

# Package ‘gwascat’

December 8, 2023

**Title** representing and modeling data in the EMBL-EBI GWAS catalog

**Version** 2.35.0

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**Description** Represent and model data in the EMBL-EBI GWAS catalog.

**Enhances** SNPlocs.Hsapiens.dbSNP144.GRCh37

**Depends** R (>= 3.5.0), methods

**Imports** S4Vectors (>= 0.9.25), IRanges, GenomeInfoDb, GenomicRanges (>= 1.29.6), GenomicFeatures, readr, Biostrings, AnnotationDbi, BiocFileCache, snpStats, VariantAnnotation, AnnotationHub

**Suggests** DO.db, DT, knitr, RBGL, testthat, rmarkdown, dplyr, Gviz, Rsamtools, rtracklayer, graph, ggbio, DelayedArray, TxDb.Hsapiens.UCSC.hg19.knownGene, org.Hs.eg.db, BiocStyle

**VignetteBuilder** knitr

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

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as\_GRanges

*produce a GRanges from gwascat tibble***Description**

produce a GRanges from gwascat tibble

**Usage**

```
as_GRanges(
  x,
  short = TRUE,
  for_short = c("PUBMEDID", "DATE", "DISEASE/TRAIT", "SNPS"),
  genome_tag = "GRCh38"
)
```

**Arguments**

x	a tibble from 'get_cached_gwascat()'
short	logical(1) if TRUE only keep selected columns in mcols
for_short	character() column names to keep in mcols
genome_tag	character(1) defaults to "GRCh38"

---

 bindcadd\_snv

*bind CADD scores of Kircher et al. 2014 to a GRanges instance*


---

**Description**

bind CADD scores of Kircher et al. 2014 to a GRanges instance; by default will use HTTP access at UW

**Usage**

```
bindcadd_snv(
  gr,
  fn = "http://krishna.gs.washington.edu/download/CADD/v1.0/1000G.tsv.gz"
)
```

**Arguments**

gr	query ranges to which CADD scores should be bound
fn	path to Tabix-indexed bgzipped TSV of CADD as distributed at krishna.gs.washington.edu on 1 April 2014

**Details**

joins CADD fields at addresses that match query; the CADD fields for query ranges that are not matched are set to NA

**Value**

GRanges instance with additional fields as obtained in the CADD resource

**Note**

This software developed in part with support from Genentech, Inc.

**Author(s)**

VJ Carey <stvjc@channing.harvard.edu>

**References**

M Kircher, DM Witten, P Jain, BJ O’Roak, GM Cooper, J Shendure, A general framework for estimating the relative pathogenicity of human genetic variants, Nature Genetics Feb 2014, PMID 24487276

**Examples**

```
## Not run:
data(ebicat_2020_04_30)
g2 = as(ebicat_2020_04_30, "GRanges")
# would need to lift over here
bindcadd_snv( g2[which(seqnames(g2)=="chr2")][1:20] )

## End(Not run)
```

---

chklocs	<i>return TRUE if all named SNPs with locations in both the SNPlocs package and the gwascat agree</i>
---------	---

---

**Description**

return TRUE if all named SNPs with locations in both the SNPlocs package and the gwascat agree

**Usage**

```
chklocs(chrtag = "20", gwwl = gwrngs19)
```

**Arguments**

chrtag	character, chromosome identifier
gwwl	instance of {gwaswloc}

---

ebicat_2020_04_30	<i>serialized gwaswloc instance from april 30 2020, sample of 50000 records</i>
-------------------	---

---

**Description**

serialized gwaswloc instance from april 30 2020, sample of 50000 records

**Usage**

ebicat\_2020\_04\_30

**Format**

gwaswloc instance

---

g17SM	<i>SnpMatrix instance from chr17</i>
-------	--------------------------------------

---

**Description**

SnpMatrix instance from chr17

**Usage**

g17SM

**Format**

snpStats SnpMatrix instance

---

getRsids	<i>generic snp name retrieval</i>
----------	-----------------------------------

---

**Description**

generic snp name retrieval

**Usage**

getRsids(x)

**Arguments**

x                      gwaswloc

getRsids,gwaswloc-method  
*specific snp name retrieval*

---

**Description**

specific snp name retrieval

**Usage**

```
## S4 method for signature 'gwaswloc'  
getRsids(x)
```

**Arguments**

x                    gwaswloc

---

getTraits            *generic trait retrieval*

---

**Description**

generic trait retrieval

**Usage**

```
getTraits(x)
```

**Arguments**

x                    gwaswloc

---

```
getTraits,gwaswloc-method
      specific trait retrieval
```

---

**Description**

specific trait retrieval

**Usage**

```
## S4 method for signature 'gwaswloc'
getTraits(x)
```

**Arguments**

x                    gwaswloc

---

```
get_cached_gwascat     use BiocFileCache to retrieve and keep an image of the tsv file distributed by EBI
```

