

# eda\_\_t2d-credible-kyle-metabo.Rmd

Jason Torres

February 2, 2017

```
"%&%&" <- function(a,b) paste0(a,b)
library("data.table")
library("dplyr")
```

```
## -----

## data.table + dplyr code now lives in dtplyr.
## Please library(dtplyr)!

## -----

##
## Attaching package: 'dplyr'

## The following objects are masked from 'package:data.table':
##
##   between, first, last

## The following objects are masked from 'package:stats':
##
##   filter, lag

## The following objects are masked from 'package:base':
##
##   intersect, setdiff, setequal, union

library("ggplot2")
```

```
## Warning: package 'ggplot2' was built under R version 3.3.2
```

```
serv.dir <- "/Users/jtorres/FUSE/"
cred.dir <- serv.dir %&% "reference/credible_sets/from_kyle/"
cred.file <- cred.dir %&% "metabochip.chr_added.vcf"

df <- fread(cred.file)
```

Evaluate and reformat data frame

```
str(df)

## Classes 'data.table' and 'data.frame':  19266 obs. of  8 variables:
## $ V1: chr  "chr1" "chr1" "chr1" "chr1" ...
## $ V2: int  120437718 120437884 120438577 120439109 120440029 120441998 120442257 120443424 12044354
## $ V3: chr  "rs2793823" "rs2641348" "rs147294252" "rs6668119" ...
## $ V4: chr  "G" "A" "G" "G" ...
## $ V5: chr  "A" "G" "A" "C" ...
## $ V6: int  100 100 100 100 100 100 100 100 100 100 ...
## $ V7: chr  "PASS" "PASS" "PASS" "PASS" ...
## $ V8: chr  "LOCUS=NOTCH2;PROB=0.00871;" "LOCUS=NOTCH2;PROB=0.01154;" "LOCUS=NOTCH2;PROB=0.00048;" "
## - attr(*, ".internal.selfref")=<externalptr>

locus <- as.character(sapply(df$V8,function(string){
  gsub("LOCUS=", "", strsplit(string, split=";")[[1]][1])
})
```

```

}))
prob <- as.character(sapply(df$V8,function(string){
  gsub("PROB=", "", strsplit(string, split=";")[[1]][2])
}))
df <- select(df, one_of("V1", "V2", "V3", "V4", "V5"))
names(df) <- c("chr", "pos", "rsid", "A1", "A2")
df <- cbind(df, locus, prob)
df <- as.data.frame(df)
df$prob <- as.numeric(df$prob)

```

There are 49 loci in this file

Build locus summary data frame

```

loci <- unique(df$locus)
loc <- loci[1]
numsnps <- as.integer(sapply(loci, function(loc){
  length(filter(df, locus==loc)$prob)
}))
prop01 <- as.numeric(sapply(loci, function(loc){
  sum(filter(df, locus==loc)$prob > 0.01)/length(filter(df, locus==loc)$prob)
}))
prop05 <- as.numeric(sapply(loci, function(loc){
  sum(filter(df, locus==loc)$prob > 0.05)/length(filter(df, locus==loc)$prob)
}))
prop10 <- as.numeric(sapply(loci, function(loc){
  sum(filter(df, locus==loc)$prob > 0.10)/length(filter(df, locus==loc)$prob)
}))
prop20 <- as.numeric(sapply(loci, function(loc){
  sum(filter(df, locus==loc)$prob > 0.20)/length(filter(df, locus==loc)$prob)
}))

loc.df <- data.frame(loci, numsnps, prop01, prop05, prop10, prop20,
  stringsAsFactors = FALSE)

