

Package ‘quincunx’

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Type Package

Title REST API Client for the 'PGS' Catalog

Version 0.1.5

Description Programmatic access to the 'PGS' Catalog.

This package provides easy access to 'PGS' Catalog data by accessing the REST API <<https://www.pgscatalog.org/rest/>>.

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Collate 'ancestry_categories.R' 'class-cohorts.R'
'class-performance_metrics.R' 'class-publications.R'
'class-releases.R' 'class-sample_sets.R' 'class-scores.R'
'class-trait_categories.R' 'class-traits.R' 'clear_cache.R'
'contains_question_mark.R' 'count.R' 'drop_metadata_cols.R'
'first_non_na.R' 'generics.R' 'get.R'
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'get_performance_metrics.R' 'get_publications.R'
'get_releases.R' 'get_sample_sets.R' 'get_scores.R'
'get_trait_categories.R' 'get_traits.R' 'id_mapping.R'
'is_json_empty.R' 'is_paginated.R' 'is_pgs_id.R'
'is_pubmed_id.R' 'messages.R' 'n_pages.R' 'nr_to_na.R'
'offsets.R' 'open_in_dbsnp.R' 'open_in_pgs_catalog.R'
'open_in_pubmed.R' 'parse-ancestry_categories.R'
'parse-cohorts.R' 'parse-performance_metrics.R'
'parse-publications.R' 'parse-releases.R' 'parse-sample_sets.R'
'parse-scores.R' 'parse-trait_categories.R' 'parse-traits.R'
'read_file_column_names.R' 'read_pgs_scoring_file.R'
'relocate_metadata_cols.R' 'remap_id.R' 'request.R'

's4-utils.R' 'stages.R' 'sure.R' 'unwrap_cohort.R'
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 'unwrap_publication.R' 'unwrap_sample.R' 'utils-pipe.R'
 'warnings.R' 'write_xlsx.R'

Depends R (>= 2.10)

URL <https://github.com/maialab/quincunx>

BugReports <https://github.com/maialab/quincunx/issues>

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ancestry_categories *Ancestry categories and classes*

Description

A dataset containing the ancestry categories defined in NHGRI-EBI GWAS Catalog framework (Table 1, [doi:10.1186/s1305901813962](https://doi.org/10.1186/s1305901813962)). Ancestry categories are assigned to samples with distinct and well-defined patterns of genetic variation. You will find these categories in the variable `ancestry_category` of the following objects: `scores`, `performance_metrics` and `sample_sets`. Ancestry categories (`ancestry_category`) are further clustered into ancestry classes (`ancestry_class`).

Usage

`ancestry_categories`

Format

A data frame with 19 ancestry categories (rows) and 6 columns:

ancestry_category Ancestry category.

ancestry_class To reduce the complexity associated with the many ancestry categories, some have been merged into higher-level groupings (`ancestry_class`). These groupings represent the current breadth of data in the PGS Catalog and are likely to change as more data is added.

ancestry_class_symbol 3-letter code for the `ancestry_class` e.g. "EUR" or "MAE".

ancestry_class_colour Hexadecimal colour code associated with ancestry groupings (`ancestry_class`). This can be useful when visually communicating about ancestries.

definition Description of the ancestry category.

examples Examples of detailed descriptions of sample ancestries included in the category.

Source

Table 1 of Morales et al. (2018): [doi:10.1186/s1305901813962](https://doi.org/10.1186/s1305901813962)

PGS Catalog Ancestry Documentation: <http://www.pgscatalog.org/docs/ancestry/>

Examples

```
ancestry_categories
```

bind	<i>Bind PGS Catalog objects</i>
------	---------------------------------

Description

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

Usage

```
bind(x, ...)
```

Arguments

x	An object of either class <code>scores</code> , <code>publications</code> , <code>traits</code> , <code>performance_metrics</code> , <code>sample_sets</code> , <code>cohorts</code> or <code>trait_categories</code> .
...	Objects of the same class as x.

Value

An object of the same class as x.

Examples

```
# Get some `scores` objects:
my_scores_1 <- get_scores(c('PGS000012', 'PGS000013'))
my_scores_2 <- get_scores(c('PGS000013', 'PGS000014'))

# NB: with `bind()`, PGS000013 is repeated (as opposed to `union()`)
bind(my_scores_1, my_scores_2)@scores
```

clear_cache	<i>Clear quincunx cache of memoised functions</i>
-------------	---

Description

quincunx uses memoised functions for the REST API calls. Use this function to reset the cache.

Usage

```
clear_cache()
```

Value

Returns a logical value, indicating whether the resetting of the cache was successful (TRUE) or not FALSE.

Examples

```
clear_cache()
```

cohorts-class	<i>An S4 class to represent a set of cohorts</i>
---------------	--

Description

The cohorts object consists of two tables (slots) that combined form a relational database of a subset of cohorts. Each cohort is an observation (row) in the cohorts table (first table).

Slots

cohorts A table of cohorts. Each cohort (row) is identified by its cohort_symbol. Columns:

cohort_symbol Cohort symbol. Example: "CECILE".

cohort_name Cohort full name. Example: "CECILE Breast Cancer Study".

pgs_ids A table of cohorts and their associated polygenic scores identifiers. Columns:

cohort_symbol Cohort symbol. Example: "CECILE".

pgs_id Polygenic Score (PGS) identifier.

stage Sample stage: either "gwas/dev" or "eval".

```
get_ancestry_categories
```

Get ancestry categories and classes

Description

Retrieves ancestry categories and classes. This function simply returns the object [ancestry_categories](#).

Usage

```
get_ancestry_categories()
```

Value

A tibble with ancestry categories, classes and associated information. See [ancestry_categories](#) for details about each column.

Examples

```
get_ancestry_categories()
```

```
get_cohorts
```

Get PGS Catalog Cohorts

Description

Retrieves cohorts via the PGS Catalog REST API. Please note that all cohort_symbol is vectorised, thus allowing for batch mode search.

Usage

```
get_cohorts(
  cohort_symbol = NULL,
  verbose = FALSE,
  warnings = TRUE,
  progress_bar = TRUE
)
```

Arguments

cohort_symbol	A cohort symbol or NULL if all cohorts are intended.
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.
progress_bar	Whether to show a progress bar indicating download progress from the REST API server.

Value

A [cohorts](#) object.