---

**Description**

use BiocFileCache to retrieve and keep an image of the tsv file distributed by EBI

**Usage**

```
get_cached_gwascat(
  url = "http://www.ebi.ac.uk/gwas/api/search/downloads/alternative",
  cache = BiocFileCache::BiocFileCache(),
  refresh = FALSE,
  ...
)
```

**Arguments**

url                    character(1) url to use  
 cache                 BiocFileCache::BiocFileCache instance  
 refresh               logical(1) force download and recaching  
 ...                    passed to bfcadd

**Value**

a tibble as produced by readr::read\_tsv, with attributes extractDate (as recorded in cache as 'access\_time', and problems (a tibble returned by read\_tsv).

**Note**

will If query of cache with 'ebi.ac.uk/gwas' returns 0-row tibble, will populate cache with bfcadd. Uses readr::read\_tsv on cache content to return tibble. The etag field does not seem to be used at EBI, thus user must check for updates.

---

`gg17N`*genotype matrix from chr17 1000 genomes*

---

**Description**

genotype matrix from chr17 1000 genomes

**Usage**`gg17N`**Format**

matrix

**Examples**

```
data(gg17N)
gg17N[1:4, 1:4]
```

---

`gr6.0_hg38`*image of locon6 in GRanges, lifted over to hg38*

---

**Description**

image of locon6 in GRanges, lifted over to hg38

**Usage**`gr6.0_hg38`**Format**

GRanges instance



---

gw6.rs_17	<i>character vector of rs numbers for SNP on chr17</i>
-----------	--

---

**Description**

character vector of rs numbers for SNP on chr17

**Usage**

```
gw6.rs_17
```

**Format**

character vector

---

gwascat_from_AHub	<i>grab an image of EBI GWAS catalog from AnnotationHub</i>
-------------------	---

---

**Description**

grab an image of EBI GWAS catalog from AnnotationHub

**Usage**

```
gwascat_from_AHub(tag = "AH91571", simple = FALSE, fixNonASCII = TRUE)
```

**Arguments**

tag	character(1) defaults to "AH91571" which is the 3.30.2021 image
simple	logical(1) if TRUE, just returns data.frame as retrieved from EBI; defaults to FALSE
fixNonASCII	logical(1) if TRUE, use iconv to identify and eliminate non-ASCII content

**Value**

If 'simple', a data.frame is returned based on TSV data produced by EBI. Otherwise, non-ASCII content is processed according to the value of 'fixNonASCII' and a 'gwaswloc' instance is returned, which has a concise show method. This can be coerced to a simple GRanges instance with as(..., "GRanges"). The reference build is GRCh38.

**Examples**

```
gwcats = gwascat_from_AHub()
gwcats
```

---

gwastagger	<i>GRanges with LD information on 9998 SNP</i>
------------	--

---

**Description**

GRanges with LD information on 9998 SNP

**Usage**

gwastagger

**Format**

GRanges

---

gwaswloc-class	<i>container for gwas hit data and GRanges for addresses</i>
----------------	--

---

**Description**

container for gwas hit data and GRanges for addresses

---

gwcatsnapshot	<i>use AnnotationHub snapshot as basis for gwaswloc structure creation</i>
---------------	--

---

**Description**

use AnnotationHub snapshot as basis for gwaswloc structure creation

**Usage**

gwcatsnapshot(x, fixNonASCII = TRUE)

**Arguments**

x	inherits from data.frame, with columns consistent with EBI table
fixNonASCII	logical(1) if TRUE, use iconv to replace non-ASCII character, important for CMD check but perhaps not important for applied use

**Examples**

```

ah = AnnotationHub::AnnotationHub()
entitytab = AnnotationHub::query(ah, "gwascatData")
cand = names(entitytab)[1]
stopifnot(nchar(cand)>0)
tab = ah[[cand]]
gww = gwascat_snapshot(tab)
gww
length(gww)

```

---

gwcecx2gviz

*Prepare salient components of GWAS catalog for rendering with Gviz*


---

**Description**

Prepare salient components of GWAS catalog for rendering with Gviz

**Usage**

```

gwcecx2gviz(
  basegr,
  contextGR = GRanges(seqnames = "chr17", IRanges::IRanges(start = 37500000, width =
    1e+06)),
  txrefobj = TxDb.Hsapiens.UCSC.hg19.knownGene::TxDb.Hsapiens.UCSC.hg19.knownGene,
  genome = "hg19",
  genesymobj = org.Hs.eg.db::org.Hs.eg.db,
  plot.it = TRUE,
  maxmlp = 25
)

