp <- merge(posred, teosinte, by = "SNP_ID")
teosintewp <- teosintewp %>% arrange(Chromosome,Position)
```

Splitting the maize data into different files by the chromosomes and SNP positions.

- The followings are using `loop` to separate the `maizewp` and `teosintewp` data frames to 10, 10, 10, and 10 files, respectively, by the chromosome and SNP positions in total 40 files. Also, change the missing genotype to be `?` or `-`.

```
chr <- 1:10
for (i in chr) {
  files_inc <- maizewp[maizewp$Chromosome == i,]
  files_inc[files_inc == "?/?"] <- "?"
  if (i < 10) { write.table(files_inc, file =
paste("Maize_Chr0",i,"_increase.txt",sep=""),row.names = FALSE,sep =
"\t",quote = FALSE) }
  else {write.table(files_inc, file =
paste("Maize_Chr",i,"_increase.txt",sep=""),row.names = FALSE, sep =
"\t",quote = FALSE)}

  files_dec <- maizewp[maizewp$Chromosome == i,]
  files_dec[files_dec == "?/?"] <- "-"
  files_dec <- files_dec %>% arrange(desc(Chromosome),desc(Position))
  if (i < 10) { write.table(files_dec, file =
paste("Maize_Chr0",i,"_decrease.txt",sep=""),row.names = FALSE,sep =
"\t",quote = FALSE) }
  else {write.table(files_dec, file =
paste("Maize_Chr",i,"_decrease.txt",sep=""),row.names = FALSE, sep =
"\t",quote = FALSE)}
}
```

Splitting the teosinte data into different files by chromosomes and SNP positions.

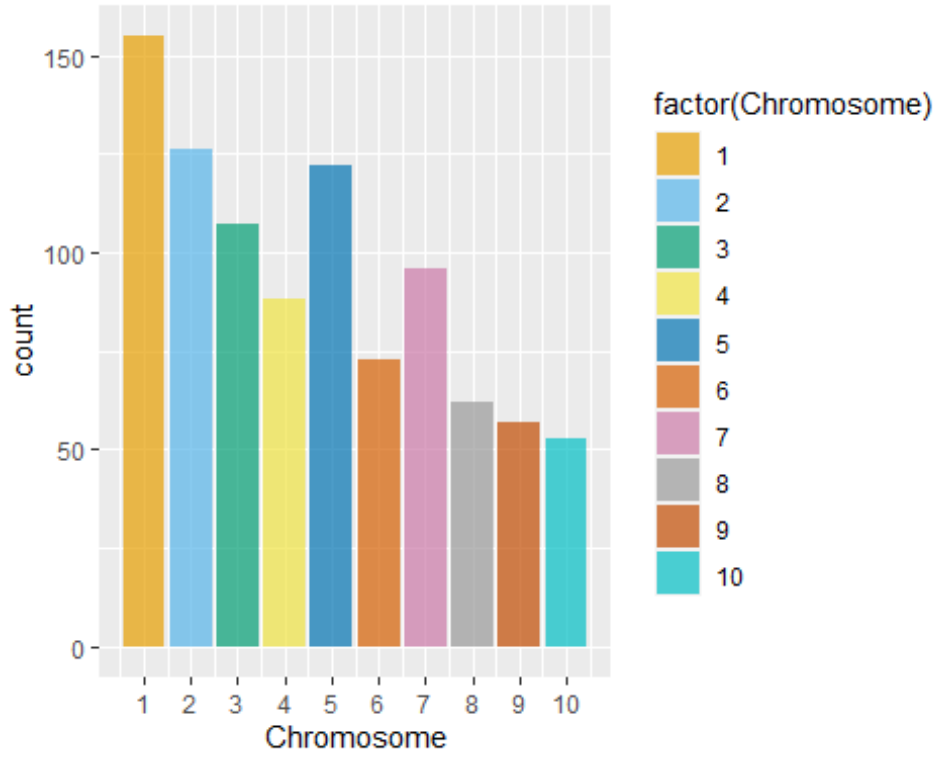
```
chr <- 1:10
for (i in chr) {
  files_inc <- teosintewp[teosintewp$Chromosome == i,]
  files_inc[files_inc == "?/?"] <- "?"
  if (i < 10) { write.table(files_inc, file =
paste("Teosinte_Chr0",i,"_increase.txt",sep=""),row.names = FALSE,sep =
"\t",quote = FALSE) }
  else {write.table(files_inc, file =
paste("Teosinte_Chr",i,"_increase.txt",sep=""),row.names = FALSE, sep =
"\t",quote = FALSE)}