Examples

```
# Get information about specific cohorts by their symbols (acronyms)
get_cohorts(cohort_symbol = c('23andMe', 'IPOBCS'))

# Get info on all cohorts (may take a few minutes to download)
## Not run:
get_cohorts()

## End(Not run)
```

```
get_performance_metrics
```

Get PGS Catalog Performance Metrics

Description

Retrieves performance metrics via the PGS Catalog REST API. The REST API is queried multiple times with the criteria passed as arguments (see below). By default all performance metrics 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_performance_metrics(
  ppm_id = NULL,
  pgs_id = NULL,
  set_operation = "union",
  interactive = TRUE,
  verbose = FALSE,
  warnings = TRUE,
  progress_bar = TRUE
)
```

Arguments

<code>ppm_id</code>	A character vector of PGS Catalog performance metrics accession identifiers.
<code>pgs_id</code>	A character vector of PGS Catalog score accession identifiers.
<code>set_operation</code>	Either 'union' or 'intersection'. This tells how performance metrics retrieved by different criteria should be combined: 'union' binds together all results removing duplicates and 'intersection' only keeps same performance metrics found with different criteria.

interactive	A logical. If all performance metrics 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.
progress_bar	Whether to show a progress bar as the queries are performed.

Details

Please note that all search criteria are vectorised, thus allowing for batch mode search.

Value

A [performance_metrics](#) object.

Examples

```
## Not run:
# Get performance metrics catalogued with identifier 'PPM000001'
get_performance_metrics(ppm_id = 'PPM000001')

# Get performance metrics associated with polygenic score id 'PGS000001'
get_performance_metrics(pgs_id = 'PGS000001')

# To retrieve all catalogued performed metrics in PGS Catalog you simply
# leave the parameters `ppm_id` and `pgs_id` as `NULL`.
get_performance_metrics()

## End(Not run)
```

get_publications	<i>Get PGS Catalog Publications</i>
------------------	-------------------------------------

Description

Retrieves PGS publications via the PGS Catalog REST API. The REST API is queried multiple times with the criteria passed as arguments (see below). By default all publications 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_publications(  
  pgp_id = NULL,  
  pgs_id = NULL,  
  pubmed_id = NULL,  
  author = NULL,  
  set_operation = "union",  
  interactive = TRUE,  
  verbose = FALSE,  
  warnings = TRUE,  
  progress_bar = TRUE  
)
```

Arguments

pgp_id	A character vector of PGS Catalog publication accession identifiers.
pgs_id	A character vector of PGS Catalog score accession identifiers.
pubmed_id	An integer vector of PubMed identifiers.
author	A character vector of author names, any author in the list of authors in a publication, .e.g. 'Mavaddat'.
set_operation	Either 'union' or 'intersection'. This tells how publications retrieved by different criteria should be combined: 'union' binds together all results removing duplicates and 'intersection' only keeps same publications found with different criteria.
interactive	A logical. If all publications 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.
progress_bar	Whether to show a progress bar as the queries are performed.

Details

Please note that all search criteria are vectorised, thus allowing for batch mode search. For more details see the help vignette: `vignette("getting-pgs-publications", package = "quincunx")`.

Value

A `publications` object.

Examples

```
## Not run:  
# Get PGS publications by their identifier  
get_publications(pgp_id = c('PGP000001', 'PGP000002'))  
  
# By polygenic score identifier
```

```

get_publications(pgs_id = 'PGS000003')

# By PubMed identifier
get_publications(pubmed_id = '30554720')

# By author's last name
get_publications(author = 'Natarajan')

## End(Not run)

```

get_releases

Get PGS Catalog Releases

Description

This function retrieves PGS Catalog release information. Note that the columns `pgs_id`, `ppm_id` and `pgp_id` contain in each element a vector. These columns can be unnested using [unnest_longer](#) (see Examples).

Usage

```

get_releases(
  date = "latest",
  verbose = FALSE,
  warnings = TRUE,
  progress_bar = TRUE
)

```

Arguments

<code>date</code>	One or more dates formatted as "YYYY-MM-DD" for respective releases, "latest" for the latest release, or "all" for all releases.
<code>verbose</code>	Whether to print information about the underlying requests to the REST API server.
<code>warnings</code>	Whether to print warnings about the underlying requests to the REST API server.
<code>progress_bar</code>	Whether to show a progress bar indicating download progress from the REST API server.

Value

A data frame where each row is a release. Columns are:

date Release date.

n_pgs Number of released Polygenic Score (PGS) identifiers (`pgs_id`).

n_ppm Number of released Performance Metric (PPM) identifiers (`ppm_id`).

n_pgp Number of released PGS Catalog Publication (PGP) identifiers (`pgp_id`).

pgs_id Released Polygenic Score (PGS) identifiers.
ppm_id Released Performance Metric (PPM) identifiers.
pgp_id Released PGS Catalog Publication (PGP) identifiers.
notes News about the release.

Examples

```
## Not run:  
# Get the latest release  
get_releases()  
get_releases(date = 'latest')  
  
# Get all releases  
get_releases(date = 'all')  
  
# Get a specific release by date  
get_releases(date = '2020-08-19')  
  
## End(Not run)
```

get_sample_sets	<i>Get PGS Catalog Sample Sets</i>
-----------------	------------------------------------

Description

Retrieves sample sets via the PGS Catalog REST API. The REST API is queried multiple times with the criteria passed as arguments (see below). By default all sample sets 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_sample_sets(  
  pss_id = NULL,  
  pgs_id = NULL,  
  set_operation = "union",  
  interactive = TRUE,  
  verbose = FALSE,  
  warnings = TRUE,  
  progress_bar = TRUE  
)
```

Arguments

pss_id	A character vector of PGS Catalog sample sets accession identifiers.
pgs_id	A character vector of PGS Catalog score accession identifiers.
set_operation	Either 'union' or 'intersection'. This tells how performance metrics retrieved by different criteria should be combined: 'union' binds together all results removing duplicates and 'intersection' only keeps same sample sets found with different criteria.
interactive	A logical. If all sample sets 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.
progress_bar	Whether to show a progress bar indicating download progress from the REST API server.

Details

Please note that all search criteria are vectorised, thus allowing for batch mode search.

Value

A [sample_sets](#) object.

Examples

```
## Not run:
# Search by PGS identifier
get_sample_sets(pgs_id = 'PGS000013')

# Search by the PSS identifier
get_sample_sets(pss_id = 'PSS000068')

## End(Not run)
```

get_scores

Get PGS Catalog Scores

Description

Retrieves polygenic scores via the PGS Catalog REST API. The REST API is queried multiple times with the criteria passed as arguments (see below). By default all scores 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_scores(  
  pgs_id = NULL,  
  efo_id = NULL,  
  pubmed_id = NULL,  
  set_operation = "union",  
  interactive = TRUE,  
  verbose = FALSE,  
  warnings = TRUE,  
  progress_bar = TRUE  
)
```

Arguments

pgs_id	A character vector of PGS Catalog score accession identifiers.
efo_id	A character vector of EFO identifiers.
pubmed_id	An integer vector of PubMed identifiers.
set_operation	Either 'union' or 'intersection'. This tells how scores retrieved by different criteria should be combined: 'union' binds together all results removing duplicates and 'intersection' only keeps same scores found with different criteria.
interactive	A logical. If all scores 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.
progress_bar	Whether to show a progress bar as the queries are performed.

Details

Please note that all search criteria are vectorised, thus allowing for batch mode search.

