

## SUMSTATS

Description of GWAS summary statistics

The repository shows how to provide input for PW-pipeline, FM-pipeline including GCTA, and possibly others here.

Briefly, the format has the following columns,

| Column | Name   | Description              |
|--------|--------|--------------------------|
| 1      | SNP    | RSid                     |
| 2      | A1     | Effect allele            |
| 3      | A2     | Other allele             |
| 4      | freqA1 | A1 frequency             |
| 5      | beta   | effect estimate          |
| 6      | se     | standard error of effect |
| 7      | P      | P-value                  |
| 8      | N      | sample size              |
| 9*     | chr    | chromosome               |
| 10*    | pos    | position                 |

\* These two columns can be obtained from UCSC as shown below.

## Information from UCSC

The chromosomal positions for the current build can be downloaded from the UCSC website, which should be helpful for GWAS summary statistics either using chromosomal positions from different build or without these at all.

```
wget http://hgdownload.soe.ucsc.edu/goldenPath/hg19/database/snp150.txt.gz
gunzip -c snp150.txt.gz | \
awk '{split($2,a,"_");sub(/chr/, "",a[1]);print a[1],$4,$5}' | \
sort -k3,3 > snp150.txt
```

where it first obtains build 37 positions, sorts them by RSid into the file snp150.txt.

## Examples

### BMI

We take data reported by Locke, et al. (2015) as example which requires build 37 positions than can be downloaded from the UCSC website described above.

```
# GWAS summary statistics
wget
http://portals.broadinstitute.org/collaboration/giant/images/1/15/SNP_gwas_mc
```

```
_merge_nogc.tbl.uniq.gz
gunzip -c SNP_gwas_mc_merge_nogc.tbl.uniq.gz |
awk 'NR>1' | \
sort -k1,1 | \
join -11 -23 - snp150.txt | \
awk '($9!="X" && $9!="Un")' > bmi.txt
```

where file containing the GWAS summary statistics is downloaded, its header dropped, sorted and positional information added leading to a file named `bmi.txt`. We also filter out nonautosomal SNPs.

## T2D

The data was reported by Scott, et al. (2017),

```
R -q --no-save <<END
```

```
library(openxlsx)
library(dplyr)
```

```
xlsx <-
"http://diabetes.diabetesjournals.org/highwire/filestream/79037/field_highwir
e_adjunct_files/1/DB161253SupplementaryData2.xlsx"
```

```
# Supplementary Table 3. Results for established, novel and additional
distinct signals from the main analysis.
```

```
ST3 <- read.xlsx(xlsx, sheet = 3, colNames=TRUE, skipEmptyRows = FALSE, cols
= 1:20, rows = 2:130) %>%
```

```
  rename(P="p-value.in.stage.1") %>% within(
  {
    beta=log(OR)
    L <- as.numeric(substr(CI,1,4))
    U <- as.numeric(substr(CI,6,9))
    se=abs(log(L)-log(U))/3.92
  }) %>% select(
  SNP=rsid,
  A1=EA,
  A2=NEA,
  freqA1=EAF,
  beta,
  se,
  P,
  N=Sample.size,
  chr=Chr,
  pos=Position_b37
)
```

```
write.table(ST3, file="ST3", row.names=FALSE, col.names=FALSE, quote=FALSE)
```

```
# Supplementary Table 4. BMI-unadjusted association analysis model
```

```
ST4 <- read.xlsx(xlsx, sheet = 4, colNames=TRUE, skipEmptyRows = FALSE, cols
= 1:12, rows = 3:132) %>% rename(
```

```

    "CI"="CI.95%",
    "P"="P-value") %>% within(
{
  beta=log(OR)
  L <- as.numeric(substr(CI,1,4))
  U <- as.numeric(substr(CI,6,9))
  se=abs(log(L)-log(U))/3.92
  P=2*(1-pnorm(abs(beta/se)))
}) %>% select(
  SNP=rsid,
  A1=allele1,
  A2=allele2,
  freqA1=freq1,
  beta,
  se,
  P,
  N,
  chr,
  pos=position_b37
)
write.table(ST4, file="ST4", row.names=FALSE, col.names=FALSE, quote=FALSE)

END

```

where we generate data based on the paper's supplementary tables ST3 and ST4; the former is in line with the paper (by specifying `_db=depict` and `p_threshold=0.00001` when calling PW-pipeline).

## References

**GIANT** (Genetic Investigation of ANthropometric Traits) data

Locke AE, et al. (2015) Genetic studies of body mass index yield new insights for obesity biology. *Nature* 518(7538):197-206. doi: 10.1038/nature14177

**DIAGRAM** (DIAbetes Genetics Replication And Meta-analysis) data

Scott R, et al. (2017) An Expanded Genome-Wide Association Study of Type 2 Diabetes in Europeans. *Diabetes* 66:2888–2902.