```

Histograms summarizing loci distributions

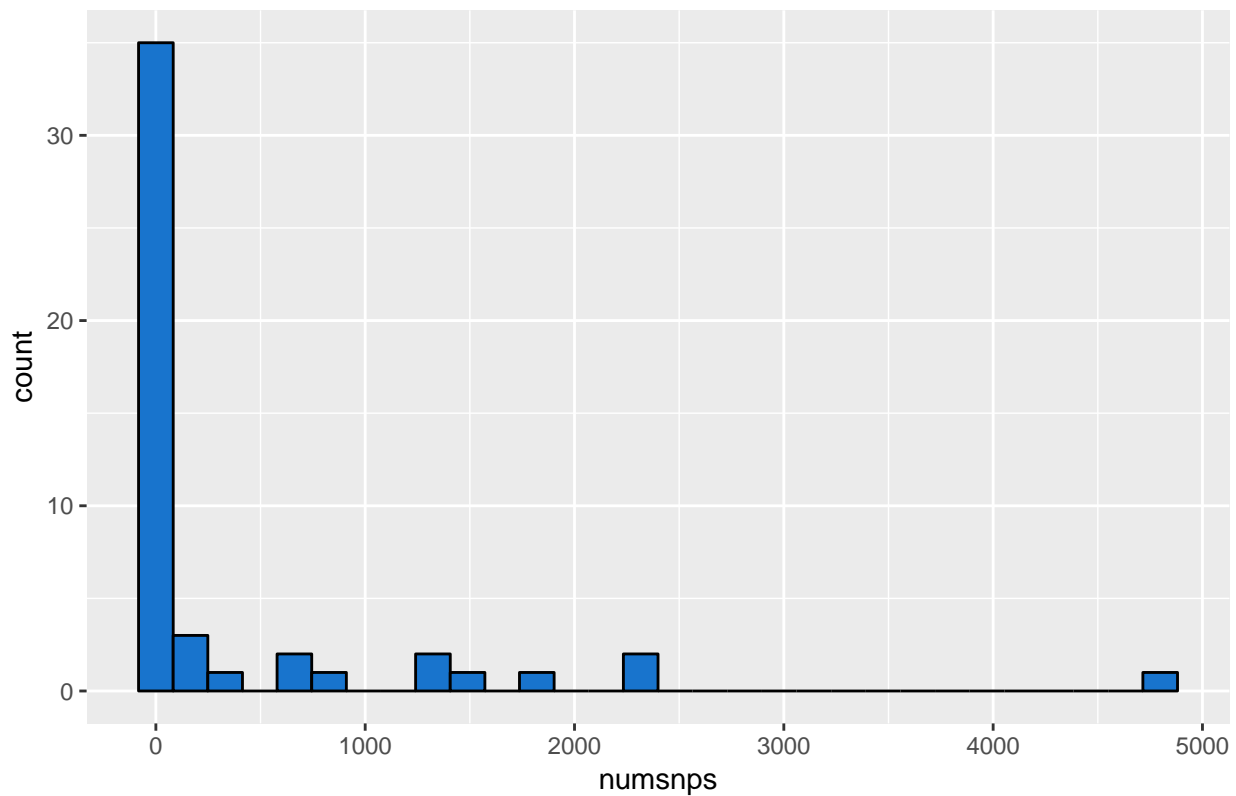
```

plt1 <- ggplot(data=loc.df) +
  geom_histogram(aes(x=numsnps), color="black",
    fill="dodgerblue3") + ggtitle("Number of Variants per Locus");plt1

## `stat_bin()` using `bins = 30`. Pick better value with `binwidth`.

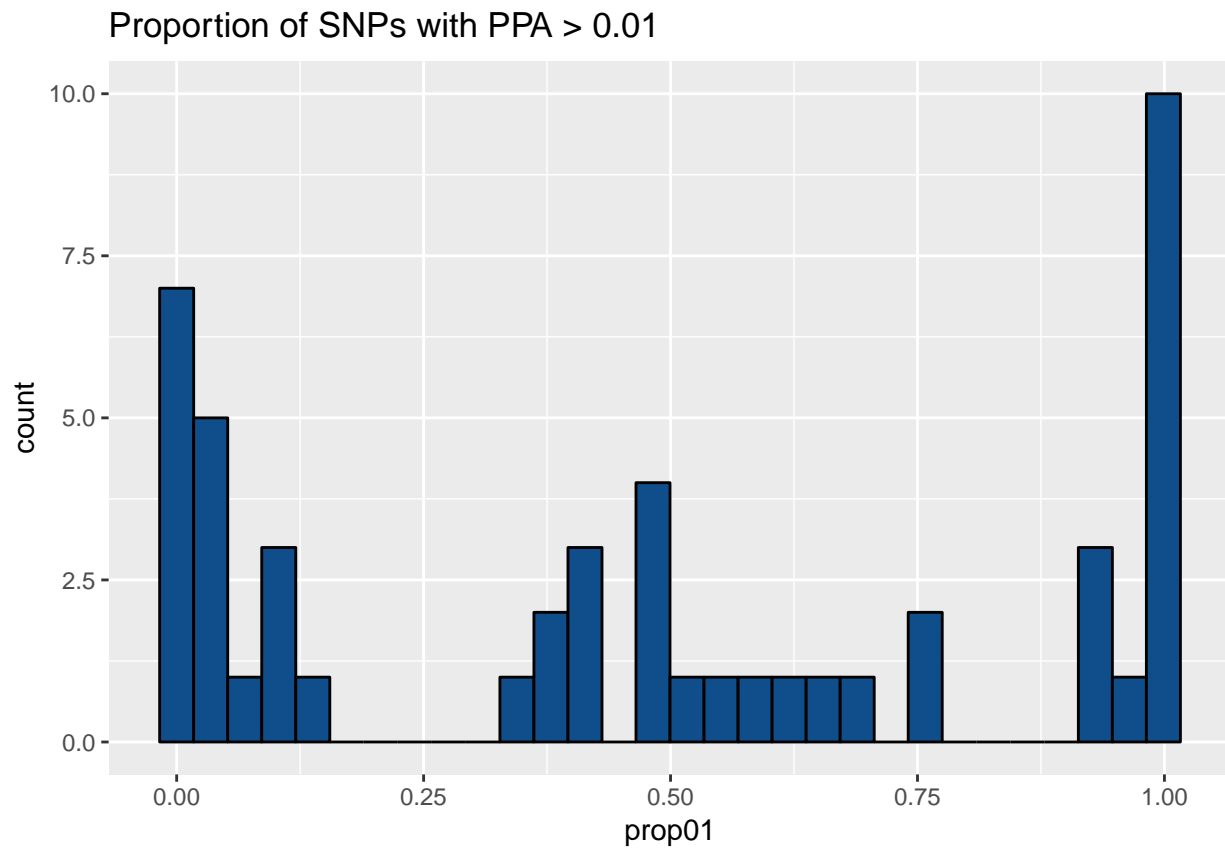
```

