

# Package ‘gQTLBase’

July 30, 2020

**Title** gQTLBase: infrastructure for eQTL, mQTL and similar studies

**Version** 1.21.1

**Author** VJ Carey <stvjc@channing.harvard.edu>

**Description** Infrastructure for eQTL, mQTL and similar studies.

**Suggests** geuvStore2, knitr, rmarkdown, BiocStyle, RUnit, Homo.sapiens, IRanges, erma, GenomeInfoDb, gwascat, geuvPack

**Imports** GenomicRanges, methods, BatchJobs, BBmisc, S4Vectors, BiocGenerics, foreach, doParallel, bit, ff, rtracklayer, ffbase, GenomicFiles, SummarizedExperiment

**Depends**

**Maintainer** VJ Carey <stvjc@channing.harvard.edu>

**License** Artistic-2.0

**LazyLoad** yes

**VignetteBuilder** knitr

**BiocViews** SNP, GenomeAnnotation, Genetics, DataImport, FunctionalGenomics

**Collate** storeS4.R cb2range.R ffapp2.R gtpath.R storeFuncs.R mergeToLoci.R ufeatByTiling.R d.R

**git\_url** <https://git.bioconductor.org/packages/gQTLBase>

**git\_branch** master

**git\_last\_commit** 7eb24a5

**git\_last\_commit\_date** 2020-06-30

**Date/Publication** 2020-07-29

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gQTLBase-package	<i>gQTLBase: infrastructure for eQTL, mQTL and similar studies</i>
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**Description**

Infrastructure for eQTL, mQTL and similar studies.

**Details**

The DESCRIPTION file: This package was not yet installed at build time.

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Purpose is to define infrastructure on a comprehensive archive of eQTL, mQTL, dsQTL, etc., association statistics.

Package will complement gQTLStats. geuvStore2 is a basic illustration relative to GEUVADIS paper.

`matprint` is exported from package ff.

**Author(s)**

VJ Carey <stvjc@channing.harvard.edu>

Maintainer: VJ Carey <stvjc@channing.harvard.edu>

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ciseStore-class	<i>Class "ciseStore"</i>
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**Description**

wrap a BatchJobs registry that manages results of a cis-eQTL search

**Objects from the Class**

Objects can be created by calls of the form `new("ciseStore", reg=reg, ...)`. All arguments must be named.

We can also use `ciseStore(reg, validJobs, addProbeMap = TRUE, addRangeMap = TRUE)` and the `probemap` and `rangeMap` slots will be populated appropriately. If `validJobs` is missing, the `validJobs` slot will be populated by `findDone(reg)`. This may be problematic for handcrafted extracts from archives.

**Slots**

`reg`: Object of class "Registry" BatchJobs Registry instance

`validJobs`: Object of class "integer" vector of valid job identifiers for the registry

`probemap`: Object of class "data.frame" a map from expression probe identifiers to job identifiers where results for the probe are stored

`rangeMap`: Object of class "GRanges" a map from ranges on chromosomes, to job identifiers, in `mcols()$jobid`

**Methods**

show

Function describeStore uses batchMapResults and reduceResults to leverage a parallel environment to collect information on numbers of tests and features. Arguments are described in the associated man page.

**Note**

the construction of the maps occurs via [storeApply](#), which

will use [foreach](#), so that registration of a parallel back end using, e.g., [registerDoParallel](#), will determine the speed of construction

Any registry job results that do not inherit from GRanges are mapped to NULL and will not be present in ultimate maps.

**Examples**

```
showClass("ciseStore")
# get the global assignment back
require(BatchJobs)
if (require(geuvStore2)) {
  store = makeGeuvStore2()
  store
}
```

---

describeStore	<i>collect basic descriptive statistics on ciseStore instances</i>
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---

**Description**

collect basic descriptive statistics on ciseStore instances

**Usage**

```
describeStore(st, genetag = "probeid", snptag = "snp", ids = NULL,
              resfilter = force, doChecks = TRUE, ...)
describeByFilts(st, filtlist, ...)
```