Value

A [scores](#) object.

Examples

```
## Not run:  
# By `pgs_id`  
get_scores(pgs_id = 'PGS000088')  
  
# By `efo_id`  
get_scores(efo_id = 'EFO_0007992')  
  
# By `pubmed_id`  
get_scores(pubmed_id = '25748612')
```

```
## End(Not run)
```

```
get_traits
```

```
Get PGS Catalog Traits
```

Description

Retrieves traits via the PGS 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(
  efo_id = NULL,
  trait_term = NULL,
  exact_term = TRUE,
  include_children = FALSE,
  set_operation = "union",
  interactive = TRUE,
  verbose = FALSE,
  warnings = TRUE,
  progress_bar = TRUE
)
```

Arguments

<code>efo_id</code>	A character vector of EFO identifiers.
<code>trait_term</code>	A character vector of terms to be matched against trait identifiers (<code>efo_id</code>), trait descriptions, synonyms thereof, externally mapped terms, or even trait categories.
<code>exact_term</code>	A logical value, indicating whether to match the <code>trait_term</code> exactly (TRUE) or not (FALSE).
<code>include_children</code>	A logical value, indicating whether to include child traits or not.
<code>set_operation</code>	Either 'union' or 'intersection'. This tells how performance metrics retrieved by different criteria should be combined: 'union' binds together all results removing duplicates and 'intersection' only keeps same sample sets found with different criteria.
<code>interactive</code>	A logical. If all traits are requested, whether to ask interactively if we really want to proceed.
<code>verbose</code>	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.
progress_bar	Whether to show a progress bar indicating download progress from the REST API server.

Details

Please note that all search criteria are vectorised, thus allowing for batch mode search.

Value

A [traits](#) object.

Examples

```
## Not run:
# Get a trait by its EFO identifier
get_traits(efo_id = 'EFO_0004631')

# Get a trait by matching a term in EFO identifier (`efo_id`), label,
# description synonyms, categories, or external mapped terms
get_traits(trait_term = 'stroke', exact_term = FALSE)

# Get a trait matching its name (`trait`) exactly (default)
get_traits(trait_term = 'stroke', exact_term = TRUE)

# Get traits, excluding its children traits (default)
get_traits(trait_term = 'breast cancer')

# Get traits, including its children traits (check column `is_child` for
# child traits)
get_traits(trait_term = 'breast cancer', include_children = TRUE)

## End(Not run)
```

get_trait_categories *Get PGS Catalog Trait Categories*

Description

Retrieves all trait categories via the PGS Catalog REST API.

Usage

```
get_trait_categories(verbose = FALSE, warnings = TRUE, progress_bar = TRUE)
```

Arguments

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.
progress_bar	Whether to show a progress bar indicating download progress from the REST API server.

Value

A [trait_categories](#) object.

Examples

```
get_trait_categories(progress_bar = FALSE)
```

n

Number of PGS Catalog entities

Description

This function returns the number of entities in a PGS Catalog object. To avoid ambiguity with `dplyr::n()` use `quincunx::n()`.

Usage

```
n(x, unique = FALSE)

## S4 method for signature 'scores'
n(x, unique = FALSE)

## S4 method for signature 'publications'
n(x, unique = FALSE)

## S4 method for signature 'traits'
n(x, unique = FALSE)

## S4 method for signature 'performance_metrics'
n(x, unique = FALSE)

## S4 method for signature 'sample_sets'
n(x, unique = FALSE)

## S4 method for signature 'cohorts'
n(x, unique = FALSE)
```



```
## S4 method for signature 'trait_categories'
n(x, unique = FALSE)

## S4 method for signature 'releases'
n(x, unique = FALSE)
```

Arguments

x	A scores , publications , traits , performance_metrics , sample_sets , cohorts , trait_categories or releases object.
unique	Whether to count only unique entries (TRUE) or not (FALSE).

Value

An integer scalar.

Examples

```
# Return the number of polygenic scores in a scores object:
my_scores <- get_scores(pgs_id = c('PGS000007', 'PGS000007', 'PGS000042'))
n(my_scores)

# If you want to count unique scores only, then use the `unique` parameter:
n(my_scores, unique = TRUE)

# Total number of curated publications in the PGS Catalog:
all_pub <- get_publications(interactive = FALSE, progress_bar = FALSE)
n(all_pub)

# Total number of curated traits in the PGS Catalog:
all_traits <- get_traits(interactive = FALSE, progress_bar = FALSE)
n(all_traits)
```

open_in_dbsnp	<i>Browse dbSNP from SNP identifiers.</i>
---------------	---

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_pgs_catalog	<i>Browse PGS Catalog entities from the PGS Catalog Web Graphical User Interface</i>
---------------------	--

Description

This function launches the web browser and opens a tab for each identifier on the PGS Catalog web graphical user interface: <https://www.pgscatalog.org/>.

Usage

```
open_in_pgs_catalog(  
  identifier = NULL,  
  pgs_catalog_entity = c("pgs", "pgp", "pss", "efo")  
)
```

Arguments

identifier	A vector of identifiers. The identifiers can be: PGS, PGP, PSS or EFO identifiers.
pgs_catalog_entity	Either 'pgs' (default), 'pgp', 'pss', 'efo'. 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 in PGS scores Catalog Web Graphical User Interface  
open_in_pgs_catalog(c('PGS000001', 'PGS000002'))  
  
# Open PGS Catalog Publications  
open_in_pgs_catalog(c('PGP000001', 'PGP000002'),  
  pgs_catalog_entity = 'pgp')  
  
# Open Sample Sets (PSS)
```

```

open_in_pgs_catalog(c('PSS000001', 'PSS000002'),
  pgs_catalog_entity = 'pss')

# Open EFO traits (EFO)
open_in_pgs_catalog(c('EFO_0001645', 'MONDO_0007254'),
  pgs_catalog_entity = 'efo')