Number of Variants per Locus



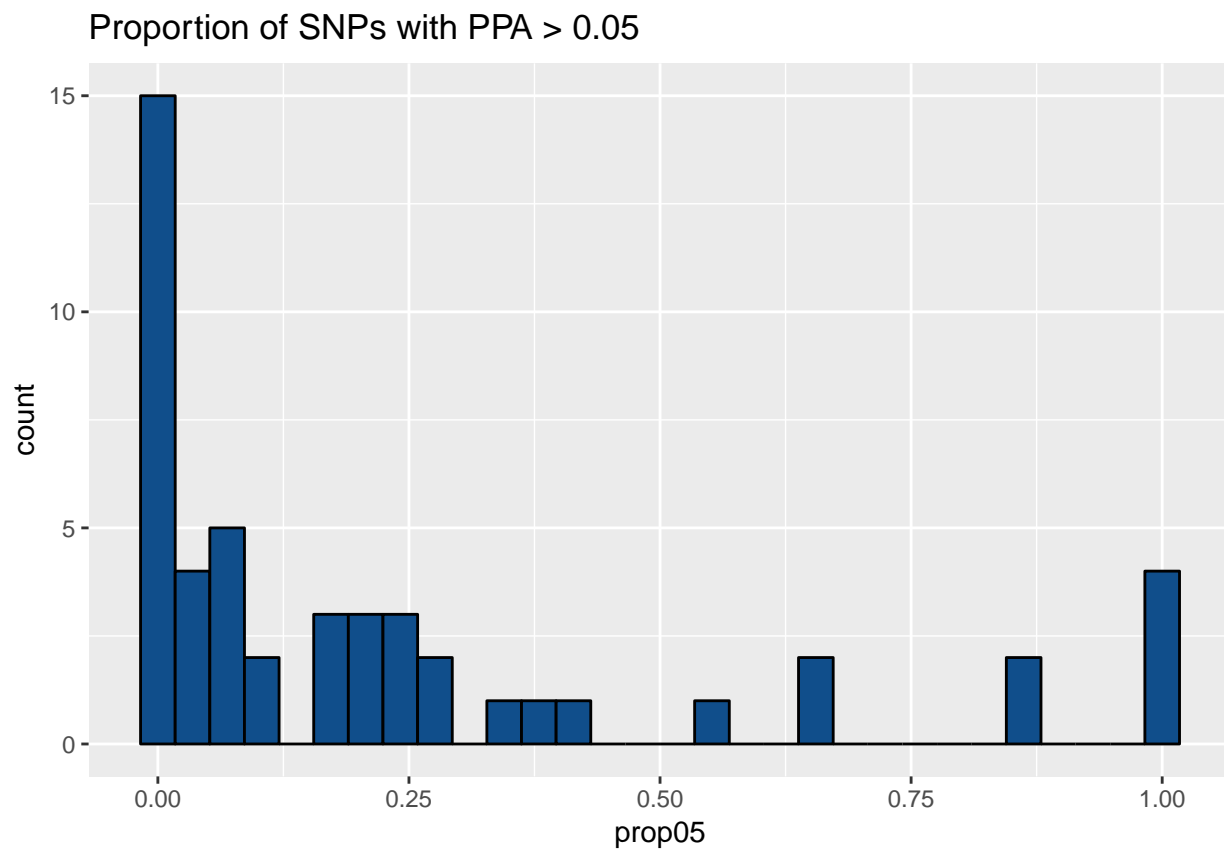
```
plt2 <- ggplot(data=loc.df) +  
  geom_histogram(aes(x=prop01),color="black",  
    fill="dodgerblue4") + ggtitle("Proportion of SNPs with PPA > 0.01");plt2
```

```
## `stat_bin()` using `bins = 30`. Pick better value with `binwidth`.
```



```
plt3 <- ggplot(data=loc.df) +  
  geom_histogram(aes(x=prop05),color="black",  
    fill="dodgerblue4") + ggtitle("Proportion of SNPs with PPA > 0.05");plt3
```