  files_dec <- teosintewp[teosintewp$Chromosome == i,]
  files_dec[files_dec == "?/?"] <- "-"
  files_dec <- files_dec %>% arrange(desc(Chromosome),desc(Position))
  if (i < 10) { write.table(files_dec, file =
paste("Teosinte_Chr0",i,"_decrease.txt",sep=""),row.names = FALSE,sep =
"\t",quote = FALSE) }
  else {write.table(files_dec, file =
paste("Teosinte_Chr",i,"_decrease.txt",sep=""),row.names = FALSE, sep =
"\t",quote = FALSE)}
}
```

Part II

Plotting

SNPs per chromosome

```
pos %>%
    dplyr::select(SNP_ID, Chromosome, Position) %>%
    drop_na() %>%
    mutate(Chromosome = as.double(Chromosome)) %>%
    ggplot() +
    geom_bar(aes(x = Chromosome, fill = factor(Chromosome)), alpha = 0.7) +
    scale_x_continuous(breaks = 1:10) +
    scale_fill_manual(values = c("#E69F00", "#56B4E9", "#009E73", "#F0E442",
"#0072B2", "#D55E00", "#CC79A7", "#999999", "#C14B00", "#00BFC4"))
```



Explanation of the above script:

This is a series of operations using the *dplyr* and *ggplot2* packages to create a bar plot of the number of SNPs on each chromosome in a data frame called pos. Here's an explanation of each operation:

- `pos %>%`: %>% is the pipe operator, which takes the output of the left-hand side of the pipe and passes it as the first argument to the function on the right-hand side of the pipe. In this case, it takes the pos data frame and passes it as the input to the next function.

- `dplyr::select(SNP_ID, Chromosome, Position) %>%`: The select() function from dplyr is used to select only the columns "SNP_ID", "Chromosome", and "Position" from the data frame. The `dplyr::` syntax specifies that we want to use the select() function from the dplyr package. The %>% operator passes the output of this function to the next function.

