Package ‘gwasrapidd’

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Type Package

Title 'REST' 'API' Client for the 'NHGRI'-'EBI' 'GWAS' Catalog

Version 0.99.12

Description 'GWAS' R 'API' Data Download.

This package provides easy access to the 'NHGRI'-'EBI' 'GWAS' Catalog data by accessing the 'REST' 'API' <https://www.ebi.ac.uk/gwas/rest/docs/api/>.

Depends R (>= 3.2.3)

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URL https://github.com/ramiromagno/gwasrapidd

BugReports https://github.com/ramiromagno/gwasrapidd/issues

Encoding UTF-8

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Imports magrittr, httr, urltools, pingr, stringr, concatenate, dplyr,
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Suggests httest, spelling, covr, knitr, rmarkdown

Collate 'browser.R' 'class-associations.R' 'class-studies.R'
   'class-traits.R' 'class-variants.R' 'data.R' 'ebi_server.R'
   'generics.R' 'get_associations.R' 'get_metadata.R'
   'get_studies.R' 'get_traits.R' 'get_variants.R'
   'gwasrapidd-package.R' 'id_mapping.R' 'list_joins.R'
   'missing.R' 'parse-associations.R' 'parse_studies.R'
   'parse-traits.R' 'parse-utils.R' 'parse-variants.R'
   'post-studies.R' 'post-traits.R' 'post-variants.R'
   'recursive_apply.R' 'request.R' 's4-utils.R' 'sure.R' 'tests.R'
   'utils-pipe.R' 'utils.R'

VignetteBuilder knitr

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associations-class

An S4 class to represent a set of GWAS Catalog associations

Description

The association object consists of six slots, each a table (tibble), that combined form a relational database of a subset of GWAS Catalog associations. Each association is an observation (row) in the associations table — main table. All tables have the column association_id as primary key.

Slots

associations A tibble listing associations. Columns:

- **association_id** GWAS Catalog association accession identifier, e.g., "20250".
- **pvalue** Reported p-value for strongest variant risk or effect allele.
- **pvalue_description** Information describing context of p-value.
- **pvalue_mantissa** Mantissa of p-value.
- **pvalue_exponent** Exponent of p-value.
- **multiple_snp_haplotype** Whether the association is for a multi-SNP haplotype.
- **snp_interaction** Whether the association is for a SNP-SNP interaction.
- **snp_type** Whether the SNP has previously been reported. Either 'known' or 'novel'.
- **standard_error** Standard error of the effect size.
- **range** Reported 95% confidence interval associated with strongest SNP risk allele, along with unit in the case of beta coefficients. If 95% CIs have not been not reported, these are estimated using the standard error, when available.
- **or_per_copy_number** Reported odds ratio (OR) associated with strongest SNP risk allele.
  Note that all ORs included in the Catalog are >1.
- **beta_number** Beta coefficient associated with strongest SNP risk allele.
- **beta_unit** Beta coefficient unit.
- **beta_direction** Beta coefficient direction, either 'decrease' or 'increase'.
- **beta_description** Additional beta coefficient comment.
- **last_mapping_date** Last time this association was mapped to Ensembl.
- **last_update_date** Last time this association was updated.

loci A tibble listing loci. Columns:

- **association_id** GWAS Catalog association accession identifier, e.g., "20250".
- **locus_id** A locus identifier referring to a single variant locus or to a multi-loci entity such as a multi-SNP haplotype.
- **haplotype_snp_count** Number of variants per locus. Most loci are single-SNP loci, i.e., there is a one to one relationship between a variant and a locus_id (haplotype_snp_count == NA). There are however cases of associations involving multiple loci at once, such as SNP-SNP interactions and multi-SNP haplotypes. This is signalled in the columns: multiple_snp_haplotype and snp_interaction with value TRUE.
**description**  Description of the locus identifier, e.g., 'Single variant', SNP x SNP interaction, or 3-SNP Haplotype.

**risk alleles**  A tibble listing risk alleles. Columns:
- **association_id**  GWAS Catalog association accession identifier, e.g., "20250".
- **locus_id**  A locus identifier referring to a single variant locus or to a multi-loci entity such as a multi-SNP haplotype.
- **variant_id**  Variant identifier, e.g., 'rs1333048'.
- **risk_allele**  Risk allele or effect allele.
- **risk_frequency**  Reported risk/effect allele frequency associated with strongest SNP in controls (if not available among all controls, among the control group with the largest sample size). If the associated locus is a haplotype the haplotype frequency will be extracted.
- **genome_wide**  Whether this variant allele has been part of a genome-wide study or not.
- **limited_list**  Undocumented.

**genes**  A tibble listing author reported genes. Columns:
- **association_id**  GWAS Catalog association accession identifier, e.g., "20250".
- **locus_id**  A locus identifier referring to a single variant locus or to a multi-loci entity such as a multi-SNP haplotype.
- **gene_name**  Gene symbol according to HUGO Gene Nomenclature (HGNC).

**ensembl_ids**  A tibble listing Ensembl gene identifiers. Columns:
- **association_id**  GWAS Catalog association accession identifier, e.g., "20250".
- **locus_id**  A locus identifier referring to a single variant locus or to a multi-loci entity such as a multi-SNP haplotype.
- **gene_name**  Gene symbol according to HUGO Gene Nomenclature (HGNC).
- **ensembl_id**  The Ensembl identifier of an Ensembl gene, see Section Gene annotation in Ensembl for more information.

**entrez_ids**  A tibble listing Entrez gene identifiers. Columns:
- **association_id**  GWAS Catalog association accession identifier, e.g., "20250".
- **locus_id**  A locus identifier referring to a single variant locus or to a multi-loci entity such as a multi-SNP haplotype.
- **gene_name**  Gene symbol according to HUGO Gene Nomenclature (HGNC).
- **entrez_id**  The Entrez identifier of a gene, see ref. doi: 10.1093/nar/gkq1237 for more information.