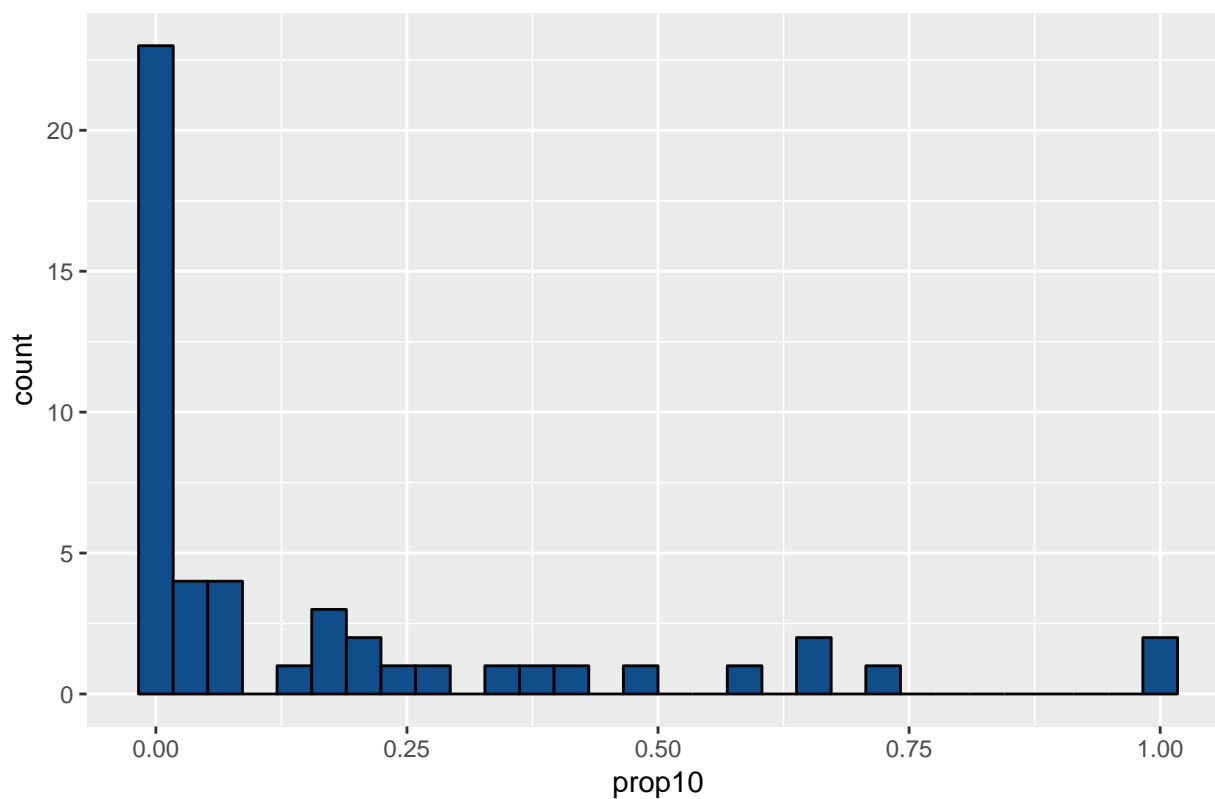
```
## `stat_bin()` using `bins = 30`. Pick better value with `binwidth`.
```



```
plt4 <- ggplot(data=loc.df) +  
  geom_histogram(aes(x=prop10),color="black",  
    fill="dodgerblue4") + ggtitle("Proportion of SNPs with PPA > 0.10");plt4
```

```
## `stat_bin()` using `bins = 30`. Pick better value with `binwidth`.
```