- `drop_na() %>%`: The drop_na() function from dplyr is used to remove any rows that contain missing values ("NA") in any of the columns. The %>% operator passes the output of this function to the next function.

- `mutate(Chromosome=as.double(Chromosome)) %>%`: The mutate() function from dplyr is used to create a new column called "Chromosome" that converts the "Chromosome" column to a numeric data type using the as.double() function. The %>% operator passes the output of this function to the next function.

- `ggplot()+`: The ggplot() function from ggplot2 is used to initialize a new plot. The + operator is used to add layers to the plot.

- `geom_bar(aes(x=Chromosome))+`: The geom_bar() function from ggplot2 is used to create a bar plot of the number of SNPs on each chromosome. The aes() function is used to map the "Chromosome" column to the x-axis. The + operator is used to add the geom_bar() layer to the plot.

- `scale_x_continuous(breaks = 1:10)`: The scale_x_continuous() function from ggplot2 is used to set the breaks (i.e., tick marks) on the x-axis to the integers 1 through 10. This function specifies that the x-axis should be a continuous scale.

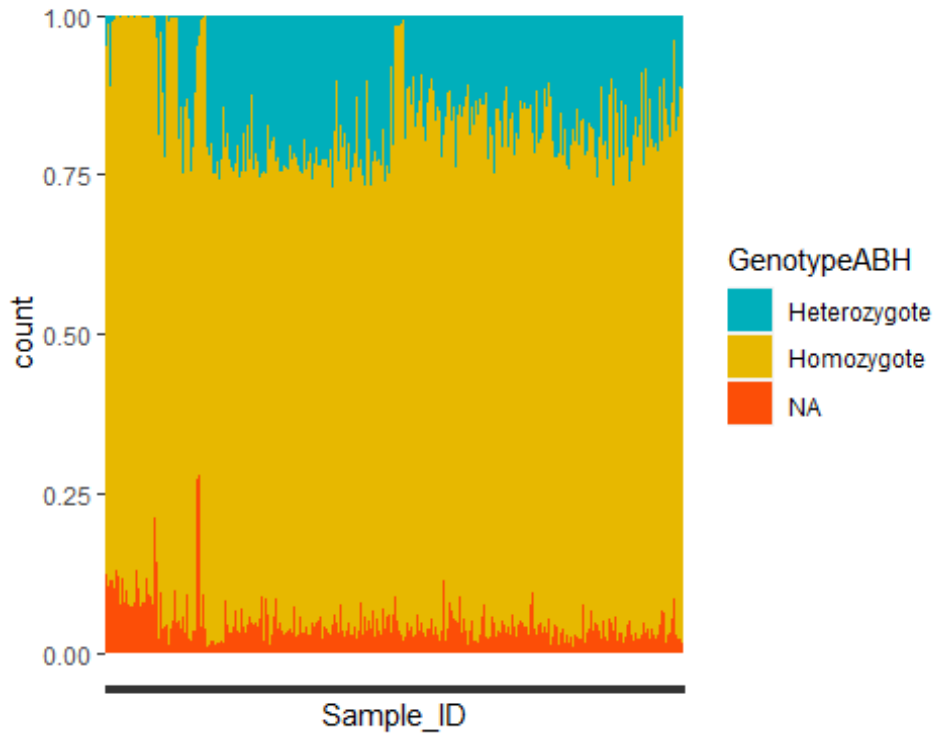
Missing data and amount of heterozygosity

- This chunk is the first version code for substituting the genotypes.

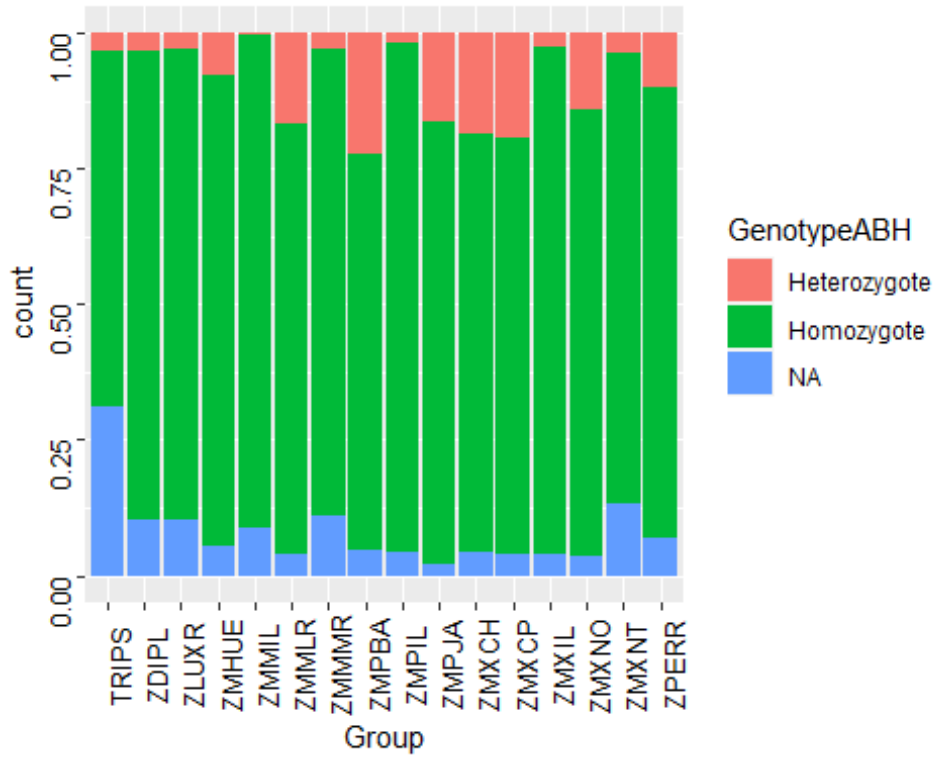
```
## create a function to detect the SNP genotypes
ABH <- function(x) {
  if ( x == "A/A" | x == "C/C" | x == "G/G" | x == "T/T") {
    return("A|B")
  }
  else if (x == "?/?") {
    return("NA")
  }
  else {return("H")}
}
ABH_V <- Vectorize(ABH) ## make the function be a vectorized function
genotype3 <- genotype2 %>%
  pivot_longer(3:last_col(), names_to = "SNP", values_to = "Genotype") %>%
  mutate( GenotypeABH = ABH_V(Genotype))