```

open_in_pubmed *Browse PubMed from PubMed identifiers.*

Description

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

Usage

```
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

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

performance_metrics-class
An S4 class to represent a set of PGS Catalog Performance Metrics

Description

The performance_metrics object consists of nine tables (slots) that combined form a relational database of a subset of performance metrics. Each performance metric is an observation (row) in the scores table (first table).

Slots

`performance_metrics` A table of PGS Performance Metrics (PPM). Each PPM (row) is uniquely identified by the `ppm_id` column. Columns:

ppm_id A PGS Performance Metrics identifier. Example: "PPM000001".

pgs_id Polygenic Score (PGS) identifier.

reported_trait The author-reported trait that the PGS has been developed to predict. Example: "Breast Cancer".

covariates Comma-separated list of covariates used in the prediction model to evaluate the PGS.

comments Any other information relevant to the understanding of the performance metrics.

`publications` A table of publications. Each publication (row) is uniquely identified by the column `pgp_id`. Columns:

ppm_id A PGS Performance Metrics identifier. Example: "PPM000001".

pgp_id PGS Publication identifier. Example: "PGP000001".

pubmed_id PubMed identifier. Example: "25855707".

publication_date Publication date. Example: "2020-09-28". Note that the class of `publication_date` is `Date`.

publication Abbreviated name of the journal. Example: "Am J Hum Genet".

title Publication title.

author_fullname First author of the publication. Example: 'Mavaddat N'.

doi Digital Object Identifier (DOI). This variable is also curated to allow unpublished work (e.g. preprints) to be added to the catalog. Example: "10.1093/jnci/djv036".

`sample_sets` A table of sample sets. Each sample set (row) is uniquely identified by the column `pss_id`. Columns:

ppm_id A PGS Performance Metrics identifier. Example: "PPM000001".

pss_id A PGS Sample Set identifier. Example: "PSS000042".

`samples` A table of samples. Each sample (row) is uniquely identified by the combination of values from the columns: `ppm_id`, `pss_id`, and `sample_id`. Columns:

ppm_id A PGS Performance Metrics identifier. Example: "PPM000001".

pss_id A PGS Sample Set identifier. Example: "PSS000042".

sample_id Sample identifier. This is a surrogate key to identify each sample.

stage Sample stage: should be always Evaluation ("eval").

sample_size Number of individuals included in the sample.

sample_cases Number of cases.

sample_controls Number of controls.

sample_percent_male Percentage of male participants.

phenotype_description Detailed phenotype description.

ancestry_category Author reported ancestry is mapped to the best matching ancestry category from the NHGRI-EBI GWAS Catalog framework (see [ancestry_categories](#)) for possible values.

ancestry A more detailed description of sample ancestry that usually matches the most specific description described by the authors (e.g. French, Chinese).

country Author reported countries of recruitment (if available).

ancestry_additional_description Any additional description not captured in the other columns (e.g. founder or genetically isolated populations, or further description of admixed samples).

study_id Associated GWAS Catalog study accession identifier, e.g., "GCST002735".

pubmed_id PubMed identifier.

cohorts_additional_description Any additional description about the samples (e.g. sub-cohort information).

demographics A table of sample demographics' variables. Each demographics' variable (row) is uniquely identified by the combination of values from the columns: ppm_id, pss_id, sample_id, and variable. Columns:

ppm_id A PGS Performance Metrics identifier. Example: "PPM000001".

pss_id A PGS Sample Set identifier. Example: "PSS000042".

sample_id Sample identifier. This is a surrogate identifier to identify each sample.

variable Demographics variable. Following columns report about the indicated variable.

estimate_type Type of statistical estimate for variable.

estimate The variable's statistical value.

unit Unit of the variable.

variability_type Measure of statistical dispersion for variable, e.g. standard error (se) or standard deviation (sd).

variability The value of the measure of dispersion.

interval_type Type of statistical interval for variable: range, iqr (interquartile), ci (confidence interval).

interval_lower Interval lower bound.

interval_upper Interval upper bound.

cohorts A table of cohorts. Each cohort (row) is uniquely identified by the combination of values from the columns: ppm_id, sample_id and cohort_symbol. Columns:

ppm_id A PGS Performance Metrics identifier. Example: "PPM000001".

sample_id Sample identifier. This is a surrogate key to identify each sample.

cohort_symbol Cohort symbol.

cohort_name Cohort full name.

pgs_effect_sizes A table of effect sizes per standard deviation change in PGS. Examples include regression coefficients (betas) for continuous traits, odds ratios (OR) and/or hazard ratios (HR) for dichotomous traits depending on the availability of time-to-event data. Each effect size is uniquely identified by the combination of values from the columns: ppm_id and effect_size_id. Columns:

ppm_id A PGS Performance Metrics identifier. Example: "PPM000001".

effect_size_id Effect size identifier. This is a surrogate identifier to identify each effect size.

estimate_type_long Long notation of the effect size (e.g. Odds Ratio).

estimate_type Short notation of the effect size (e.g. OR).

estimate The estimate's value.

unit Unit of the estimate.

variability_type Measure of statistical dispersion for variable, e.g. standard error (se) or standard deviation (sd).

variability The value of the measure of dispersion.

interval_type Type of statistical interval for variable: range, iqr (interquartile), ci (confidence interval).

interval_lower Interval lower bound.

interval_upper Interval upper bound.

pgs_classification_metrics A table of classification metrics. Examples include the Area under the Receiver Operating Characteristic (AUROC) or Harrell's C-index (Concordance statistic). Columns:

ppm_id A PGS Performance Metrics identifier. Example: "PPM000001".

classification_metrics_id Classification metric identifier. This is a surrogate identifier to identify each classification metric.

estimate_type_long Long notation of the classification metric (e.g. Concordance Statistic).

estimate_type Short notation classification metric (e.g. C-index).

estimate The estimate's value.

unit Unit of the estimate.

variability_type Measure of statistical dispersion for variable, e.g. standard error (se) or standard deviation (sd).

variability The value of the measure of dispersion.

interval_type Type of statistical interval for variable: range, iqr (interquartile), ci (confidence interval).

interval_lower Interval lower bound.

interval_upper Interval upper bound.

pgs_other_metrics A table of other metrics that are neither effect sizes nor classification metrics. Examples include: R^2 (proportion of the variance explained), or reclassification metrics. Columns:

ppm_id A PGS Performance Metrics identifier. Example: "PPM000001".

other_metrics_id Other metric identifier. This is a surrogate identifier to identify each metric.

estimate_type_long Long notation of the metric. Example: "Proportion of the variance explained".

estimate_type Short notation metric. Example: " R^2 ".

estimate The estimate's value.

unit Unit of the estimate.

variability_type Measure of statistical dispersion for variable, e.g. standard error (se) or standard deviation (sd).

variability The value of the measure of dispersion.

interval_type Type of statistical interval for variable: range, iqr (interquartile), ci (confidence interval).

interval_lower Interval lower bound.

interval_upper Interval upper bound.

pgp_to_pgs

Map PGP identifiers to PGS identifiers

Description

Map PGP identifiers to PGS identifiers.

Usage

```
pgp_to_pgs(  
  pgp_id = NULL,  
  verbose = FALSE,  
  warnings = TRUE,  
  progress_bar = TRUE  
)
```

Arguments

pgp_id	A character vector of PGS Catalog Publication identifiers, e.g., "PGP000001". If NULL then returns results for all PGP identifiers in the Catalog.
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.
progress_bar	Whether to show a progress bar as the queries are performed.

Value

A data frame of two columns: `pgp_id` and `pgs_id`.

Examples

```
## Not run:  
pgp_to_pgs('PGP000001')  
pgp_to_pgs(c('PGP000017', 'PGP000042'))  
  
## End(Not run)
```

pgp_to_ppm

Map PGP identifiers to PPM identifiers

Description

Map PGP identifiers to PPM identifiers.

Usage