**Arguments**

st	instance of <a href="#">ciseStore-class</a>
genetag	string for field name for name of quantitatively assayed feature, defaults to "probeid"; for GTEx application "gene" is used
snptag	string for field name for name of genotype feature
ids	integerish vector of ids, can be left NULL to survey entire store
resfilter	function applied to job results prior to summarization, defaults to force()
filtlist	a list of functions suitable as resfilter arguments
doChecks	logical – if true, will collect information on match between number probes requested and number reported on, and two scans of VCF loci in cis to probes. See details.
...	used with describeByFilts, pass to storeApply

**Details**

uses parallel infrastructure of foreach on contents managed by st@reg

describeByFilt returns a matrix of descriptions with one row per filtlist element

storeDescription holds results of a describe task and includes information on noncongruence of features with cis tests and of results of two distinct scans of VCF: one with readGT on a single sample, the other with readVcf on all samples. If there are discrepancies between features given and tests returned, [storeDescription]@reqfail will give the job ids for these. If there are discrepancies between the numbers of loci retrieved on the two VCF scans, @locfail will give the job ids for these. @reqfail events may be legitimate when a feature has no SNP in cis at the given radius. @locfail events usually indicate an I/O problem and the jobs should be resubmitted.

**Value**

list with elements ntests, ngene.uniq, nsnp.uniq

**Examples**

```
## Not run:
library(geuvStore2)
mm = makeGeuvStore2()
describeStore(mm, ids=1:10, resfilter=function(x) x[x$mindist < 50000])

## End(Not run)
```

---

extractByProbes	<i>retrieve eqtlTest results from a ciseStore instance</i>
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**Description**

retrieve eqtlTest results from a ciseStore instance

**Usage**

```
extractByProbes(store, probeids, extractTag = "probeid")
extractByRanges(store, gr)
extractBySymbols(store, symbols, sym2probe, extractTag = "probeid")
```

**Arguments**

store	instance of <a href="#">ciseStore-class</a>
probeids, symbols	vector character tokens
gr	instance of <a href="#">GRanges-class</a>
sym2probe	named character vector of probeids with names given by corresponding symbols
extractTag	character atom telling what field in the archived GRanges is regarded as the probe or gene identifier

**Details**

an index will be searched if created by the ciseStore constructor

**Value**

a GRanges instance

**Author(s)**

VJ Carey <stvjc@channing.harvard.edu>

**Examples**

```
if (require(geuvStore2)) {
  store = makeGeuvStore2()
  ebp = extractByProbes(store, c("ENSG00000183814.10", "ENSG00000174827.9"))
  ebp
  rr = range(ebp)
  ebr = extractByRanges(store, rr)
  ebr
  s2p = structure(c("ENSG00000183814.10", "ENSG00000163207.5", "ENSG00000228449.1",
"ENSG00000137962.8", "ENSG00000232848.1", "ENSG00000227280.1",
"ENSG00000238081.1", "ENSG00000117480.10", "ENSG00000253368.2",
"ENSG00000174827.9"), .Names = c("LIN9", "IVL", "RP11-177A2.4",
"ARHGAP29", "CTA-215D11.4", "RP11-458D21.2", "RP4-620F22.3",
"FAAH", "TRNP1", "PDZK1"))
  ss = extractBySymbols(store, c("IVL", "FAAH", "PDZK1"), s2p)
  ss
}
```

---

mergeCIstates	<i>merge ChromImpute chromatin states, or GWAS hit indicators, to a GRanges</i>
---------------	---

---

**Description**

merge ChromImpute chromatin states, or GWAS hit indicators, to a GRanges

**Usage**

```
mergeCIstates(gr, ermaset=NULL, epig, genome = "hg19", importFull=FALSE, useErma = TRUE, stateGR=NULL)
mergeGWhits(gr, gwcat, use=c("both", "addr", "name")[1],
  grSnpField="SNP")
```