```

**Arguments**

basegr	gwaswloc instance containing information about GWAS in catalog
contextGR	A GRanges instance delimiting the visualization in genomic coordinates
txrefobj	a TxDb instance
genome	character tag like 'hg19'
genesymobj	an OrgDb instance
plot.it	logical, if FALSE, just return list
maxmlp	maximum value of $-10 \log p$ – winsorization of all larger values is performed, modifying the contents of Pvalue\_mlogp in the elementMetadata for the call

**Examples**

```

data(ebicat_2020_04_30)
# GenomeInfoDb::seqlevelsStyle(ebicat_2020_04_30) = "UCSC" # no more
GenomeInfoDb::seqlevels(ebicat_2020_04_30) = paste0("chr", GenomeInfoDb::seqlevels(ebicat_2020_04_30))
gwcecx2gviz(ebicat_2020_04_30)

```

---

ldtagr                      *expand a list of variants by including those in a VCF with LD exceeding some threshold; uses snpStats ld()*

---

### Description

expand a list of variants by including those in a VCF with LD exceeding some threshold; uses snpStats ld()

### Usage

```
ldtagr(
  snprng,
  tf,
  samples,
  genome = "hg19",
  lbmaf = 0.05,
  lbR2 = 0.8,
  radius = 1e+05
)
```

### Arguments

snprng	a named GRanges for a single SNP. The name must correspond to the name that will be assigned by genotypeToSnpMatrix (from VariantTools) to the corresponding column of a SnpMatrix.
tf	TabixFile instance pointing to a bgzipped tabix-indexed VCF file
samples	a vector of sample identifiers, if excluded, all samples used
genome	tag like 'hg19'
lbmaf	lower bound on variant MAF to allow consideration
lbR2	lower bound on R squared for regarding SNP to be incorporated
radius	radius of search in bp around the input range

### Value

a GRanges with names corresponding to 'new' variants and mcols fields 'paramRangeID' (base variant input) and 'R2'

### Note

slow but safe approach. probably a matrix method could be substituted using the nice sparse approach already in snpStats

### Author(s)

VJ Carey

**Examples**

```

cand = GenomicRanges::GRanges("1", IRanges::IRanges(113038694, width=1))
names(cand) = "rs883593"
requireNamespace("VariantAnnotation")
expath = dir(system.file("vcf", package="gwascat"), patt="*exon.*gz$", full=TRUE)
tf = Rsamtools::TabixFile(expath)
ldtagr( cand, tf, lbR2 = .8)

```

---

locon6	<i>location data for 10000 SNP</i>
--------	------------------------------------

---

**Description**

location data for 10000 SNP

**Usage**

locon6

**Format**

data.frame, coordinates are hg19

---

locs4trait	<i>get locations for SNP affecting a selected trait</i>
------------	---

---

**Description**

get locations for SNP affecting a selected trait

**Usage**

```
locs4trait(gwvl, trait, tag = "DISEASE/TRAIT")
```

**Arguments**

gwvl	instance of {gwaswloc}
trait	character, name of trait
tag	character, name of field to be used for trait enumeration

---

low17	<i>SnpMatrix instance from chr17</i>
-------	--------------------------------------

---

**Description**

SnpMatrix instance from chr17

**Usage**

low17

**Format**

snpStats SnpMatrix instance

---

makeCurrentGwascat	<i>read NHGRI GWAS catalog table and construct associated GRanges instance records for which clear genomic position cannot be determined are dropped from the ranges instance an effort is made to use reasonable data types for GRanges metadata, so some qualifying characters such as (EA) in Risk allele frequency field will simply be omitted during coercion of contents of that field to numeric.</i>
--------------------	---

---

**Description**

read NHGRI GWAS catalog table and construct associated GRanges instance records for which clear genomic position cannot be determined are dropped from the ranges instance an effort is made to use reasonable data types for GRanges metadata, so some qualifying characters such as (EA) in Risk allele frequency field will simply be omitted during coercion of contents of that field to numeric.