```
pgp_to_ppm(  
  pgp_id = NULL,  
  verbose = FALSE,  
  warnings = TRUE,  
  progress_bar = TRUE  
)
```

Arguments

pgp_id	A character vector of PGS Catalog Publication identifiers, e.g., "PGP000001". If NULL then returns results for all PGP identifiers in the Catalog.
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.
progress_bar	Whether to show a progress bar as the queries are performed.

Value

A data frame of two columns: `pgp_id` and `ppm_id`.

Examples

```
## Not run:  
pgp_to_ppm('PGP000001')  
pgp_to_ppm(c('PGP000017', 'PGP000042'))  
  
## End(Not run)
```

pgp_to_pss	<i>Map PGP identifiers to PSS identifiers</i>
------------	---

Description

Map PGP identifiers to PSS identifiers.

Usage

```
pgp_to_pss(  
  pgp_id = NULL,  
  verbose = FALSE,  
  warnings = TRUE,  
  progress_bar = TRUE  
)
```

Arguments

pgp_id	A character vector of PGS Catalog Publication identifiers, e.g., "PGP000001". If NULL then returns results for all PGP identifiers in the Catalog.
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.
progress_bar	Whether to show a progress bar as the queries are performed.

Value

A data frame of two columns: `pgp_id` and `pss_id`.

Examples

```
## Not run:  
pgp_to_pss('PGP000001')  
pgp_to_pss(c('PGP000017', 'PGP000042'))  
  
## End(Not run)
```

pgs_to_pgp

Map PGS identifiers to PGP identifiers

Description

Map PGS identifiers to PGP identifiers.

Usage

```
pgs_to_pgp(  
  pgs_id = NULL,  
  verbose = FALSE,  
  warnings = TRUE,  
  progress_bar = TRUE  
)
```

Arguments

pgs_id	A character vector of PGS identifiers, e.g., "PGS000001". If NULL then returns results for all PGS identifiers in the Catalog.
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.
progress_bar	Whether to show a progress bar as the queries are performed.

Value

A data frame of two columns: pgs_id and pgp_id.

Examples

```
## Not run:  
pgs_to_pgp('PGS000001')  
pgs_to_pgp(c('PGS000017', 'PGS000042'))  
  
## End(Not run)
```

pgs_to_ppm *Map PGS identifiers to PPM identifiers*

Description

Map PGS identifiers to PPM identifiers.

Usage

```
pgs_to_ppm(pgs_id, verbose = FALSE, warnings = TRUE, progress_bar = TRUE)
```

Arguments

pgs_id	A character vector of PGS identifiers, e.g., "PGS000001".
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.
progress_bar	Whether to show a progress bar as the queries are performed.

Value

A data frame of two columns: pgs_id and ppm_id.

Examples

```
## Not run:
pgs_to_ppm('PGS000001')
pgs_to_ppm(c('PGS000017', 'PGS000042'))

## End(Not run)
```

pgs_to_pss *Map PGS identifiers to PSS identifiers*

Description

Map PGS identifiers to PSS identifiers.

Usage

```
pgs_to_pss(
  pgs_id = NULL,
  verbose = FALSE,
  warnings = TRUE,
  progress_bar = TRUE
)
```

Arguments

pgs_id	A character vector of PGS identifiers, e.g., "PGS000001". If NULL then returns results for all PGS identifiers in the Catalog.
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.
progress_bar	Whether to show a progress bar as the queries are performed.

Value

A data frame of two columns: pgs_id and pss_id.

Examples

```
## Not run:
pgs_to_pss('PGS000001')
pgs_to_pss(c('PGS000017', 'PGS000042'))

## End(Not run)
```

pgs_to_study

Map PGS identifiers to GWAS study identifiers

Description

Map PGS identifiers to GWAS study identifiers. Retrieves GWAS study identifiers associated with samples used in the discovery stage of queried PGS identifiers.

Usage

```
pgs_to_study(
  pgs_id = NULL,
  verbose = FALSE,
  warnings = TRUE,
  progress_bar = TRUE
)
```

Arguments

pgs_id	A character vector of PGS Catalog score accession identifiers., e.g., "PGS000001". If NULL then returns results for all PGS identifiers in the Catalog.
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.
progress_bar	Whether to show a progress bar as the queries are performed.

Value

A data frame of two columns: pgs_id and study_id.

Examples

```
## Not run:
pgs_to_study('PGS000001')
# Unmappable pgs ids will be missing, e.g., PGS000023
pgs_to_study(c('PGS000013', 'PGS000023'))

## End(Not run)
```

ppm_to_pgp

*Map PPM identifiers to PGP identifiers***Description**

Map PPM identifiers to PGP identifiers.

Usage

```
ppm_to_pgp(ppm_id, verbose = FALSE, warnings = TRUE, progress_bar = TRUE)
```

Arguments

ppm_id	A character vector of PPM identifiers, e.g., "PPM000001".
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.
progress_bar	Whether to show a progress bar as the queries are performed.

Value

A data frame of two columns: ppm_id and pgp_id.

Examples

```
## Not run:
ppm_to_pgp('PPM000001')
ppm_to_pgp(c('PPM000017', 'PPM000042'))

## End(Not run)
```

`ppm_to_pgs`*Map PPM identifiers to PGS identifiers*

Description

Map PPM identifiers to PGS identifiers.

Usage

```
ppm_to_pgs(ppm_id, verbose = FALSE, warnings = TRUE, progress_bar = TRUE)
```

Arguments

<code>ppm_id</code>	A character vector of PPM identifiers, e.g., "PPPM000001".
<code>verbose</code>	A logical indicating whether the function should be verbose about the different queries or not.
<code>warnings</code>	A logical indicating whether to print warnings, if any.
<code>progress_bar</code>	Whether to show a progress bar as the queries are performed.

Value

A data frame of two columns: `ppm_id` and `pgs_id`.

Examples

```
## Not run:  
ppm_to_pgs('PPM000001')  
ppm_to_pgs(c('PPM000017', 'PPM000042'))  
  
## End(Not run)
```

`ppm_to_pss`*Map PPM identifiers to PSS identifiers*

Description

Map PPM identifiers to PSS identifiers.

Usage

```
ppm_to_pss(ppm_id, verbose = FALSE, warnings = TRUE, progress_bar = TRUE)
```

Arguments

ppm_id	A character vector of PPM identifiers, e.g., "PPM000001".
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.
progress_bar	Whether to show a progress bar as the queries are performed.

Value

A data frame of two columns: ppm_id and pss_id.

Examples

```
## Not run:
ppm_to_pss('PPM000001')
ppm_to_pss(c('PPM000017', 'PPM000042'))

## End(Not run)
```

pss_to_pgp

Map PSS identifiers to PGP identifiers

Description

Map PSS identifiers to PGP identifiers. This is a slow function because it starts by downloading first all Performance Metrics, as this is the linkage between PSS and PGP.

Usage