---

**association_to_study**  Map an association id to a study id

**Description**

Map an association accession identifier to a study accession identifier.

**Usage**

`association_to_study(association_id, verbose = FALSE, warnings = TRUE)`
**association_to_trait**

**Arguments**

- **association_id** A character vector of association accession identifiers.
- **verbose** Whether the function should be verbose about the different queries or not.
- **warnings** Whether to print warnings.

**Value**

A dataframe of two identifiers. First column is the association identifier and the second column is the study identifier.

**Examples**

```r
# Map GWAS association identifiers to study identifiers
association_to_study(c('24300097', '24299759'))
```

---

**association_to_trait**  
*Map an association id to an EFO trait id*

**Description**

Map an association accession identifier to an EFO trait id.

**Usage**

```r
association_to_trait(association_id, verbose = FALSE, warnings = TRUE)
```

**Arguments**

- **association_id** A character vector of association accession identifiers.
- **verbose** Whether the function should be verbose about the different queries or not.
- **warnings** Whether to print warnings.

**Value**

A dataframe of two identifiers. First column is the association identifier and the second column is the EFO trait identifier.

**Examples**

```r
# Map GWAS association identifiers to EFO trait identifiers
association_to_trait(c('24300097', '24299759'))
```
association_to_variant

Map an association id to a variant id

Description

Map an association accession identifier to a variant identifier.

Usage

association_to_variant(association_id, verbose = FALSE, warnings = TRUE)

Arguments

association_id  A character vector of association accession identifiers.
verbose        Whether the function should be verbose about the different queries or not.
warnings       Whether to print warnings.

Value

A dataframe of two identifiers. First column is the association identifier and the second column is the variant identifier.

Examples

# Map GWAS association identifiers to variant identifiers
association_to_variant(c("24300097", "24299759"))

bind

Bind GWAS Catalog objects

Description

Binds together GWAS Catalog objects of the same class. Note that bind() preserves duplicates whereas union does not.

Usage

bind(x, ...)

Arguments

x        An object of class: studies, associations, variants, or traits.
...      Objects of the same class as x.
Value
An object of the same class as x.

Examples

# Join two studies objects.
bind(studies_ex01, studies_ex02)

# Join two associations objects.
bind(associations_ex01, associations_ex02)

# Join two variants objects.
bind(variants_ex01, variants_ex02)

# Join two traits objects.
bind(traits_ex01, traits_ex02)

cytoegenetic_bands

A dataset containing the GRCh38 human cytogenetic bands and their genomic coordinates.

Usage
cytogenetic_bands

Format
A data frame with 862 rows and 8 variables:

cytoegenetic_band  Cytogenetic band name. See Cytogenetic Nomenclature below.
chromosome  Chromosome name: 1 through 22 (the autosomes), X or Y.
start  Genomic start position of the cytogenetic band. Starts at 1.
end  Genomic end position of the cytogenetic band. End position is included in the band interval.
length  Length of the genomic interval of cytogenetic band.
assembly  Assembly version, should be 'GRCh38'.
stain  Giemsa stain results: Giemsa negative, 'gneg'; Giemsa positive, of increasing intensities, 'gpos25', 'gpos50', 'gpos75', and 'gpos100'; centromeric region, 'acen'; heterochromatin, either pericentric or telomeric, 'gvar'; and short arm of acrocentric chromosomes 13, 14, 15, 21, and 22 are coded as 'stalk'.
last_download_date  Time stamp of last time this dataset was downloaded from Ensembl.
Details

Genomic coordinates are for fully closed intervals.

Cytogenetic Nomenclature

Cytogenetic bands are numbered from the centromere outwards in both directions towards the
telomeres on the shorter p arm and the longer q arm.
The first number or letter represents the chromosome. Chromosomes 1 through 22 (the autosomes)
are designated by their chromosome number. The sex chromosomes are designated by X or Y. The
next letter represents the arm of the chromosome: p or q.
The numbers cannot be read in the normal decimal numeric system e.g. 36, but rather 3-6 (region
3 band 6). Counting starts at the centromere as region 1 (or 1-0), to 11 (1-1) to 21 (2-1) to 22 (2-2)
etc. Subbands are added in a similar way, e.g. 21.1 to 21.2, if the bands are small or only appear at
a higher resolution.

Source

https://rest.ensembl.org/info/assembly/homo_sapiens?content-type=application/json&

exists_variant

Check if a variant exists in the Catalog.

Description

This function attempts to get a variant by its variant identifier and checks the response code. If the
response code is 200 then the response has been successful, meaning that the variant does exist in
the GWAS Catalog. If the response is 404 then the variant is not found in the Catalog database.
Other errors are mapped to NA.

Usage

exists_variant(variant_id = NULL, verbose = FALSE, page_size = 20L)

Arguments

variant_id A character vector of GWAS Catalog variant identifiers.
verbose Whether the function should be verbose about the different queries or not.
page_size An integer scalar indicating the page value to be used in the JSON requests, can
be between 1 and 1000.

Value

A named logical vector, TRUE indicates that the variant does exist in the Catalog, FALSE other-
wise. NA codes other types of errors. The names of the vector are the variant identifiers passed as
variant_id.
Examples

exists_variant('rs12345')

exists_variant('rs11235813')

Description

These are examples of GWAS Catalog entities shipped with gwasrapidd:

Usage

studies_ex01
studies_ex02
associations_ex01
associations_ex02
variants_ex01
variants_ex02
traits_ex01
traits_ex02

Format

**studies_ex01** An S4 studies object of 2 studies: 'GCST001585' and 'GCST003985'.

**studies_ex02** An S4 studies object of 2 studies: 'GCST001585' and 'GCST006655'.

**associations_ex01** An S4 associations object of 4 associations: '22509', '22505', '19537565' and '19537593'.

**associations_ex02** An S4 associations object of 3 associations: '19537593', '31665940' and '34944736'.

**variants_ex01** An S4 variants object of 3 variants: 'rs146992477', 'rs56261590' and 'rs4725504'.

**variants_ex02** An S4 variants object of 4 variants: 'rs56261590', 'rs4725504', 'rs11099757' and 'rs16871509'.

**traits_ex01** An S4 traits object of 3 traits: 'EFO_0004884', 'EFO_0004343' and 'EFO_0005299'.

**traits_ex02** An S4 traits object of 3 traits: 'EFO_0004884', 'EFO_0004343' and 'EFO_0005299'.

**traits_ex03** An S4 traits object of 3 traits: 'EFO_0004884', 'EFO_0004343' and 'EFO_0005299'.
traits_ex02 An S4 traits object of 4 traits: 'EFO_0007845', 'EFO_0004699', 'EFO_0004884' and 'EFO_0004875'.