**Usage**

```
makeCurrentGwascat(
  table.url = "http://www.ebi.ac.uk/gwas/api/search/downloads/alternative",
  fixNonASCII = TRUE,
  genome = "GRCh38",
  withOnt = TRUE
)
```

**Arguments**

<code>table.url</code>	string identifying the .txt file curated at EBI/EMBL
<code>fixNonASCII</code>	logical, if TRUE, non-ASCII characters as identified by <code>iconv</code> will be replaced by asterisk
<code>genome</code>	character string: 'GRCh38' is default and yields current image as provided by EMBL/EBI; 'GRCh37' yields a realtime liftOver to hg19 coordinates, via AnnotationHub storage of the chain files. Any other value yields an error.
<code>withOnt</code>	logical indicating whether 'alternative' (ontology-present, includes repetition of loci with one:many ontological mapping) or 'full' (ontology-absent, one record per locus report) version of distributed table

**Value**

a slightly extended `GRanges` instance, with class name 'gwaswloc'; the purpose of the introduction of this class is to support a concise `show` method that does not produce very long lines owing to large numbers of fields in the `mcols` component.

**Note**

'`readr::read_tsv`' records problems when some records have field contents that are inconsistent with the column specification. This information can be retrieved from the metadata slot of the returned object, as noted in a message produced when this function is run.

**Author(s)**

VJ Carey

**Examples**

```
# if you have good internet access
if (interactive()) {
  newcatr = makeCurrentGwascat()
  newcatr
}
```

---

obo2graphNEL

*convert a typical OBO text file to a graphNEL instance (using Term elements)*

---

**Description**

convert a typical OBO text file to a graphNEL instance (using Term elements)

## Usage

```
obo2graphNEL(  
  obo = "human-phenotype-ontology.obo",  
  kill = "\\[Typedef\\]",  
  killTrailSp = TRUE  
)
```

## Arguments

obo	string naming a file in OBO format
kill	entity types to be excluded from processing – probably this should be in a 'keep' form, but for now this works.
killTrailSp	In the textual version of EFO ca. Aug 2015, there is a trailing blank in the tag field defining EFO:0000001, which is not present in references to this term. Set this to TRUE to eliminate this, or graphNEL construction will fail to validate.

## Details

Very rudimentary list and grep operations are used to retain sufficient information to map the DAG to a graphNEL, using formal term identifiers as node names and 'is-a' relationships as edges, and term names and other metadata are assigned to nodeData components.

## Value

a graphNEL instance

## Note

The OBO for Human Disease ontology is serialized as text with this package.

## Author(s)

VJ Carey <stvjc@channing.harvard.edu>

## References

For use with human disease ontology, [http://www.obofoundry.org/cgi-bin/detail.cgi?id=disease\\_ontology](http://www.obofoundry.org/cgi-bin/detail.cgi?id=disease_ontology)

## Examples

```
data(efo.obo.g)  
requireNamespace("graph")  
hn = graph::nodes(efo.obo.g)[1:5]  
hn  
graph::nodeData(efo.obo.g, hn[5])
```



---

process\_gwas\_dataframe

*convert GWAS catalog data.frame to gwaswloc, a GRanges extension with simple show method*

---

### Description

convert GWAS catalog data.frame to gwaswloc, a GRanges extension with simple show method

### Usage

```
process_gwas_dataframe(df)
```

### Arguments

df                    data.frame

---

riskyAlleleCount      *given a matrix of subjects x SNP calls, count number of risky alleles*

---

### Description

given a matrix of subjects x SNP calls, count number of risky alleles for various conditions, relative to NHGRI GWAS catalog

### Usage

```
riskyAlleleCount(
  callmat,
  matIsAB = TRUE,
  chr,
  gwwl,
  snpap = "SNPlocs.Hsapiens.dbSNP144.GRCh37",
  gencode = c("A/A", "A/B", "B/B")
)
```

### Arguments

callmat	matrix with subjects as rows, SNPs as columns; entries can be generic A/A, A/B, B/B, or specific nucleotide calls
matIsAB	logical, FALSE if nucleotide codes are present, TRUE if generic call codes are present; in the latter case, gwascat::ABmat2nuc will be run
chr	code for chromosome, should work with the SNP annotation getSNPlocs function, so likely "ch[nn]"
gwwl	an instance of {gwaswloc}
snpap	name of a Bioconductor SNPlocs.Hsapiens.dbSNP.* package
gencode	codes used for generic SNP call

**Value**

matrix with rows corresponding to subjects , columns corresponding to SNP

**Examples**

```
## Not run:
data(gg17N) # translated from GGdata chr 17 calls using ABmat2nuc
data(ebicat37)
library(GenomeInfoDb)
seqlevelsStyle(ebicat37) = "UCSC"
h17 = riskyAlleleCount(gg17N, matIsAB=FALSE, chr="ch17", gwwl=ebicat37)
h17[1:5,1:5]
table(as.numeric(h17))

## End(Not run)
```

