

# Package ‘echotabix’

August 31, 2021

**Type** Package

**Title** echoverse module: Tabix indexing and querying.

**Version** 0.99.0

**Description** echoverse module: Tabix indexing and querying.

**URL** <https://github.com/RajLabMSSM/echotabix>

**BugReports** <https://github.com/RajLabMSSM/echotabix/issues>

**Encoding** UTF-8

**LazyData** true

**Depends** R (>= 3.6.0)

**SystemRequirements** Python (>= 3.7.0)

**biocViews**

**Imports** magrittr,  
dplyr,  
utils,  
R.utils,  
stats,  
methods,  
Matrix,  
parallel,  
BiocManager,  
data.table,  
GenomeInfoDb,  
IRanges,  
GenomicRanges,  
VariantAnnotation,  
Rsamtools,  
seqminer,  
rtracklayer

**Suggests** markdown,  
rmarkdown,  
remotes,  
knitr,  
BiocStyle,  
covr,  
testthat (>= 3.0.0)

**RoxygenNote** 7.1.1  
**VignetteBuilder** knitr  
**License** GPL (>= 3) + file LICENSE  
**Config/testthat/edition** 3

**R topics documented:**

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BST1	<b>echolocatoR</b> output example: <i>BST1</i> locus
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**Description**

An example results file after running `finemap_loci` on the *BST1* locus.

**Usage**

```
data("BST1")
```

**Format**

data.table  
**SNP** SNP RSID  
**CHR** Chromosome  
**POS** Genomic position (in basepairs)  
... Optional: extra columns  
**Nalls2019**  
data.table

**Details**

Data originally comes from the Parkinson’s disease GWAS by [Nalls et al., \(bioRxiv\)](#).

**Source**

```
root_dir <- "~/Desktop/Fine_Mapping/Data/GWAS/Nalls23andMe_2019/BST1/Multi-finemap"  
BST1 <- data.table::fread(file.path(root_dir, "Multi-finemap_results.txt"))  
BST1 <- update_cols(dat = BST1)  
BST1 <- find_consensus_SNPs(dat = BST1) usethis::use_data(BST1, overwrite = TRUE)
```

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convert	<i>Convert summary stats file to tabix format</i>
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### Description

Convert summary stats file to tabix format

### Usage

```
convert(fullSS_path, chrom_col = "CHR", position_col = "POS", verbose = TRUE)
```

### See Also

Other tabix: [convert\\_and\\_query\(\)](#)

### Examples

```
## Not run:
fullSS_path <- echolocator::example_fullSS()
fullSS_tabix <- convert(fullSS_path = fullSS_path, position_col = "BP")

## End(Not run)
```

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convert_and_query	<i>Convert and query</i>
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### Description

If it is not tabix format already (determined by checking for a [.tbi](#) file of the same name in the same directory), the full summary statistics file is converted into tabix format for super fast querying. A query is then made using the min/max genomic positions to extract a locus-specific summary stats file.

### Usage

```
convert_and_query(
  fullSS_path,
  study_dir = NULL,
  subset_path = tempfile(".tsv.gz"),
  is_tabix = FALSE,
  chrom_col = "CHR",
  position_col = "BP",
  min_POS,
  max_POS,
  chrom,
  save_subset = TRUE,
  nThread = 1,
  conda_env = "echoR",
  verbose = TRUE
)
```

**Value**

data.table of locus subset summary statistics

**See Also**

Other tabix: [convert\(\)](#)

**Examples**

```
## Not run:
data("BST1")
fullSS_path <- echolocator::example_fullSS()

subset_path <- file.path(tempdir(), "BST1_Nalls23andMe_2019_subset.tsv.gz")
dat <- convert_and_query(
  fullSS_path = fullSS_path,
  subset_path = subset_path,
  min_POS = min(BST1$POS),
  max_POS = max(BST1$POS),
  chrom = BST1$CHR[1]
)

## End(Not run)
```

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liftover

*Genome build liftover*


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**Description**

Transfer genomic coordinates from one genome build to another.

**Usage**

```
liftover(
  dat,
  chrom_col = "CHR",
  start_col = "POS",
  end_col = start_col,
  build_conversion = c("hg38ToHg19", "hg19ToHg38"),
  as_granges = FALSE,
  verbose = TRUE
)
```

**Arguments**

dat	SNP-level data table.
chrom_col	Name of the chromosome column.
start_col	Name of the start position column.
end_col	Name of the end position column (can be same as start_col if all data is SNP-level).

build\_conversion      From which to which genome build to lift over dat.  
 as\_granges            Return lifted dat as [GenomicRanges](#) object.  
 verbose                Print messages.

### Source

[liftOver](#)  
[UCSC chain files](#)

### Examples

```
data("BST1")
dat_lifted <- liftover(dat = BST1, build_conversion = "hg19ToHg38")
```

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locus_dir	<i>Example results path for BST1 locus</i>
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### Description

Example results path for BST1 locus

### Usage

```
data("locus_dir")
```

### Format

path string

### Source

```
locus_dir <- "results/GWAS/Nalls23andMe_2019/BST1" usethis::use_data(locus_dir, overwrite = T)
```

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query_tabular	<i>Query a tabix file</i>
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### Description

Query by genomic coordinates.

### Usage

```
query_tabular(fullSS_tabix, chrom, start_pos, end_pos, verbose = TRUE)
```

Examples

```
## Not run:
data("BST1")
fullSS_path <- echolocator::example_fullSS()
fullSS_tabix <- convert(fullSS_path = fullSS_path, position_col = "BP")
tab <- query_tabular(
  fullSS_tabix = fullSS_tabix,
  chrom = BST1$CHR[1],
  start_pos = min(BST1$POS),
  end_pos = max(BST1$POS)
)

## End(Not run)
```

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query_vcf	<i>Query VCF file</i>
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Description

Query a (remote) Variant Call Format (VCF) file.

Usage

```
query_vcf(
  dat,
  vcf_url,
  locus_dir = file.path(tempdir(), "LD"),
  vcf_name = gsub(".vcf|.gz|.bgz", "", basename(vcf_url)),
  ref_genome = "GRCh37",
  samples = NULL,
  force_new_vcf = FALSE,
  verbose = TRUE
)
```

Arguments

dat	SNP-level data table.
vcf_url	URL or path to VCF.
locus_dir	Directory to store LD in.
vcf_name	VCF reference name (e.g. "1KGphase1").
ref_genome	Genome build of the VCF file.
samples	Sample names to subset the VCF by before computing LD.
force_new_vcf	Force the creation of a new LD file even if one exists.
verbose	Print messages.

Value

VCF object.

See Also

Other LD: [construct\\_subset\\_vcf\\_name\(\)](#), [get\\_locus\\_vcf\\_folder\(\)](#)

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