An object of class studies of length 1.
An object of class associations of length 1.
An object of class associations of length 1.
An object of class variants of length 1.
An object of class variants of length 1.
An object of class traits of length 1.
An object of class traits of length 1.

getAssociations An S4 traits object of 4 traits: 'EFO_0007845', 'EFO_0004699', 'EFO_0004884' and 'EFO_0004875'.

An object of class studies of length 1.
An object of class associations of length 1.
An object of class associations of length 1.
An object of class variants of length 1.
An object of class variants of length 1.
An object of class traits of length 1.
An object of class traits of length 1.

get_associations Get GWAS Catalog associations

Description

Retrieves associations via the NHGRI-EBI GWAS Catalog REST API. The REST API is queried multiple times with the criteria passed as arguments (see below). By default all associations that match the criteria supplied in the arguments are retrieved: this corresponds to the default option set_operation set to 'union'. If you rather have only the associations that match simultaneously all criteria provided, then set set_operation to 'intersection'.

Usage

get_associations(
  study_id = NULL,
  association_id = NULL,
  variant_id = NULL,
  efo_id = NULL,
  pubmed_id = NULL,
  efo_trait = NULL,
  set_operation = "union",
  interactive = TRUE,
  verbose = FALSE,
  warnings = TRUE
)

Arguments

study_id A character vector of GWAS Catalog study accession identifiers.
association_id A character vector of GWAS Catalog association identifiers.
variant_id A character vector of GWAS Catalog variant identifiers.
efo_id A character vector of EFO identifiers.
pubmed_id An integer vector of PubMed identifiers.
get_child_efo

- **efo_trait**: A character vector of EFO trait descriptions, e.g., 'uric acid measurement'.
- **set_operation**: Either 'union' or 'intersection'. This tells how associations retrieved by different criteria should be combined: 'union' binds together all results removing duplicates and 'intersection' only keeps same associations found with different criteria.
- **interactive**: A logical. If all associations are requested, whether to ask interactively if we really want to proceed.
- **verbose**: A logical indicating whether the function should be verbose about the different queries or not.
- **warnings**: A logical indicating whether to print warnings, if any.

**Details**

Please note that all search criteria are vectorised, thus allowing for batch mode search, e.g., one can search by multiple variant identifiers at once by passing a vector of identifiers to `variant_id`.

**Value**

An `associations` object.

**Examples**

```r
# Get an association by study identifier
get_associations(study_id = 'GCST001085', warnings = FALSE)

# Get an association by association identifier
get_associations(association_id = '25389945', warnings = FALSE)

# Get associations by variant identifier
get_associations(variant_id = 'rs3798440', warnings = FALSE)

# Get associations by EFO trait identifier
get_associations(efo_id = 'EFO_0005537', warnings = FALSE)
```

**get_child_efo**

*Get all child terms of this trait in the EFO hierarchy*

**Description**

Get all child terms of this trait in the EFO hierarchy
Usage

get_child_efo(
  efo_id,
  verbose = FALSE,
  warnings = TRUE,
  page_size = 20L,
  progress_bar = TRUE
)

Arguments

  efo_id          A character vector of EFO identifiers.
  verbose         A logical indicating whether the function should be verbose about the different
                   queries or not.
  warnings        A logical indicating whether to print warnings, if any.
  page_size       An integer scalar indicating the page value to be used in the JSON requests,
                   can be between 1 and 1000.
  progress_bar    Whether to show a progress bar as the paginated resources are retrieved.

Value

A named list whose values are character vectors of EFO identifiers.

Examples

get_child_efo(c(‘EFO_0004884’, ‘EFO_0004343’, ‘EFO_0005299’))

get_metadata

Get GWAS Catalog metadata

Description

Provides a list of the resources the GWAS Catalog data is currently mapped against: Ensembl release number, Genome build version and dbSNP version. In addition, the date since this combination of resource versions has been in use is also returned.

Usage

get_metadata(VERBOSE = FALSE, warnings = TRUE)

Arguments

  verbose         Whether to be chatty.
  warnings        Whether to trigger a warning if the request is not successful.
Value

A named list whose names are:

- `ensembl_release_number`: Ensembl release number;
- `genome_build_version`: Genome build version;
- `dbsnp_version`: dbsNP version.
- `usage_start_date`: Date since this combination of resource versions has been in use.

Examples

```r
get_metadata(warnings = FALSE)
```

---

**get_studies**

*Get GWAS Catalog studies*

Description

Retrieves studies via the NHGRI-EBI GWAS Catalog REST API. The REST API is queried multiple times with the criteria passed as arguments (see below). By default all studies that match the criteria supplied in the arguments are retrieved: this corresponds to the default option `set_operation` set to 'union'. If you rather have only the studies that match simultaneously all criteria provided, then set `set_operation` to 'intersection'.