```
pss_to_pgp(pss_id, verbose = FALSE, warnings = TRUE, progress_bar = TRUE)
```

Arguments

pss_id	A character vector of PSS identifiers, e.g., "PSS000001".
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.
progress_bar	Whether to show a progress bar as the queries are performed.

Value

A data frame of two columns: pss_id and pgp_id.

Examples

```
## Not run:
pss_to_pgp('PSS000001')
pss_to_pgp(c('PSS000017', 'PSS000042'))

## End(Not run)
```

pss_to_pgs

Map PSS identifiers to PGS identifiers

Description

Map PSS identifiers to PGS identifiers. This is a slow function because it starts by downloading first all Performance Metrics, as this is the linkage between PSS and PGS.

Usage

```
pss_to_pgs(pss_id, verbose = FALSE, warnings = TRUE, progress_bar = TRUE)
```

Arguments

pss_id	A character vector of PSS identifiers, e.g., "PSS000001".
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.
progress_bar	Whether to show a progress bar as the queries are performed.

Value

A data frame of two columns: pss_id and pgs_id.

Examples

```
## Not run:
pss_to_pgs('PSS000001')
pss_to_pgs(c('PSS000017', 'PSS000042'))

## End(Not run)
```

pss_to_ppm

Map PSS identifiers to PPM identifiers

Description

Map PSS identifiers to PPM identifiers. This is a slow function because it starts by downloading first all Performance Metrics.

Usage

```
pss_to_ppm(pss_id, verbose = FALSE, warnings = TRUE, progress_bar = TRUE)
```

Arguments

pss_id	A character vector of PSS identifiers, e.g., "PSS000001".
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.
progress_bar	Whether to show a progress bar as the queries are performed.

Value

A data frame of two columns: pss_id and ppm_id.

Examples

```
## Not run:
pss_to_ppm('PSS000001')
pss_to_ppm(c('PSS000017', 'PSS000042'))

## End(Not run)
```

publications-class

An S4 class to represent a set of PGS Catalog Publications

Description

The publications object consists of two tables (slots), each a table that combined form a relational database of a subset of PGS Catalog Publications. Each publication is an observation (row) in the publications table (first table).

Slots

publications A table of publications. Each publication (row) is uniquely identified by the `pgp_id` column. Columns:

pgp_id PGS Publication identifier. Example: "PGP000001".

pubmed_id PubMed identifier. Example: "25855707".

publication_date Publication date. Example: "2020-09-28". Note that the class of `publication_date` is `Date`.

publication Abbreviated name of the journal. Example: "Am J Hum Genet".

title Publication title.

author_fullname First author of the publication. Example: 'Mavaddat N'.

doi Digital Object Identifier (DOI). This variable is also curated to allow unpublished work (e.g. preprints) to be added to the catalog. Example: "10.1093/jnci/djv036".

authors Concatenated list of all the publication authors.

pgs_ids A table of publication and associated PGS identifiers. Columns:

pgp_id PGS Publication identifier. Example: "PGP000001".

pgs_id Polygenic Score (PGS) identifier.

stage PGS stage: either "gwas/dev" or "eval".

`read_scoring_file` *Read a polygenic scoring file*

Description

This function imports a PGS scoring file. For more information about the scoring file schema check `vignette("pgs-scoring-file", package = "quincunx")`.

Usage

```
read_scoring_file(source, protocol = "http", metadata_only = FALSE)
```

Arguments

<code>source</code>	PGS scoring file. This can be specified in three forms: (i) a PGS identifier, e.g. "PGS000001", (ii) a path to a local file, e.g. "~/PGS000001.txt" or "~/PGS000001.txt.gz" or (iii) a direct URL to the PGS Catalog FTP server, e.g. "http://ftp.ebi.ac.uk/pub/databases/spot/pgs/scores/PGS000001/ScoringFiles/PGS000001.txt.gz".
<code>protocol</code>	Network protocol for communication with the PGS Catalog FTP server: either "http" or "ftp".
<code>metadata_only</code>	Whether to read only the comment block (header) from the scoring file.

Value

The returned value is a named list. The names are copied from the arguments passed in source. Each element of the list contains another list of two elements: "metadata" and "data". The "metadata" element contains data parsed from the header of the PGS scoring file. The "data" element contains a data frame with as many rows as variants that constitute the PGS score. The columns can vary. There are mandatory and optional columns. The mandatory columns are those that identify the variant, effect allele (`effect_allele`), and its respective weight (`effect_weight`) in the score. The columns that identify the variant can either be the `rsID` or the combination of `chr_name` and `chr_position`. The "data" element will be NULL if argument `metadata_only` is TRUE. For more information about the scoring file schema check `vignette("pgs-scoring-file", package = "quincunx")`.

Examples

```
## Not run:
# Read a PGS scoring file by PGS ID
# (internally, it translates the PGS ID
# to the corresponding FTP URL)
try(read_scoring_file("PGS000655"))

# Equivalent to `read_scoring_file("PGS000655")`
url <- paste0(
  "http://ftp.ebi.ac.uk/",
  "pub/databases/spot/pgs/scores/",
  "PGS000655/ScoringFiles/",
  "PGS000655.txt.gz"
)
read_scoring_file(url)