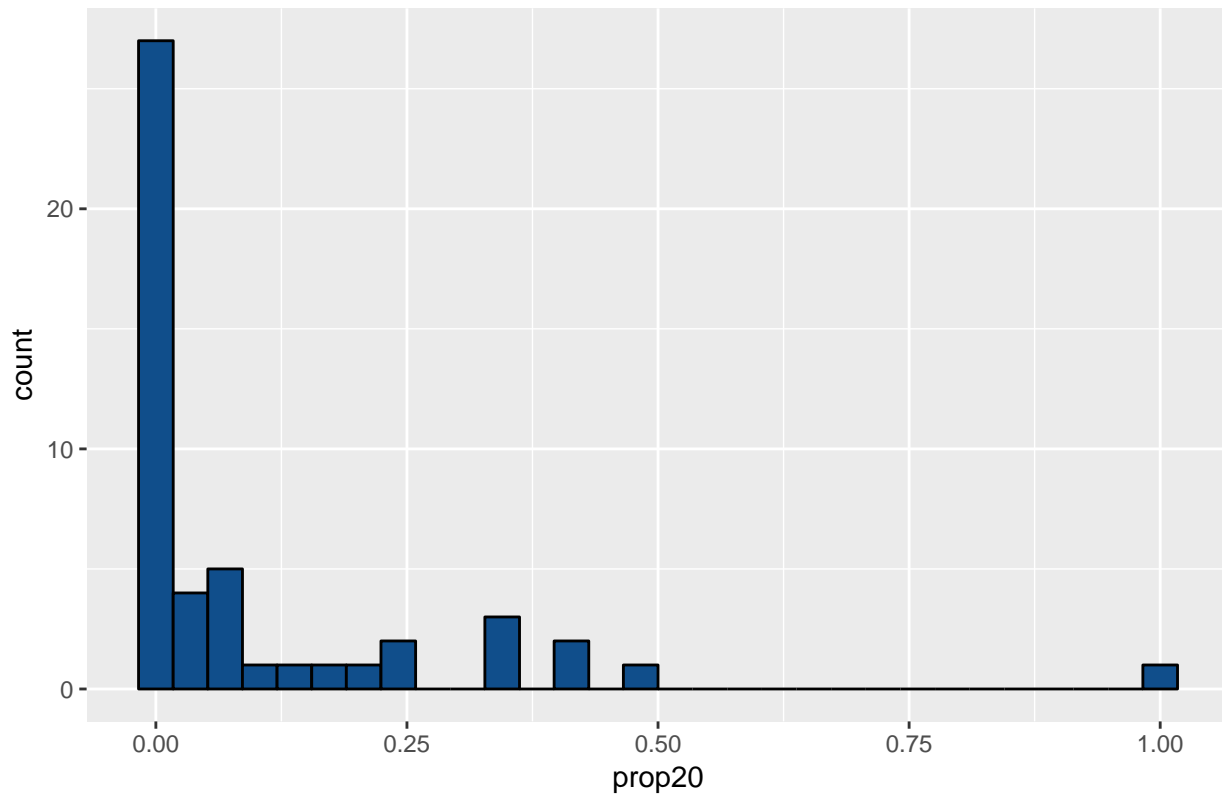
Proportion of SNPs with PPA > 0.10



```
plt5 <- ggplot(data=loc.df) +  
  geom_histogram(aes(x=prop20),color="black",  
    fill="dodgerblue4") + ggtitle("Proportion of SNPs with PPA > 0.20");plt5
```

```
## `stat_bin()` using `bins = 30`. Pick better value with `binwidth`.
```