**Arguments**

gr	a GRanges instance
ermaset	an instance of <a href="#">ErmaSet-class</a> . if NULL, supply a GRanges as stateGR, with fields states and statecols
gwcat	an instance of <a href="#">gwaswloc-class</a> , or any compliant GRanges instance – must have mcols field SNPS with snp identifier
epig	the standardized epigenome name of the epigenome to use
genome	a tag for genome build
importFull	logical, set to TRUE to acquire entire content (for LNG.FET, 800K ranges), to avoid contention for connections in parallel applications

useErma	logical – at the moment, must be TRUE; plan is to allow use of elements of AnnotationHub
use	character string selecting approach for linking loci in gr to those in gwcat – if "both", coincidence in address or name are both checked and used; if "addr", only address is checked, if "name", only SNP name.
grSnpField	character string naming the field in mcols(gr) with SNP id
stateGR	a GRanges instance as imported from erma package or from AnnotationHub, with mcols field states denoting chromatin state and statecols the associated colors for rendering

### Value

for mergeCIstates, a GRanges instance with additional fields in mcols: fullStates, states, and statecols, denoting respectively the full annotation of ChromImpute for the inferred state, an abbreviated tag that collapses related states, and a color tag for rendering, that does not replicate the colors in the ChromImpute bed files. The states field is a factor with levels c("Het", "DNase", "Enh", "Prom", "Quies", "ReprPC", "

for mergeGWhits, a single mcols field is added, isGwasHit, that is 1 for coincident hit and 0 otherwise. Eventually phenotype information will be collected and added.

### Examples

```
## Not run: # needs to use updated gwascats
if (require(gwascats) && require(erma)) {
#
# demonstrate Tx state for exon starts
#
gm = resize(genemodel("ORMDL3"),1)
es = makeErmaSet()
g1 = mergeCIstates(gm, es, "LNG.FET")
g1
#
# set up for GWAS
#
require(GenomeInfoDb)
data(ebicat37)
genome(ebicat37) = "hg19"
seqlevelsStyle(ebicat37) = "UCSC"
g1 = c(g1, g1[1]) # add a known hit
start(g1[length(g1)]) = 38062196
mergeGWhits(g1, ebicat37)
}

## End(Not run)
```

---

storeApply

*apply a function over job results in a ciseStore instance*

---

### Description

apply a function over job results in a ciseStore instance

**Usage**

```
storeApply(store, f, n.chunks, ids=NULL, ..., verbose = FALSE, flatten1=TRUE)
```

**Arguments**

store	instance of <a href="#">ciseStore-class</a>
f	function on GRanges stored in ciseStore
n.chunks	Number of chunks into which the jobs are to be broken; the series of chunks is handed to <a href="#">foreach</a> to extract results and apply f to them. If missing, the value of <code>getDoParWorkers()</code> used.
ids	defaults to NULL; if non-null, the jobs to be processed are limited to those identified in this vector.
...	additional arguments to foreach
verbose	if TRUE will allow progressbars and other messages to display
flatten1	if TRUE will execute <code>unlist(...,recursive=FALSE)</code> on output, defaulted to FALSE in previous version

**Details**

The chunking of job identifiers will determine the degree of parallelization of application, and the form of the list that is returned. `flatten1` will eventually default to TRUE.

**Value**

A list whose structure depends on the chunking of job identifiers. See the examples.

**Note**

`eqtlStore` imports `BiocParallel`'s `bpparam` function, and this determines in real time the number of workers to be employed by `storeApply`.

**See Also**

[storeMapResults](#) will apply over the store using the batch jobs submission infrastructure and can target specific results via `ids`; `storeApply` uses `bplapply` over the entire store

**Examples**

```
if (require(geuvStore2)) {
  require(BatchJobs)
  store = makeGeuvStore2()
  storeApply(store, length)
  storeApply(store, length, ids=c(1:3,603))
}
```

---

storeMapResults	<i>use batchMapResults infrastructure to process results in a ciseStore instance</i>
-----------------	--

---

### Description

use batchMapResults infrastructure to process results in a ciseStore instance

### Usage

```
storeMapResults(store, reg2, fun, ...,
  ids = NULL, part = NA_character_, more.args = list())
loadAndFilterResult(reg,
  id, filter=force, part = NA_character_, missing.ok = FALSE)
```

### Arguments

store	an instance of <a href="#">ciseStore-class</a>
reg	instance of BatchJobs Registry class
reg2	an empty instance of the Registry class (see <a href="#">makeRegistry</a> )
fun	A function to map over results in store, with formals (job, res, ...).
filter	a function that accepts and returns a GRanges instance, to be applied just after loading a result from the store
...	additional arguments to vectorize over (should be same length as length(findDone(store@reg)))
ids	ids of job results to be mapped; if missing, map all job results
id	a single job id
part	see <a href="#">batchMapResults</a>
missing.ok	see <a href="#">loadResult</a>
more.args	a list of other arguments to be passed to fun; default is empty list.

