

variantBedOverlap

Leland Taylor

2018-03-12

This document shows a demo of how to use variantBedOverlap. There is also a command line script.

```
# get the lib dir for variantBedOverlap
install_dir=$(R --slave -e 'cat(find.package("variantBedOverlap"))')

# see help options of command line script
Rscript "$install_dir/exec/variant_bed_overlap.R" --help
```

(1) Get variants in LD

Get proxies from the 1000 Genomes Project via proxysnps.

```
snps_q <- proxysnps::get_proxies(query = "rs2072014", pop = "FIN")
snps <- subset(snps_q, R.squared >= 0.8)

#knitr::kable( head(snps, 10) )
snps # enabled by setting --> df_print: paged
#> # A tibble: 8 x 9
#>   CHROM      POS ID      REF  ALT      MAF R.squared D.prime CHOSEN
#> * <int>    <int> <chr>    <chr> <chr>    <dbl>    <dbl>    <dbl> <lgl>
#> 1     22 26231688 rs2072014  G     A      0.343     1.00      1. TRUE
#> 2     22 26228757 rs35045598 TA     T      0.348     0.978    -1. FALSE
#> 3     22 26227039 rs13056293 T      C      0.364     0.915    -1. FALSE
#> 4     22 26228540 rs6004788  T      C      0.364     0.915    -1. FALSE
#> 5     22 26226815 rs12166250 A      G      0.369     0.896    -1. FALSE
#> 6     22 26226935 rs12166358 A      G      0.374     0.877    -1. FALSE
#> 7     22 26227108 rs13056441 C      T      0.374     0.877    -1. FALSE
#> 8     22 26228281 rs9624913  A      G      0.374     0.877    -1. FALSE
```

(2) Get BED overlaps

Get the overlaps of each variant (row) and genomic regions from a list of BED files. Here we load example BED files included in this package taken from Varsheny et al. 2017 (<https://doi.org/10.1073/pnas.1621192114>).

```
# few pre-packaged bed files from
# https://theparkerlab.med.umich.edu/data/papers/doi/10.1073/pnas.1621192114/
dir <- system.file("extdata", package = "variantBedOverlap", mustWork = TRUE)

# get overlaps with all bed files in directory
snps_overlap <- variantBedOverlap::get_bed_overlaps(
  df = snps,
  dir = dir,
  col_itemRgb = 5
)
```