## Proportion of SNPs with PPA > 0.20



```
loc.df <- arrange(loc.df, desc(prop01))
summary(loc.df)
```

```
##      loci      numsnps      prop01      prop05
## Length:49      Min.   : 1.0      Min.   :0.001042      Min.   :0.000000
## Class :character 1st Qu.: 13.0      1st Qu.:0.058824      1st Qu.:0.004905
## Mode  :character Median : 27.0      Median :0.476191      Median :0.100000
##                Mean  : 393.2      Mean  :0.488629      Mean  :0.241847
##                3rd Qu.: 171.0      3rd Qu.:0.923077      3rd Qu.:0.259259
##                Max.  :4799.0      Max.  :1.000000      Max.  :1.000000
##      prop10      prop20
## Min.   :0.00000      Min.   :0.000000
## 1st Qu.:0.00000      1st Qu.:0.000000
## Median :0.03704      Median :0.002941
## Mean   :0.16971      Mean   :0.100857
## 3rd Qu.:0.21053      3rd Qu.:0.105263
## Max.   :1.00000      Max.   :1.000000
```

```
loc.df
```

```
##      loci numsnps      prop01      prop05      prop10
## 1      TCF7L2      3 1.000000000 1.000000000 1.000000000
## 2      KCNQ1.rs74046911 3 1.000000000 1.000000000 0.666666667
## 3      MTNR1B      1 1.000000000 1.000000000 1.000000000
## 4      HNF1B      7 1.000000000 0.8571428571 0.7142857143
## 5      PPARG      27 1.000000000 0.2222222222 0.0000000000
## 6      ZBED3      5 1.000000000 0.4000000000 0.4000000000
## 7      CDKAL1      8 1.000000000 0.8750000000 0.5000000000
```