### Value

integer vector with job ids. Main purpose is to prepare the registry for submitJobs.

### Note

loadAndFilterResult is not intended to be exported and may be removed in future versions.

### Author(s)

VJ Carey <stvjc@channing.harvard.edu>



**Examples**

```
## Not run:
if (require(geuvStore2)) {
  require(BatchJobs)
  store = makeGeuvStore2()
  fd = tempfile()
  tempreg = makeRegistry("tempSMR", file.dir=fd)
  storeMapResults( store, tempreg, fun=function(job, res, ...) length(res) )
  showStatus(tempreg)
  submitJobs(tempreg, 1:2)
  loadResults(tempreg)
  unlink(fd)
}

## End(Not run)
```

---

storeToFf	<i>extract a vector from store results as ff (out of memory reference); support statistical reductions</i>
-----------	--

---

**Description**

extract a vector from store results as ff (out of memory reference); support statistical reductions

**Usage**

```
storeToFf(store, field, ids = NULL, filter=force, ..., checkField = FALSE,
          ischar=FALSE)
```

**Arguments**

store	instance of <a href="#">ciseStore-class</a>
field	character tag, length one. If name of a numeric field in the result set (typically something like 'chisq' in the GRanges generated by cisAssoc), ff is applied directly. Character variables are converted to factors before ff is applied.
ids	job ids to be used; if NULL, process all jobs
filter	function to be applied when GRanges is loaded from results store, should accept and return a GRanges instance
...	supplied to makeRegistry for a temporary registry: typically will be a vector of package names if additional packages are needed to process results
checkField	if TRUE steps will be taken to verify that the tag to which 'field' evaluates is present in result in the first job
ischar	must be true for character vector to be handled properly as a factor, otherwise NA will be returned

**Details**

uses current BatchJobs configuration to parallelize extraction; reduceResults could be used for a sequential solution

**Value**

a vector as ff reference

**Note**

uses ffbase:::c.ff explicitly to concatenate outputs; there is no guarantee of order among elements

**Examples**

```
if (require(geuvStore2)) {
  require(BatchJobs)
  store = makeGeuvStore2()
  smchisq = storeToFF( store, "chisq", ids=store@validJobs[1:3])
  smchisq
}
```

---

ufeatByTiling

*split featurenames of SummarizedExperiment according to tiling, or to achieve simple balance within seqnames*

---

**Description**

split featurenames of SummarizedExperiment according to tiling, without redundancies

**Usage**

```
ufeatByTiling(se, tiling, maxlen=20)
balancedFeatList(se, maxlen=20)
```

**Arguments**

se	instance of SummarizedExperiment
tiling	GRanges instance corresponding to a genomic tiling
maxlen	numeric ... list elements longer than maxlen are chopped up to have this length, to foster load balancing

**Details**

ufeatByTiling uses findOverlaps, balancedFeatList uses split on seqnames and BBmisc::chunk

**Value**

a list with elements of names(rowRanges(se)) corresponding to the elements of the tiling

**Examples**

```
## Not run:
library(geuvPack)
data(geuFPKM)
library(Homo.sapiens)
au = paste0("chr", 1:22)
tg_500k = tileGenome(seqinfo(TxDb(Homo.sapiens))[au,], tilewidth=500000,
  cut.last.tile.in.chrom=TRUE)
sn = ufeatByTiling(geuFPKM, tg_500k)
summary(sapply(sn,length))
sn2 = balancedFeatList(geuFPKM)
summary(sapply(sn2,length))

## End(Not run)
```

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