color_Palette = c("#00AFBB", "#E7B800", "#FC4E07")
genotype2 <- genotype[,-2]
genotype2[1:6,1:6]
```

```
##   Sample_ID Group abph1.20 abph1.22 ae1.3 ae1.4
## 1     SL-15 TRIPS      ?/?      ?/?   T/T   G/G
## 2     SL-16 TRIPS      ?/?      ?/?   T/T   ?/?
## 3     SL-11 TRIPS      ?/?      ?/?   T/T   G/G
## 4     SL-12 TRIPS      ?/?      ?/?   T/T   G/G
## 5     SL-18 TRIPS      ?/?      ?/?   T/T   G/G
## 6      SL-2 TRIPS      ?/?      ?/?   T/T   G/G
```

```
genotype3 <- genotype2 %>%
  pivot_longer(3:last_col(), names_to = "SNP", values_to = "Genotype") %>%
  mutate( GenotypeABH = ifelse(Genotype%in% c("C/C", "G/G", "A/A", "T/T"),
"Homozygote", ifelse(Genotype == "?/?", "NA", "Heterozygote")))
ggplot(genotype3)+
  geom_bar(aes(x=Sample_ID, fill=GenotypeABH), position = "fill", width=1)+
  scale_x_discrete(labels=NULL)+
  scale_fill_manual(values = c("#00AFBB", "#E7B800", "#FC4E07")) ## change
the color palette
```




```
ggplot(genotype3)+
  geom_bar(aes(x=Group, fill=GenotypeABH), position = "fill")+
  theme(axis.text = element_text( angle =90, color="black", size=10, face=1))
```



The distribution of SNP maker postions on each of chromosomes

```
sample_size = posred %>% group_by(Chromosome) %>% summarize(num=n())
library(viridis)
```

```
## Loading required package: viridisLite
```

```
posred %>%
  left_join(sample_size) %>%
  mutate(myaxis = paste0(Chromosome, "\n", "n=", num)) %>%
  ggplot( aes(x=myaxis, y=Position, fill=as.character(Chromosome)))+
  geom_violin(width=1.4) +
  geom_boxplot(width=0.1, color="grey", alpha=0.2) +
  scale_fill_viridis(discrete = TRUE) +
    theme(
      legend.position="none",
      plot.title = element_text(size=11)
    ) +
    ggtitle("The distribution of SNP postion on each chromosomes") +
    xlab("Chromosome")
```

```
## Joining with `by = join_by(Chromosome)`
```


The distribution of SNP postion on each chromosomes
Position
3e+08
2e+08
1e+08
0e+00
1
n=155
10
n=53
2
n=126
3
n=107
4
n=88
5
n=122
6
n=73
7
n=96
8
n=62
9
n=57
Chromosome