

Package ‘gQTLBase’

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Title gQTLBase: infrastructure for eQTL, mQTL and similar studies

Version 1.2.1

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

Description Infrastructure for eQTL, mQTL and similar studies.

Suggests geuvStore, knitr, rmarkdown, BiocStyle, RUnit, GGtools,
Homo.sapiens, IRanges, erma, GenomeInfoDb, gwascats, geuvPack

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

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

NeedsCompilation no

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gQTLBase-package

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

Infrastructure for eQTL, mQTL and similar studies.

Details

The DESCRIPTION file:

```

Package:      gQTLBase
Title:        gQTLBase: infrastructure for eQTL, mQTL and similar studies
Version:      1.2.1
Author:       VJ Carey <stvjc@channing.harvard.edu>
Description:  Infrastructure for eQTL, mQTL and similar studies.
Suggests:    geuvStore, knitr, rmarkdown, BiocStyle, RUnit, GGtools, Homo.sapiens, IRanges, erma, GenomeInfoDb, g
Imports:      GenomicRanges, methods, BatchJobs, BBmisc, S4Vectors, BiocGenerics, foreach, doParallel, bit, ff, rtrack
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

```

Index of help topics:

```

ciseStore-class      Class '"ciseStore"'
describeStore        collect basic descriptive statistics on
                     ciseStore instances
extractByProbes      retrieve eqtlTest results from a ciseStore
                     instance
gQTLBase-package     gQTLBase: infrastructure for eQTL, mQTL and
                     similar studies
mergeCIstates        merge ChromImpute chromatin states, or GWAS hit
                     indicators, to a GRanges
storeApply           apply a function over job results in a
                     ciseStore instance
storeMapResults      use batchMapResults infrastructure to process
                     results in a ciseStore instance
storeToFF            extract a vector from store results as ff (out
                     of memory reference); support statistical
                     reductions
ufeatByTiling        split featurenames of SummarizedExperiment
                     according to tiling, or to achieve simple

```

balance within seqnames

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

Package will complement gQTLStats. geuvStore is a basic illustration relative to GEUVADIS paper. `matprint` is exported from package ff.

Author(s)

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ciseStore-class	Class "ciseStore"
-----------------	-------------------

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(geuvStore)) {
  store = makeGeuvStore()
  store
}
```

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

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 <code>ciseStore-class</code>
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(geuvStore)
mm = makeGeuvStore()
describeStore(mm, ids=partialIds()[1:10], resfilter=function(x) x[x$mindist < 50000])

## End(Not run)
```

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

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 ciseStore-class
probeids, symbols	vector character tokens
gr	instance of GRanges-class
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
...	extra arguments to extractByProbes

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(geuvStore)) {
  store = makeGeuvStore()
  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

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

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 ErmaSet-class . if NULL, supply a GRanges as stateGR, with fields states and statecols
gwcats	an instance of gwaswloc-class , 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 gwcats – 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

```
if (require(gwascat) && 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)
}

```

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=FALSE)
```

Arguments

store	instance of ciseStore-class
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 foreach 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 <code>foreach</code>
verbose	if TRUE will allow progressbars and other messages to display
flatten1	if TRUE will execute <code>unlist(...,recursive=FALSE)</code> on output, so that stratification by host-level chunk is eliminated.

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(geuvStore)) {
  require(BatchJobs)
  store = makeGeuvStore()
  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 ciseStore-class
reg	instance of BatchJobs Registry class
reg2	an empty instance of the Registry class (see makeRegistry)
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 batchMapResults
missing.ok	see loadResult
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)

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Examples

```
## Not run:
if (require(geuvStore)) {
  require(BatchJobs)
  store = makeGeuvStore()
  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 ciseStore-class
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(geuvStore)) {
  require(BatchJobs)
  store = makeGeuvStore()
  smchisq = storeToFF( store, "chisq", ids=store@validJobs[1:3])
  smchisq
}
```

ufeatByTiling	<i>split featurenames of SummarizedExperiment according to tiling, or to achieve simple balance within seqnames</i>
---------------	---

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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