## 8	SLC30A8	6	1.000000000	0.666666667	0.666666667
## 9	CDKN2B.rs10811660	6	1.000000000	0.666666667	0.333333333
## 10	CDKN2B.rs10757283	5	1.000000000	1.000000000	0.600000000
## 11	ADAMTS9	27	0.962962963	0.2592592593	0.000000000
## 12	ADCY5	17	0.941176471	0.3529411765	0.1764705882
## 13	GCKR	13	0.923076923	0.5384615385	0.3846153846
## 14	CDC123	12	0.916666667	0.2500000000	0.2500000000
## 15	HHEX	40	0.775000000	0.1000000000	0.000000000
## 16	IGF2BP2	50	0.760000000	0.0400000000	0.000000000
## 17	GLIS3	10	0.700000000	0.2000000000	0.200000000
## 18	GRB14	24	0.666666667	0.2500000000	0.083333333
## 19	CENTD2	27	0.629629630	0.2592592593	0.1481481481
## 20	KCNQ1.chr11_2692322	12	0.583333333	0.166666667	0.166666667
## 21	JAZF1	16	0.562500000	0.1875000000	0.187500000
## 22	WFS1	82	0.524390244	0.0365853659	0.000000000
## 23	IRS1	65	0.492307692	0.0153846154	0.000000000
## 24	CILP2	29	0.482758621	0.0689655172	0.0689655172
## 25	KCNJ11	21	0.476190476	0.3809523810	0.2857142857
## 26	PRC1	51	0.470588235	0.1568627451	0.000000000
## 27	PROX1	19	0.421052632	0.2105263158	0.2105263158
## 28	HMGA2	72	0.416666667	0.0555555556	0.000000000
## 29	BCL11A	35	0.400000000	0.2571428571	0.0285714286
## 30	NOTCH2	108	0.388888889	0.000000000	0.000000000
## 31	HNF1A.rs1169288	27	0.370370370	0.0740740741	0.0370370370
## 32	FTO	72	0.361111111	0.000000000	0.000000000
## 33	DGKB.rs1974620	266	0.124060150	0.000000000	0.000000000
## 34	TSPAN8	76	0.118421053	0.1052631579	0.0394736842
## 35	GCK	18	0.111111111	0.0555555556	0.0555555556
## 36	KLF14	171	0.111111111	0.0058479532	0.000000000
## 37	HNF4A	17	0.058823529	0.0588235294	0.0588235294
## 38	GIPR.rs2238689	26	0.038461538	0.0384615385	0.0384615385
## 39	THADA	247	0.036437247	0.0242914980	0.0080971660
## 40	GIPR.rs4399645	704	0.031250000	0.0028409091	0.0014204545
## 41	HNF1A.rs1800574	899	0.027808676	0.0022246941	0.0022246941
## 42	MC4R.rs17066842	1275	0.025098039	0.000000000	0.000000000
## 43	HNF1A.chr12_121440833	1427	0.011212334	0.0049053959	0.000000000
## 44	C2CD4B	1851	0.008103728	0.000000000	0.000000000
## 45	KCNQ1.rs2237895	680	0.007352941	0.0029411765	0.0029411765
## 46	KCNQ1.rs2283220	2258	0.003985828	0.0004428698	0.000000000
## 47	KCNQ1.rs458069	2309	0.001732352	0.0004330879	0.000000000
## 48	MC4R.chr18_57739289	1343	0.001489203	0.000000000	0.000000000
## 49	DGKB.rs10276674	4799	0.001041884	0.0006251302	0.0004167535
##	prop20				
## 1	0.333333333				
## 2	0.333333333				
## 3	1.000000000				
## 4	0.142857143				
## 5	0.000000000				
## 6	0.400000000				
## 7	0.250000000				
## 8	0.500000000				
## 9	0.333333333				
## 10	0.400000000				
## 11	0.000000000				



```
## 12 0.058823529
## 13 0.076923077
## 14 0.250000000
## 15 0.000000000
## 16 0.000000000
## 17 0.200000000
## 18 0.041666667
## 19 0.000000000
## 20 0.083333333
## 21 0.187500000
## 22 0.000000000
## 23 0.000000000
## 24 0.034482759
## 25 0.000000000
## 26 0.000000000
## 27 0.105263158
## 28 0.000000000
## 29 0.000000000
## 30 0.000000000
## 31 0.037037037
## 32 0.000000000
## 33 0.000000000
## 34 0.013157895
## 35 0.055555556
## 36 0.000000000
## 37 0.058823529
## 38 0.038461538
## 39 0.004048583
## 40 0.000000000
## 41 0.001112347
## 42 0.000000000
## 43 0.000000000
## 44 0.000000000
## 45 0.002941176
## 46 0.000000000
## 47 0.000000000
## 48 0.000000000
## 49 0.000000000
```

```
write.table(df,cred.dir%>%>"metabohip.chr_added.txt",row.names=FALSE,
            sep="\t",quote=FALSE)
write.table(loc.df,cred.dir%>%>"metabohip.chr_added.locusSummary.txt",row.names=FALSE,
            sep="\t",quote=FALSE)
```