Usage

```r
get_studies(
  study_id = NULL,
  association_id = NULL,
  variant_id = NULL,
  efo_id = NULL,
  pubmed_id = NULL,
  user_requested = NULL,
  full_pvalue_set = NULL,
  efo_uri = NULL,
  efo_trait = NULL,
  reported_trait = NULL,
  set_operation = "union",
  interactive = TRUE,
  verbose = FALSE,
  warnings = TRUE
)
```
get_studies

Arguments

study_id A character vector of GWAS Catalog study accession identifiers.
association_id A character vector of GWAS Catalog association identifiers.
variant_id A character vector of GWAS Catalog variant identifiers.
efo_id A character vector of EFO identifiers.
pubmed_id An integer vector of PubMed identifiers.
user_requested A logical (scalar!) indicating to retrieve either studies requested by users of the Catalog (TRUE) or otherwise (FALSE).
full_pvalue_set A logical (scalar!) indicating to retrieve studies with full summary statistics (TRUE) or studies without it (FALSE).
efo_uri A character vector of EFO URIs.
efo_trait A character vector of EFO trait descriptions, e.g., 'uric acid measurement'.
reported_trait A character vector of phenotypic traits as reported by the original authors of the study.
set_operation Either 'union' or 'intersection'. This tells how studies retrieved by different criteria should be combined: 'union' binds together all results removing duplicates and 'intersection' only keeps same studies found with different criteria.
interactive A logical. If all studies are requested, whether to ask interactively if we really want to proceed.
verbose Whether the function should be verbose about the different queries or not.
warnings Whether to print warnings.

Details

Please note that all search criteria are vectorised, thus allowing for batch mode search, e.g., one can search by multiple variant identifiers at once by passing a vector of identifiers to variant_id.

Value

A studies object.

Examples

# Get a study by its accession identifier
get_studies(study_id = 'GCST001085', warnings = FALSE)

# Get a study by association identifier
get_studies(association_id = '25389945', warnings = FALSE)

# Get studies by variant identifier
get_studies(variant_id = 'rs3798440', warnings = FALSE)

# Get studies by EFO trait identifier
get_studies(efo_id = 'EFO_0005537', warnings = FALSE)
get_traits

Get GWAS Catalog EFO traits

Description

Retrieves traits via the NHGRI-EBI GWAS Catalog REST API. The REST API is queried multiple times with the criteria passed as arguments (see below). By default all traits that match the criteria supplied in the arguments are retrieved: this corresponds to the default option set_operation set to 'union'. If you rather have only the traits that match simultaneously all criteria provided, then set set_operation to 'intersection'.

Usage

get_traits(
  study_id = NULL,
  association_id = NULL,
  efo_id = NULL,
  pubmed_id = NULL,
  efo_uri = NULL,
  efo_trait = NULL,
  set_operation = "union",
  verbose = FALSE,
  warnings = TRUE
)

Arguments

study_id A character vector of GWAS Catalog study accession identifiers.
association_id A character vector of GWAS Catalog association identifiers.
efo_id A character vector of EFO identifiers.
pubmed_id An integer vector of PubMed identifiers.
efo_uri A character vector of EFO URIs.
efo_trait A character vector of EFO trait descriptions, e.g., 'uric acid measurement'.
set_operation Either 'union' or 'intersection'. This tells how traits retrieved by different criteria should be combined: 'union' binds together all results removing duplicates and 'intersection' only keeps same traits found with different criteria.
verbose A logical indicating whether the function should be verbose about the different queries or not.
warnings A logical indicating whether to print warnings, if any.

Details

Please note that all search criteria are vectorised, thus allowing for batch mode search, e.g., one can search by multiple trait identifiers at once by passing a vector of identifiers to efo_id.
get_variants

Value
A traits object.

Examples

# Get traits by study identifier
get_traits(study_id = 'GCST001085', warnings = FALSE)

# Get traits by association identifier
get_traits(association_id = '25389945', warnings = FALSE)

# Get a trait by its EFO identifier
get_traits(efo_id = 'EFO_0005537', warnings = FALSE)

get_variants

Get GWAS Catalog variants

Description
Retrieves variants via the NHGRI-EBI GWAS Catalog REST API. The REST API is queried multiple times with the criteria passed as arguments (see below). By default all variants that match the criteria supplied in the arguments are retrieved: this corresponds to the default option set_operation set to 'union'. If you rather have only the variants that match simultaneously all criteria provided, then set set_operation to 'intersection'.

Usage

get_variants(
  study_id = NULL,
  association_id = NULL,
  variant_id = NULL,
  efo_id = NULL,
  pubmed_id = NULL,
  genomic_range = NULL,
  cytogenetic_band = NULL,
  gene_name = NULL,
  efo_trait = NULL,
  reported_trait = NULL,
  set_operation = "union",
  interactive = TRUE,
  std_chromosomes_only = TRUE,
  verbose = FALSE,
  warnings = TRUE
)
get_variants

Arguments

study_id    A character vector of GWAS Catalog study accession identifiers.
association_id    A character vector of GWAS Catalog association identifiers.
variant_id    A character vector of GWAS Catalog variant identifiers.
efo_id    A character vector of EFO identifiers.
pubmed_id    An integer vector of PubMed identifiers.
genomic_range A named list of three vectors:
    chromosome  A character vector of chromosome names of the form 1–22, X or Y.
    start  A numeric vector of start positions, starting at 1.
    end  A numeric vector of end positions.

The three vectors need to be of the same length so that chromosome names, start and end positions can be matched by position.

cytogenetic_band    A character vector of cytogenetic bands of the form '1p36.11'.
gene_name    Gene symbol according to HUGO Gene Nomenclature (HGNC).
efo_trait    A character vector of EFO trait descriptions, e.g., 'uric acid measurement'.
reported_trait A character vector of phenotypic traits as reported by the original authors of the study.
set_operation Either 'union' or 'intersection'. This tells how variants retrieved by different criteria should be combined: 'union' binds together all results removing duplicates and 'intersection' only keeps same variants found with different criteria.
interactive    A logical. If all variants are requested, whether to ask interactively if we really want to proceed.
std_chromosomes_only Whether to return only variants mapped to standard chromosomes: 1 thru 22, X, Y, and MT.
verbose Whether the function should be verbose about the different queries or not.
warnings Whether to print warnings.