# Reading from a local file
try(read_scoring_file("~/PGS000655.txt.gz"))

## End(Not run)
```

releases-class

An S4 class to represent a set of PGS Catalog Releases

Description

The releases object consists of four tables (slots) that combined form a relational database of a subset of PGS Catalog releases. Each release is an observation (row) in the releases table (first table).

Slots

`releases` A table of PGS Catalog releases. Each release (row) is uniquely identified by the release date (`date`). Columns:

date Release date.

n_pgs Number of newly released Polygenic Scores.

n_ppm Number of newly released PGS Performance Metrics.

n_pgp Number of newly released PGS Publications.

pgs_ids A table of released Polygenic Scores (PGS) identifiers. Columns:

date Release date.

pgs_id Polygenic Score (PGS) identifier. Example: "PGS000001".

ppm_ids A table of the released PGS Performance Metrics identifiers. Columns:

date Release date.

ppm_id A PGS Performance Metrics identifier. Example: "PPM000001".

pgp_ids A table of the released PGS Publication identifiers. Columns:

date Release date.

pgp_id PGS Publication identifier. Example: "PGP000001".

sample_sets-class *An S4 class to represent a set of PGS Catalog Sample Sets*

Description

The `sample_sets` object consists of four tables (slots) that combined form a relational database of a subset of PGS Catalog sample sets. Each sample set is an observation (row) in the `sample_sets` table (first table).

Slots

`sample_sets` A table of sample sets. Each sample set (row) is uniquely identified by the column `pss_id`. Columns:

pss_id A PGS Sample Set identifier. Example: "PSS000042".

`samples` A table of samples. Each sample (row) is uniquely identified by the combination of values from the columns: `pss_id` and `sample_id`. Columns:

pss_id A PGS Sample Set identifier. Example: "PSS000042".

sample_id Sample identifier. This is a surrogate key to identify each sample.

stage Sample stage: should be always Evaluation ("eval").

sample_size Number of individuals included in the sample.

sample_cases Number of cases.

sample_controls Number of controls.

sample_percent_male Percentage of male participants.

phenotype_description Detailed phenotype description.

ancestry_category Author reported ancestry is mapped to the best matching ancestry category from the NHGRI-EBI GWAS Catalog framework (see [ancestry_categories](#)) for possible values.

ancestry A more detailed description of sample ancestry that usually matches the most specific description described by the authors (e.g. French, Chinese).

country Author reported countries of recruitment (if available).

ancestry_additional_description Any additional description not captured in the other columns (e.g. founder or genetically isolated populations, or further description of admixed samples).

study_id Associated GWAS Catalog study accession identifier, e.g., "GCST002735".

pubmed_id PubMed identifier.

cohorts_additional_description Any additional description about the samples (e.g. sub-cohort information).

demographics A table of sample demographics' variables. Each demographics' variable (row) is uniquely identified by the combination of values from the columns: pss_id, sample_id, and variable. Columns:

pss_id A PGS Sample Set identifier. Example: "PSS000042".

sample_id Sample identifier. This is a surrogate identifier to identify each sample.

variable Demographics variable. Following columns report about the indicated variable.

estimate_type Type of statistical estimate for variable.

estimate The variable's statistical value.

unit Unit of the variable.

variability_type Measure of statistical dispersion for variable, e.g. standard error (se) or standard deviation (sd).

variability The value of the measure of dispersion.

interval_type Type of statistical interval for variable: range, iqr (interquartile), ci (confidence interval).

interval_lower Interval lower bound.

interval_upper Interval upper bound.

cohorts A table of cohorts. Each cohort (row) is uniquely identified by the combination of values from the columns: pss_id, sample_id and cohort_symbol. Columns:

pss_id A PGS Sample Set identifier. Example: "PSS000042".

sample_id Sample identifier. This is a surrogate key to identify each sample.

cohort_symbol Cohort symbol.

cohort_name Cohort full name.

Description

The scores object consists of six tables (slots) that combined form a relational database of a subset of PGS Catalog polygenic scores. Each score is an observation (row) in the scores table (the first table).

Slots

scores A table of polygenic scores. Each polygenic score (row) is uniquely identified by the `pgs_id` column. Columns:

pgs_id Polygenic Score (PGS) identifier. Example: "PGS000001".

pgs_name This may be the name that the authors describe the PGS with in the source publication, or a name that a curator of the PGS Catalog has assigned to identify the score during the curation process (before a PGS identifier has been given). Example: PRS77_BC.

scoring_file URL to the scoring file on the PGS FTP server. Example: "http://ftp.ebi.ac.uk/pub/databases/spo

matches_publication Indicate if the PGS data matches the published polygenic score (TRUE). If not (FALSE), the authors have provided an alternative polygenic for the Catalog and some other data, such as performance metrics, may differ from the publication.

reported_trait The author-reported trait that the PGS has been developed to predict. Example: "Breast Cancer".

trait_additional_description Any additional description not captured in the other columns. Example: "Femoral neck BMD (g/cm2)".

pgs_method_name The name or description of the method or computational algorithm used to develop the PGS.

pgs_method_params A description of the relevant inputs and parameters relevant to the PGS development method/process.

n_variants Number of variants used to calculate the PGS.

n_variants_interactions Number of higher-order variant interactions included in the PGS.

assembly The version of the genome assembly that the variants present in the PGS are associated with. Example: GRCh37.

license The PGS Catalog distributes its data according to EBI's standard Terms of Use. Some PGS have specific terms, licenses, or restrictions (e.g. non-commercial use) that we highlight in this field, if known.

publications A table of publications. Each publication (row) is uniquely identified by the `pgp_id` column. Columns:

pgs_id Polygenic Score (PGS) identifier.

pgp_id PGS Publication identifier. Example: "PGP000001".

pubmed_id PubMed identifier. Example: "25855707".

publication_date Publication date. Example: "2020-09-28". Note that the class of `publication_date` is `Date`.

publication Abbreviated name of the journal. Example: "Am J Hum Genet".

title Publication title.

author_fullname First author of the publication. Example: 'Mavaddat N'.

doi Digital Object Identifier (DOI). This variable is also curated to allow unpublished work (e.g. preprints) to be added to the catalog. Example: "10.1093/jnci/djv036".

samples A table of samples. Each sample (row) is uniquely identified by the combination of values from the columns: `pgs_id` and `sample_id`. Columns:

pgs_id Polygenic score identifier. An identifier that starts with 'PGS' and is followed by six digits, e.g. 'PGS000001'.

sample_id Sample identifier. This is a surrogate key to identify each sample.

stage Sample stage: either "discovery" or "training".

sample_size Number of individuals included in the sample.

sample_cases Number of cases.

sample_controls Number of controls.

sample_percent_male Percentage of male participants.

phenotype_description Detailed phenotype description.

ancestry_category Author reported ancestry is mapped to the best matching ancestry category from the NHGRI-EBI GWAS Catalog framework (see [ancestry_categories](#)) for possible values.

ancestry A more detailed description of sample ancestry that usually matches the most specific description described by the authors (e.g. French, Chinese).

country Author reported countries of recruitment (if available).

ancestry_additional_description Any additional description not captured in the other columns (e.g. founder or genetically isolated populations, or further description of admixed samples).

study_id Associated GWAS Catalog study accession identifier, e.g., "GCST002735".

pubmed_id PubMed identifier.

cohorts_additional_description Any additional description about the samples (e.g. sub-cohort information).

demographics A table of sample demographics' variables. Each demographics' variable (row) is uniquely identified by the combination of values from the columns: pgs_id, sample_id and variable. Columns:

pgs_id Polygenic Score (PGS) identifier.

sample_id Sample identifier. This is a surrogate identifier to identify each sample.

variable Demographics variable. Following columns report about the indicated variable.

estimate_type Type of statistical estimate for variable.

estimate The variable's statistical value.

unit Unit of the variable.

variability_type Measure of statistical dispersion for variable, e.g. standard error (se) or standard deviation (sd).

variability The value of the measure of dispersion.

interval_type Type of statistical interval for variable: range, iqr (interquartile), ci (confidence interval).

interval_lower Interval lower bound.

interval_upper Interval upper bound.

cohorts A table of cohorts. Each cohort (row) is uniquely identified by the combination of values from the columns: pgs_id, sample_id and cohort_symbol. Columns:

pgs_id Polygenic Score (PGS) identifier.

sample_id Sample identifier. This is a surrogate key to identify each sample.

cohort_symbol Cohort symbol.

cohort_name Cohort full name.

traits A table of EFO traits. Each trait (row) is uniquely identified by the combination of the columns pgs_id and efo_id. Columns:

pgs_id Polygenic Score (PGS) identifier.
efo_id An **EFO** identifier.
trait Trait name.
description Detailed description of the trait from EFO.
url External link to the EFO entry.

stages_tally A table of sample sizes and number of samples sets at each stage.
pgs_id Polygenic Score (PGS) identifier.
stage Sample stage: either "gwas", "dev" or "eval".
sample_size Sample size.
n_sample_sets Number of sample sets (only meaningful for the evaluation stage "eval")

ancestry_frequencies This table describes the ancestry composition at each stage.
pgs_id Polygenic Score (PGS) identifier.
stage Sample stage: either "gwas", "dev" or "eval".
ancestry_class_symbol Ancestry class symbol.
frequency Ancestry fraction (percentage).

multi_ancestry_composition A table of a breakdown of the ancestries included in multi-ancestries.
pgs_id Polygenic Score (PGS) identifier.
stage Sample stage: either "gwas", "dev" or "eval".
multi_ancestry_class_symbol Multi-ancestry class symbol.
ancestry_class_symbol Ancestry class symbol.

 setop

Set operations on PGS Catalog objects

Description

Performs set union, intersection, and (asymmetric!) difference on two objects of either class [scores](#), [publications](#), [traits](#), [performance_metrics](#), [sample_sets](#), [cohorts](#) or [trait_categories](#). Note that `union()` removes duplicated entities, whereas `bind()` does not.

Usage