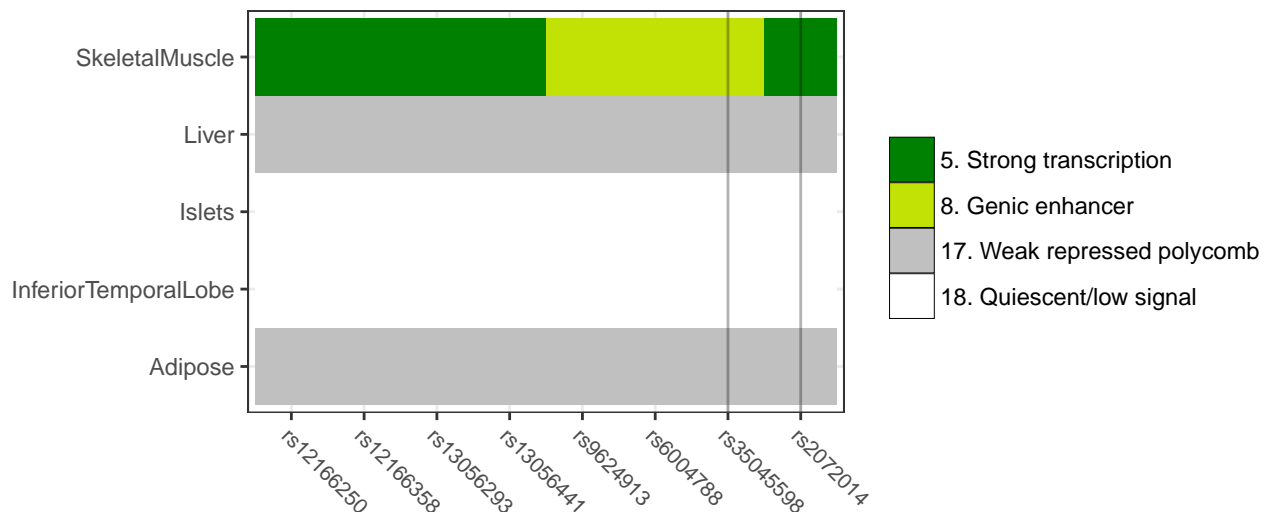
processing file: Adipose processing file: InferiorTemporalLobe processing file: Islets processing file: Liver
processing file: SkeletalMuscle

```
snps_overlap
#> # A tibble: 8 x 14
#>   ID          CHROM      POS REF  ALT    MAF R.squared D.prime CHOSEN
#>   <chr>        <int>    <int> <chr> <chr> <dbl>   <dbl>   <dbl> <lgl>
#> 1 rs12166250    22 26226815 A    G    0.369   0.896   -1. FALSE
#> 2 rs12166358    22 26226935 A    G    0.374   0.877   -1. FALSE
#> 3 rs13056293    22 26227039 T    C    0.364   0.915   -1. FALSE
#> 4 rs13056441    22 26227108 C    T    0.374   0.877   -1. FALSE
#> 5 rs2072014     22 26231688 G    A    0.343   1.00    1. TRUE
#> 6 rs35045598    22 26228757 TA   T    0.348   0.978   -1. FALSE
#> 7 rs6004788     22 26228540 T    C    0.364   0.915   -1. FALSE
#> 8 rs9624913     22 26228281 A    G    0.374   0.877   -1. FALSE
#> # ... with 5 more variables: bed.Adipose <chr>,
#> #   bed.InferiorTemporalLobe <chr>, bed.Islets <chr>, bed.Liver <chr>,
#> #   bed.SkeletalMuscle <chr>
```

(3) Plot the data

Plot the overlap data.

```
# xid_solid_line = list of x-axis IDs to add a line through
# varshney_chrhmm = flag to say assume BED file names are from Varshney et al
#                   2017. Given that assumption clean up the names to make them
#                   publication ready.
lst <- variantBedOverlap::plot_overlaps(
  df = snps_overlap,
  xid_solid_line = c("rs2072014", "rs35045598"),
  varshney_chrhmm = TRUE
)
print( lst$plt )
```



The output of `plot_overlaps` also contains the data underlying the plot. Note that `ID` is a factor now, sorted by `POS`. If `varshney_chrhmm == TRUE`, `bed_feature` will also be a factor sorted by `chrhmm_state`.

```
lst$df
#> # A tibble: 40 x 6
#>   ID          POS bed_file          bed_feature color chrhmm_state
```

```

#>      <fct>          <int> <chr>          <fct>      <chr>          <dbl>
#> 1 rs12166250 26226815 Adipose      17. Weak r~ #COC~      17.
#> 2 rs12166358 26226935 Adipose      17. Weak r~ #COC~      17.
#> 3 rs13056293 26227039 Adipose      17. Weak r~ #COC~      17.
#> 4 rs13056441 26227108 Adipose      17. Weak r~ #COC~      17.
#> 5 rs2072014  26231688 Adipose      17. Weak r~ #COC~      17.
#> 6 rs35045598 26228757 Adipose      17. Weak r~ #COC~      17.
#> 7 rs6004788  26228540 Adipose      17. Weak r~ #COC~      17.
#> 8 rs9624913  26228281 Adipose      17. Weak r~ #COC~      17.
#> 9 rs12166250 26226815 InferiorTemporalLobe 18. Quiesc~ #FFF~      18.
#> 10 rs12166358 26226935 InferiorTemporalLobe 18. Quiesc~ #FFF~      18.
#> # ... with 30 more rows

```