Details

Please note that all search criteria are vectorised, thus allowing for batch mode search, e.g., one can search by multiple variant identifiers at once by passing a vector of identifiers to variant_id.

Value

A variants object.
is_ebi_reachable

**Description**

Check if the EBI server where the GWAS Catalog REST API server is running is reachable. This function attempts to connect to https://www.ebi.ac.uk, returning TRUE on success, and FALSE otherwise. Set chatty = TRUE for a step by step description of the connection attempt.

**Usage**

```r
is_ebi_reachable(url = "https://www.ebi.ac.uk", port = 443L, chatty = FALSE)
```

**Arguments**

- `url` NHGRI-EBI GWAS Catalog server URL. Default is https://www.ebi.ac.uk. You should not need to change this parameter.
- `port` Network port on which to ping the server. You should not need to change this parameter.
- `chatty` Whether to be verbose (TRUE) or not (FALSE).

**Value**

A logical value: TRUE if EBI server is reachable, FALSE otherwise.

**Examples**

```r
# Check if the GWAS Catalog Server is reachable
is_ebi_reachable() # Returns TRUE or FALSE.

# Check if the GWAS Catalog Server is reachable
# and show exactly at what step is it failing (if that is the case)
is_ebi_reachable(chatty = TRUE)
```
**Number of GWAS Catalog entities**

**Description**

This function returns the number of unique entities in a GWAS Catalog object.

**Usage**

\[
\text{n}(x, \text{unique} = \text{FALSE})
\]

---

## S4 method for signature 'studies'

\[
\text{n}(x, \text{unique} = \text{FALSE})
\]

## S4 method for signature 'associations'

\[
\text{n}(x, \text{unique} = \text{FALSE})
\]

## S4 method for signature 'variants'

\[
\text{n}(x, \text{unique} = \text{FALSE})
\]

## S4 method for signature 'traits'

\[
\text{n}(x, \text{unique} = \text{FALSE})
\]

**Arguments**

- **x**: A `studies`, an `associations`, a `variants`, or a `traits` object.
- **unique**: Whether to count only unique entries (TRUE) or not (FALSE).

**Value**

An integer scalar.

**Examples**

# Determine number of studies
\[\text{n(studies\_ex01)}\]

# Determine number of associations
\[\text{n(associations\_ex01)}\]

# Determine number of variants
\[\text{n(variants\_ex01)}\]

# Determine number of traits
\[\text{n(traits\_ex01)}\]
open_in_dbsnp

Browse dbSNP from SNP identifiers.

Description
This function launches the web browser at dbSNP and opens a tab for each SNP identifier.

Usage
open_in_dbsnp(variant_id)

Arguments
variant_id A variant identifier, a character vector.

Value
Returns TRUE if successful. Note however that this function is run for its side effect.

Examples
open_in_dbsnp(‘rs56261590’)

open_in_gtex

Browse GTEx from SNP identifiers.

Description
This function launches the web browser at the GTEx Portal and opens a tab for each SNP identifier.

Usage
open_in_gtex(variant_id)

Arguments
variant_id A variant identifier, a character vector.

Value
Returns TRUE if successful. Note however that this function is run for its side effect.

Examples
open_in_gtex(‘rs56261590’)
open_in_gwas_catalog

Browse GWAS Catalog entities from the GWAS Web Graphical User Interface

Description
This function launches the web browser and opens a tab for each identifier on the GWAS web graphical user interface: https://www.ebi.ac.uk/gwas.

Usage
open_in_gwas_catalog(
  identifier,
  gwas_catalog_entity = c("study", "variant", "trait", "gene", "region", "publication")
)

Arguments

identifier
A vector of identifiers. The identifiers can be: study accession identifiers, variant identifiers, EFO trait identifiers, gene symbol names, cytogenetic regions, or PubMed identifiers.

gwas_catalog_entity
Either 'study' (default), 'variant', 'trait', 'gene', 'region' or 'publication', a scalar character. This argument indicates the type of the identifiers passed in identifier.

Value
Returns TRUE if successful, or FALSE otherwise. But note that this function is run for its side effect.

Examples

# Open studies in GWAS Web Graphical User Interface
open_in_gwas_catalog(c('GCST000016', 'GCST001115'))

# Open variants
open_in_gwas_catalog(c('rs146992477', 'rs56261590'),
  gwas_catalog_entity = 'variant')

# Open EFO traits
open_in_gwas_catalog(c('EFO_0004884', 'EFO_0004343'),
  gwas_catalog_entity = 'trait')

# Open genes
open_in_gwas_catalog(c('DPP6', 'MCCC2'),
  gwas_catalog_entity = 'gene')

# Open cytogenetic regions
open_in_gwas_catalog(c('2q37.1', '1p36.11'),)
setop

```r
gwas_catalog_entity = 'region')

# Open publications
open_in_gwas_catalog(c('25533513', '24376627'),
                      gwas_catalog_entity = 'publication')
```

**open_in_pubmed**  
_Browse PubMed from PubMed identifiers._

**Description**

This function launches the web browser and opens a tab for each PubMed citation.

**Usage**

```r
open_in_pubmed(pubmed_id)
```

**Arguments**

- `pubmed_id`  
  A PubMed identifier, either a character or an integer vector.

**Value**

Returns TRUE if successful. Note however that this function is run for its side effect.

**Examples**

```r
open_in_pubmed(c('26301688', '30595370'))
```

**setop**  
_Set operations on GWAS Catalog objects._

**Description**

Performs set union, intersection, and (asymmetric!) difference on two objects of either class studies, associations, variants, or traits. Note that `union()` removes duplicated entities, whereas `bind()` does not.