---

 si.hs.37

*Seqinfo for GRCh37*


---

**Description**

Seqinfo for GRCh37

**Usage**

si.hs.37

**Format**

GenomeInfoDb Seqinfo instance

---

 si.hs.38

*Seqinfo for GRCh38*


---

**Description**

Seqinfo for GRCh38

**Usage**

si.hs.38

**Format**

GenomeInfoDb Seqinfo instance

---

subsetByChromosome      *generic trait subsetting*

---

**Description**

generic trait subsetting

**Usage**

```
subsetByChromosome(x, ch)
```

**Arguments**

x	gwaswloc
ch	character vector of chromosomes

---

subsetByChromosome, gwaswloc-method  
*specific trait subsetting*

---

**Description**

specific trait subsetting

**Usage**

```
## S4 method for signature 'gwaswloc'  
subsetByChromosome(x, ch)
```

**Arguments**

x	gwaswloc
ch	character vector of chromosomes

---

subsetByTraits	<i>generic trait subsetting</i>
----------------	---------------------------------

---

**Description**

generic trait subsetting

**Usage**

```
subsetByTraits(x, tr)
```

**Arguments**

x	gwaswloc
tr	character vector of traits

---

subsetByTraits,gwaswloc-method	<i>specific trait subsetting</i>
--------------------------------	----------------------------------

---

**Description**

specific trait subsetting

**Usage**

```
## S4 method for signature 'gwaswloc'  
subsetByTraits(x, tr)
```

**Arguments**

x	gwaswloc
tr	character vector of traits

---

topTraits	<i>operations on GWAS catalog</i>
-----------	-----------------------------------

---

**Description**

operations on GWAS catalog

**Usage**

```
topTraits(gwwl, n = 10, tag = "DISEASE/TRAIT")
```

**Arguments**

gwwl	instance of {gwaswloc}
n	numeric, number of traits to report
tag	character, name of field to be used for trait enumeration

**Value**

topTraits returns a character vector of most frequently occurring traits in the database

locs4trait returns a {gwaswloc} object with records defining associations to the specified trait

chklocs returns a logical that is TRUE when the asserted locations of SNP in the GWAS catalog agree with the locations given in the dbSNP package SNPlocs.Hsapiens.dbSNP144.GRCh37

**Author(s)**

VJ Carey <stvjc@channing.harvard.edu>

**Examples**

```
data(ebicat_2020_04_30)
topTraits(ebicat_2020_04_30)
```

---

traitsManh	<i>use ggbio facilities to display GWAS results for selected traits in genomic coordinates</i>
------------	--

---

**Description**

use ggbio facilities to display GWAS results for selected traits in genomic coordinates

**Usage**

```
traitsManh(
  gwr,
  selr = GRanges(seqnames = "chr17", IRanges(3e+07, 5e+07)),
  traits = c("Asthma", "Parkinson's disease", "Height", "Crohn's disease"),
  truncmlp = 25,
  ...
)
```

**Arguments**

<code>gwr</code>	GRanges instance as managed by the gwaswloc container design, with Disease.Trait and Pvalue_mlog among elementMetadata columns
<code>selr</code>	A GRanges instance to restrict the gwr for visualization. Not tested for noncontiguous regions.
<code>traits</code>	Character vector of traits to be exhibited; GWAS results with traits not among these will be labeled "other".
<code>truncmlp</code>	Maximum value of $-\log_{10} p$ to be displayed; in the raw data this ranges to the hundreds and can cause bad compression.
<code>...</code>	not currently used

**Details**

uses a ggbio autoplot

**Value**

autoplot value

**Note**

An xlab is added, concatenating genome tag with seqnames tag.

**Author(s)**

VJ Carey <stvjc@channing.harvard.edu>

**Examples**

```
# do a p-value truncation if you want to reduce compression
## Not run: # ggbio July 2018
data(ebicat_2020_04_30)
library(GenomeInfoDb)
seqlevelsStyle(ebicat_2020_04_30) = "UCSC"
traitsManh(ebicat_2020_04_30)

## End(Not run)
```

---

[,gwaswloc,ANY,ANY,ANY-method  
*extractor for gwaswloc*

---

### **Description**

extractor for gwaswloc

### **Usage**

```
## S4 method for signature 'gwaswloc,ANY,ANY,ANY'  
x[i, j, ..., drop = FALSE]
```

### **Arguments**

x	gwaswloc
i	index
j	index
...	addtl indices
drop	logical(1)

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