```
union(x, y, ...)
```

```
intersect(x, y, ...)
```

```
setdiff(x, y, ...)
```

```
setequal(x, y, ...)
```

Arguments

<code>x, y</code>	Objects of either class scores , publications , traits , performance_metrics , sample_sets , cohorts or trait_categories .
<code>...</code>	other arguments passed on to methods.

Value

In the case of `union()`, `intersect()`, or `setdiff()`: an object of the same class as `x` and `y`. In the case of `setequal()`, a logical scalar.

Examples

```
# Get some `scores` objects:
my_scores_1 <- get_scores(c('PGS000012', 'PGS000013'))
my_scores_2 <- get_scores(c('PGS000013', 'PGS000014'))

#
# union()
#
# NB: with `union()`, PGS000013 is not repeated.
union(my_scores_1, my_scores_2)@scores

#
# intersect()
#
intersect(my_scores_1, my_scores_2)@scores

#
# setdiff()
#
setdiff(my_scores_1, my_scores_2)@scores

#
# setequal()
#
setequal(my_scores_1, my_scores_2)
setequal(my_scores_1, my_scores_1)
setequal(my_scores_2, my_scores_2)
```

stages

Study stages

Description

A dataset containing the various study stages assigned to samples in the PGS Catalog.

Usage

stages

Format

A data frame with 5 stages (rows) and 4 columns:

stage Study stage.

symbol One-letter symbol for the stage, or a comma separated combination thereof.

name Stage name.

definition Stage description.

Source

<https://www.pgscatalog.org/docs/ancestry>

Examples

stages

study_to_pgs	<i>Map GWAS studies identifiers to PGS identifiers</i>
--------------	--

Description

Map GWAS studies identifiers to PGS identifiers.

Usage

```
study_to_pgs(study_id, verbose = FALSE, warnings = TRUE, progress_bar = TRUE)
```

Arguments

study_id	A character vector of GWAS Catalog study accession identifiers, e.g., "GCST001937".
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.
progress_bar	Whether to show a progress bar as the queries are performed.

Value

A data frame of two columns: study_id and pgs_id.

Examples

```
## Not run:
study_to_pgs('GCST001937')
study_to_pgs(c('GCST000998', 'GCST000338'))

## End(Not run)
```

traits-class	<i>An S4 class to represent a set of PGS Catalog Traits</i>
--------------	---

Description

The traits object consists of six slots, each a table ([tibble](#)), that combined form a relational database of a subset of PGS Catalog traits. Each trait is an observation (row) in the traits table — main table. All tables have the column `efo_id` as primary key.

Slots

`traits` A table of traits. Columns:

- efo_id** An **EFO** identifier.
- parent_efo_id** An **EFO** identifier of the parent trait.
- is_child** Is this trait obtained because it is a child of other trait?
- trait** Trait name.
- description** Detailed description of the trait from EFO.
- url** External link to the EFO entry.

`pgs_ids` A table of associated polygenic score identifiers. Columns:

- efo_id** An **EFO** identifier.
- parent_efo_id** An **EFO** identifier of the parent trait.
- is_child** Is this trait obtained because it is a child of other trait?
- pgs_id** Polygenic Score (PGS) identifier.

`child_pgs_ids` A table of polygenic score identifiers associated with the child traits. Columns:

- efo_id** An **EFO** identifier.
- parent_efo_id** An **EFO** identifier of the parent trait.
- is_child** Is this trait obtained because it is a child of other trait?
- child_pgs_id** Polygenic Score (PGS) identifiers associated with child traits.

`trait_categories` A table of associated trait categories. Columns:

- efo_id** An **EFO** identifier.
- parent_efo_id** An **EFO** identifier of the parent trait.
- is_child** Is this trait obtained because it is a child of other trait?
- trait_category** Trait category name.

`trait_synonyms` A table of associated trait synonyms. Columns:

- efo_id** An **EFO** identifier.
- parent_efo_id** An **EFO** identifier of the parent trait.
- is_child** Is this trait obtained because it is a child of other trait?
- trait_synonyms** Trait synonyms.

`trait_mapped_terms` A table of associated external references, identifiers or other terms. Columns:

- efo_id** An **EFO** identifier.
- parent_efo_id** An **EFO** identifier of the parent trait.
- is_child** Is this trait obtained because it is a child of other trait?
- trait_mapped_terms** Trait mapped terms.

 trait_categories-class

An S4 class to represent a set of PGS Catalog Trait Categories

Description

The trait_categories object consists of two tables (slots) that combined form a relational database of a subset of PGS Catalog trait categories. Each score is an observation (row) in the trait_categories table (first table).

Slots

trait_categories A table of trait categories. Columns:

trait_category Trait category name.

traits A table of associated traits. Columns:

trait_category Trait category name.

efo_id An **EFO** identifier.

trait Trait name.

description Detailed description of the trait from EFO.

url External link to the EFO entry.

 write_xlsx

Export a PGS Catalog object to xlsx

Description

This function exports a PGS Catalog object to Microsoft Excel xlsx file. Each table (slot) is saved in its own sheet.

Usage

```
write_xlsx(x, file = stop("`file` must be specified"))
```

Arguments

x A [scores](#), [publications](#), [traits](#), [performance_metrics](#), [sample_sets](#), [cohorts](#), [trait_categories](#) or [releases](#) object.

file A file name to write to.

Value

No return value, called for its side effect.

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