**Usage**

```r
union(x, y, ...)
intersect(x, y, ...)
setdiff(x, y, ...)
setequal(x, y, ...)
```
Arguments

x, y  Objects of either class studies, associations, variants, or traits.
...  other arguments passed on to methods.

Value

An object of the same class as x and y, i.e., studies, associations, variants, or traits.

Examples

```
# union()
# Combine studies and remove duplicates
union(studies_ex01, studies_ex02)

# Combine associations and remove duplicates
union(associations_ex01, associations_ex02)

# Combine variants and remove duplicates
union(variants_ex01, variants_ex02)

# Combine traits and remove duplicates
union(traits_ex01, traits_ex02)

# intersect()
# Intersect common studies
intersect(studies_ex01, studies_ex02)

# Intersect common associations
intersect(associations_ex01, associations_ex02)

# Intersect common variants
intersect(variants_ex01, variants_ex02)

# Intersect common traits
intersect(traits_ex01, traits_ex02)

# setdiff()
# Remove studies from ex01 that are also present in ex02
setdiff(studies_ex01, studies_ex02)

# Remove associations from ex01 that are also present in ex02
setdiff(associations_ex01, associations_ex02)

# Remove variants from ex01 that are also present in ex02
setdiff(variants_ex01, variants_ex02)
```
# Remove traits from ex01 that are also present in ex02
setdiff(traits_ex01, traits_ex02)

# setequal()

# Compare two studies objects
setequal(studies_ex01, studies_ex01)
setequal(studies_ex01, studies_ex02)

# Compare two associations objects
setequal(associations_ex01, associations_ex01)
setequal(associations_ex01, associations_ex02)

# Compare two variants objects
setequal(variants_ex01, variants_ex01)
setequal(variants_ex01, variants_ex02)

# Compare two traits objects
setequal(traits_ex01, traits_ex01)
setequal(traits_ex01, traits_ex02)

---

# An S4 class to represent a set of GWAS Catalog studies

## Description

The studies object consists of eight slots, each a table (tibble), that combined form a relational database of a subset of GWAS Catalog studies. Each study is an observation (row) in the studies table — main table. All tables have the column study_id as primary key.

## Slots

- **studies**
  - **study_id**  GWAS Catalog study accession identifier, e.g., "GCST002735".
  - **reported_trait**  Phenotypic trait as reported by the authors of the study, e.g. "Breast cancer".
  - **initial_sample_size**  Free text description of the initial cohort sample size.
  - **replication_sample_size**  Free text description of the replication cohort sample size.
  - **gxe**  Whether the study investigates a gene-environment interaction.
  - **gxg**  Whether the study investigates a gene-gene interaction.
  - **snp_count**  Number of variants passing quality control.
  - **qualifier**  Qualifier of number of variants passing quality control.
  - **imputed**  Whether variants were imputed.
  - **pooled**  Whether samples were pooled.
  - **study_design_comment**  Any other relevant study design information.
  - **full_pvalue_set**  Whether full summary statistics are available for this study.
user_requested  Whether the addition of this study to the GWAS Catalog was requested by a user.

genotyping_techs  A tibble listing genotyping technologies employed in each study. Columns:
  study_id  GWAS Catalog study accession identifier.
  genotyping_technology  Genotyping technology employed, e.g., "Exome genotyping array", "Exome-wide sequencing", "Genome-wide genotyping array", "Genome-wide sequencing", or "Targeted genotyping array".

platforms  A tibble listing platforms used per study.
  study_id  GWAS Catalog study accession identifier.
  manufacturer  Platform manufacturer, e.g., "Affymetrix", "Illumina", or "Perlegen".

ancestries  A tibble listing ancestry of samples used in each study.
  study_id  GWAS Catalog study accession identifier.
  ancestry_id  Ancestry identifier.
  type  Stage of the ancestry sample: either 'initial' or 'replication'.
  number_of_individuals  Number of individuals comprising this ancestry sample.

ancestral_groups  A tibble listing ancestral groups used in each ancestry.
  study_id  GWAS Catalog study accession identifier.
  ancestry_id  Ancestry identifier.
  ancestral_group  Genetic ancestry groups present in the sample.

countries_of_origin  A tibble listing countries of origin of samples.
  study_id  GWAS Catalog study accession identifier.
  ancestry_id  Ancestry identifier.
  country_name  Country name, according to The United Nations M49 Standard of Geographic Regions.
  major_area  Region name, according to The United Nations M49 Standard of Geographic Regions.
  region  Sub-region name, according to The United Nations M49 Standard of Geographic Regions.

countries_of_recruitment  A tibble listing countries of recruitment of samples.
  study_id  GWAS Catalog study accession identifier.
  ancestry_id  Ancestry identifier.
  country_name  Country name, according to The United Nations M49 Standard of Geographic Regions.
  major_area  Region name, according to The United Nations M49 Standard of Geographic Regions.
  region  Sub-region name, according to The United Nations M49 Standard of Geographic Regions.

publications  A tibble listing publications associated with each study.
  study_id  GWAS Catalog study accession identifier.
  pubmed_id  PubMed identifier.
  publication_date  Publication date (online date if available) formatted as ymd.
### study_to_association

Map a study id to an association id

**Description**

Map a study accession identifier to an association accession identifier.

**Usage**

```r
study_to_association(study_id, verbose = FALSE, warnings = TRUE)
```

**Arguments**

- `study_id`: A character vector of study accession identifiers.
- `verbose`: Whether the function should be verbose about the different queries or not.
- `warnings`: Whether to print warnings.

**Value**

A dataframe of two identifiers. First column is the study identifier and the second column is the association identifier.

**Examples**

```r
# Map GWAS study identifiers to association identifiers
study_to_association(c("GCST001084", "GCST001085"))
```

### study_to_trait

Map a study id to an EFO trait id

**Description**

Map a study accession identifier to an EFO trait identifier.

**Usage**

```r
study_to_trait(study_id, verbose = FALSE, warnings = TRUE)
```
Arguments

- **study_id**: A character vector of study accession identifiers.
- **verbose**: Whether the function should be verbose about the different queries or not.
- **warnings**: Whether to print warnings.

Value

A dataframe of two identifiers. First column is the study identifier and the second column is the EFO identifier.

Examples

```r
# Map GWAS study identifiers to EFO trait identifiers
study_to_trait(c("GCST001084", "GCST001085"))
```

---

**study_to_variant**

*Map a study id to a variant id*

Description

Map a study accession identifier to a variant accession identifier.

Usage

```r
study_to_variant(study_id, verbose = FALSE, warnings = TRUE)
```

Arguments

- **study_id**: A character vector of study accession identifiers.
- **verbose**: Whether the function should be verbose about the different queries or not.
- **warnings**: Whether to print warnings.

Value

A dataframe of two identifiers. First column is the study identifier and the second column is the variant identifier.

Examples

```r
# Map GWAS study identifiers to variant identifiers
study_to_variant(c("GCST001084", "GCST001085"))
```
subset-associations  Subset an associations object

Description
You can subset associations by identifier or by position using the `[]` operator.

Usage
```r
## S4 method for signature 'associations,missing,missing,missing'
x[i, j, ..., drop = FALSE]

## S4 method for signature 'associations,numeric,missing,missing'
x[i, j, ..., drop = FALSE]

## S4 method for signature 'associations,character,missing,missing'
x[i, j, ..., drop = FALSE]
```

Arguments
- `x`: A associations object.
- `i`: Position of the identifier or the name of the identifier itself.
- `j`: Not used.
- `...`: Additional arguments not used here.
- `drop`: Not used.

Value
A associations object.

Examples
```r
# Subset an associations object by identifier
associations_ex01["22505"]

# Or by its position in table associations
associations_ex01[2]

# Keep all associations except the second
associations_ex01[-2]
```
Description

You can subset studies by identifier or by position using the `[]` operator.

Usage

```r
## S4 method for signature 'studies,missing,missing,missing'
x[i, j, ..., drop = FALSE]
## S4 method for signature 'studies,numeric,missing,missing'
x[i, j, ..., drop = FALSE]
## S4 method for signature 'studies,character,missing,missing'
x[i, j, ..., drop = FALSE]
```

Arguments

- `x`: A studies object.
- `i`: Position of the identifier or the name of the identifier itself.
- `j`: Not used.
- `...`: Additional arguments not used here.
- `drop`: Not used.

Value

A studies object.

Examples

```r
# Subset a studies object by identifier
studies_ex01['GCST001585']

# Or by its position in table studies
studies_ex01[1]

# Keep all studies except the first
studies_ex01[-1]
```
Description

You can subset traits by identifier or by position using the `[]` operator.

Usage

```r
## S4 method for signature 'traits,missing,missing,missing'
x[i, j, ..., drop = FALSE]

## S4 method for signature 'traits,numeric,missing,missing'
x[i, j, ..., drop = FALSE]

## S4 method for signature 'traits,character,missing,missing'
x[i, j, ..., drop = FALSE]
```

Arguments

- `x`: A traits object.
- `i`: Position of the identifier or the name of the identifier itself.
- `j`: Not used.
- `...`: Additional arguments not used here.
- `drop`: Not used.

Value

A traits object.

Examples

```r
# Subset a traits object by identifier
traits_ex01['EFO_0004884']

# Or by its position in table traits
traits_ex01[1]

# Keep all traits except the second
traits_ex01[-2]
```
## Description

You can subset `variants` by identifier or by position using the `\[\` operator.

## Usage

```r
## S4 method for signature 'variants,missing,missing,missing'
x[i, j, ..., drop = FALSE]
```

```r
## S4 method for signature 'variants,numeric,missing,missing'
x[i, j, ..., drop = FALSE]
```

```r
## S4 method for signature 'variants,character,missing,missing'
x[i, j, ..., drop = FALSE]
```

## Arguments

- `x`: A `variants` object.
- `i`: Position of the identifier or the name of the identifier itself.
- `j`: Not used.
- `...`: Additional arguments not used here.
- `drop`: Not used.

## Value

A `variants` object.

## Examples

```r
# Subset a variants object by identifier
variants_ex01[rs4725504]

# Or by its position in table variants
variants_ex01[3]

# Keep all variants except the third
variants_ex01[-3]
```
traits-class

An S4 class to represent a set of GWAS Catalog EFO traits.

Description

The traits object consists of one slot only, a table (tibble) of GWAS Catalog EFO traits. Each EFO trait is an observation (row) in the traits table — main table.

Slots

- **traits**: A tibble listing EFO traits. Columns:
  - **efo_id**: EFO identifier.
  - **trait**: EFO trait description.
  - **uri**: The full URI of the EFO term.

trait_to_association

Map an EFO trait id to an association id

Description

Map an EFO trait id to an association identifier.

Usage

```r
trait_to_association(efo_id, verbose = FALSE, warnings = TRUE)
```

Arguments

- **efo_id**: A character vector of EFO trait identifiers.
- **verbose**: Whether the function should be verbose about the different queries or not.
- **warnings**: Whether to print warnings.

Value

A dataframe of two identifiers. First column is the EFO trait identifier and the second column is the association identifier.

Examples

```r
# Map EFO trait identifiers to association identifiers
trait_to_association(c('EFO_0005108', 'EFO_0005109'))
```
trait_to_study  

Map an EFO trait id to a study id

Description

Map an EFO trait id to a study accession identifier.

Usage

```
trait_to_study(efo_id, verbose = FALSE, warnings = TRUE)
```

Arguments

- `efo_id`: A character vector of EFO trait identifiers.
- `verbose`: Whether the function should be verbose about the different queries or not.
- `warnings`: Whether to print warnings.

Value

A dataframe of two identifiers. First column is the EFO trait identifier and the second column is the study identifier.

Examples

```
# Map EFO trait identifiers to study identifiers
trait_to_study(c('EFO_0005108', 'EFO_0005109'))
```

trait_to_variant  

Map an EFO trait id to a variant id

Description

Map an EFO trait id to a variant identifier.

Usage

```
trait_to_variant(efo_id, verbose = FALSE, warnings = TRUE)
```

Arguments

- `efo_id`: A character vector of EFO trait identifiers.
- `verbose`: Whether the function should be verbose about the different queries or not.
- `warnings`: Whether to print warnings.
variants-class

Value

A dataframe of two identifiers. First column is the EFO trait identifier and the second column is the variant identifier.

Examples

```r
# Map EFO trait identifiers to variant identifiers
trait_to_variant('EFO_0005229')
```

variants-class

An S4 class to represent a set of GWAS Catalog variants

Description

The variants object consists of four slots, each a table (tibble), that combined form a relational database of a subset of GWAS Catalog variants. Each variant is an observation (row) in the variants table — main table. All tables have the column variant_id as primary key.

Slots

variants A tibble listing variants. Columns:

- **variant_id** Variant identifier, e.g., 'rs1333048'.
- **merged** Whether this SNP has been merged with another SNP in a newer genome build.
- **functional_class** Class according to Ensembl’s predicted consequences that each variant allele may have on transcripts. See Ensembl Variation - Calculated variant consequences.
- **chromosome_name** Chromosome name.
- **chromosome_position** Chromosome position.
- **chromosome_region** Cytogenetic location.
- **last_update_date** Last time this variant was updated.

genomic_contexts A tibble listing genomic contexts associated with each variant. Columns:

- **variant_id** Variant identifier.
- **gene_name** Gene symbol according to HUGO Gene Nomenclature (HGNC).
- **chromosome_name** Chromosome name.
- **chromosome_position** Chromosome position.
- **distance** Genomic distance between the variant and the gene (in base pairs).
- **is_mapped_gene** Whether this is a mapped gene to this variant. A mapped gene is either an overlapping gene with the variant or the two closest genes upstream and downstream of the variant. Moreover, only genes whose mapping source is 'Ensembl' are considered.
- **is_closest_gene** Whether this is the closest gene to this variant.
- **is_intergenic** Whether this variant is intergenic, i.e., if there is no gene up or downstream within 100kb.
- **is_upstream** Whether this variant is upstream of this gene.
- **is_downstream** Whether this variant is downstream of this gene.
source Gene mapping source, either Ensembl or NCBI.
mapping_method Gene mapping method.

ensmbil_ids A tibble listing gene Ensembl identifiers associated with each genomic context. Columns:
  variant_id Variant identifier.
  gene_name Gene symbol according to HUGO Gene Nomenclature (HGNC).
  ensembl_id The Ensembl identifier of an Ensembl gene, see Section Gene annotation in Ensembl for more information.

entrez_ids A tibble listing gene Entrez identifiers associated with each genomic context. Columns:
  variant_id Variant identifier.
  gene_name Gene symbol according to HUGO Gene Nomenclature (HGNC).
  entrez_id The Entrez identifier of a gene, see ref. doi: 10.1093/nar/gkq1237 for more information.

```
variant_to_association

   Map a variant id to an association id
```

Description

Map a variant identifier to an association identifier.

Usage

```
variant_to_association(variant_id, verbose = FALSE, warnings = TRUE)
```

Arguments

  variant_id A character vector of variant identifiers.
  verbose Whether the function should be verbose about the different queries or not.
  warnings Whether to print warnings.

Value

A dataframe of two identifiers. First column is the variant identifier and the second column is the association identifier.

Examples

```
# Map GWAS variant identifiers to association identifiers
variant_to_association(c('rs7904579', 'rs138331350'))
```
**variant_to_study**  \hspace{1cm} Map a variant id to a study id

### Description
Map a variant identifier to a study accession identifier.

### Usage
```r
variant_to_study(variant_id, verbose = FALSE, warnings = TRUE)
```

### Arguments
- **variant_id**: A character vector of variant identifiers.
- **verbose**: Whether the function should be verbose about the different queries or not.
- **warnings**: Whether to print warnings.

### Value
A dataframe of two identifiers. First column is the variant identifier and the second column is the study identifier.

### Examples
```r
# Map GWAS variant identifiers to study identifiers
variant_to_study(c("rs7904579", "rs138331350"))
```

---

**variant_to_trait**  \hspace{1cm} Map a variant id to an EFO trait

### Description
Map a variant identifier to an EFO trait identifier. Variants are first mapped to association identifiers, and then to EFO traits. Set the option `keep_association_id = TRUE` to keep the intermediate mapping, i.e., the association identifiers.

### Usage
```r
variant_to_trait(
  variant_id,
  keep_association_id = FALSE,
  verbose = FALSE,
  warnings = TRUE
)
```
Arguments

- **variant_id**: A character vector of variant identifiers.
- **keep_association_id**: Whether to keep the association identifier in the final output (default is `FALSE`).
- **verbose**: Whether the function should be verbose about the different queries or not.
- **warnings**: Whether to print warnings.

**Value**

A dataframe of two or three identifiers. If `keep_association_id` is set to `FALSE`, the first column is the variant identifier and the second column is the EFO trait identifier, otherwise the variable `association_id` is also included as the second column.

**Examples**

```r
# Map GWAS variant identifiers to EFO trait identifiers
variant_to_trait(c('rs7904579', 'rs138331350'))

# Map GWAS variant identifiers to EFO trait identifiers
# but keep the intermediate association identifier
variant_to_trait(c('rs7904579', 'rs138331350'), keep_association_id = TRUE)
```

---

**Pipe operator**

**Description**

See `magrittr::%>%` for details.

**Value**

The same as the rhs.

**Examples**

```r
c(1,2,3) %>% mean()
```
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