Package ‘genieBPC’

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Type Package
Title Project GENIE BioPharma Collaborative Data Processing Pipeline
Version 2.0.1
Description The American Association Research (AACR) Project Genomics Evidence Neoplasia Information Exchange (GENIE) BioPharma Collaborative represents a multi-year, multi-institution effort to build a pan-cancer repository of linked clinico-genomic data. The genomic and clinical data are provided in multiple releases (separate releases for each cancer cohort with updates following data corrections), which are stored on the data sharing platform ‘Synapse’ <https://www.synapse.org/>.
The ‘genieBPC’ package provides a seamless way to obtain the data corresponding to each release from ‘Synapse’ and to prepare datasets for analysis.
License MIT + file LICENSE
BugReports https://github.com/GENIE-BPC/genieBPC/issues
Depends R (>= 3.4)
Imports cli (>= 2.5.0), dplyr (>= 1.0.6), dtplyr (>= 1.1.0), htr, jsonlite, purrr (>= 0.3.4), rlang (>= 1.0.0), stringr (>= 1.4.0), sunburstR, tibble (>= 3.1.2), tidyr
Suggests covr (>= 3.5.1), ggplot2 (>= 3.3.5), gt (>= 0.3.0), gtsummary (>= 1.5.2), knitr (>= 1.33), magrittr (>= 2.0.1), plotly (>= 4.10.0), rmarkdown (>= 2.8), testthat (>= 3.0.0), markdown, spelling
VignetteBuilder knitr
Config/testthat/edition 3
Encoding UTF-8
LazyData TRUE
RoxygenNote 7.3.1
URL https://genie-bpc.github.io/genieBPC/
Language en-US
check_genie_access

Check Access to GENIE Data

Description
Check Access to GENIE Data

Usage
check_genie_access(
    username = NULL,
    password = NULL,
    pat = NULL,
    check_consortium_access = FALSE
)
Arguments

username 'Synapse' username. If NULL, package will search package environment for "username".

password 'Synapse' password. If NULL, package will search package environment for "password".

pat 'Synapse' Personal Access Token. If NULL, package will search package environment for "pat".

check_consortium_access Specifies whether access to GENIE BPC consortium data releases (vs. public data releases) is checked. Default is FALSE, indicating that access to GENIE BPC public data releases is checked instead.

Value

A success message if you are able to access GENIE BPC data; otherwise an error

Author(s)

Karissa Whiting

Examples

## Not run:
# if credentials are saved:
check_genie_access()

## End(Not run)

create_analytic_cohort

Select cohort of patients for analysis

Description

This function allows the user to create a cohort from the GENIE BPC data based on cancer diagnosis information such as cancer cohort, treating institution, histology, and stage at diagnosis, as well as cancer-directed regimen information including regimen name and regimen order. This function returns each of the clinical and genomic data files subset on the patients that met criteria for the analytic cohort. Documentation regarding the structure and contents of each file can be found in the Analytic Data Guide corresponding to each data release, as well as in the Clinical Data Structure vignette.
create_analytic_cohort

data_synapse,
index_ca_seq = 1,
institution,
stage_dx,
histology,
regimen_drugs,
regimen_type = "Exact",
regimen_order,
regimen_order_type,
return_summary = FALSE
)

Arguments

data_synapse  The item from the nested list returned from pull_data_synapse() that corresponds to the cancer cohort of interest.

index_ca_seq  Index cancer sequence. Default is 1, indicating the patient’s first index cancer. The index cancer is also referred to as the BPC Project cancer in the GENIE BPC Analytic Data Guide; this is the cancer that met the eligibility criteria for the project and was selected at random for PRISSMM phenomic data curation. Specifying multiple index cancer sequences, e.g. index_ca_seq = c(1, 2) will return index cancers to patients with 1 index cancer and will return the first AND second index cancers to patients with multiple.

institution  GENIE BPC participating institution. Must be one of "DFCI", "MSK", "UHN", or "VICC" for NSCLC, BLADDER, Prostate, and PANC cohorts; must be one of "DFCI", "MSK", "VICC" for CRC and BrCa. Default selection is all institutions. This parameter corresponds to the variable ‘institution’ in the Analytic Data Guide.

stage_dx  Stage at diagnosis. Must be one of "Stage I", "Stage II", "Stage III", "Stage I-III NOS", "Stage IV". The default selection is all stages. Note that if this parameter is specified, any cases that are missing stage information are automatically excluded from the resulting cohort. This parameter corresponds to the variable ‘stage_dx’ in the Analytic Data Guide.

histology  Cancer histology. For all cancer cohorts except for BrCa (breast cancer), this parameter corresponds to the variable ‘ca_hist_adeno_squamous’ and must be one of "Adenocarcinoma", "Squamous cell", "Sarcoma", "Small cell carcinoma", "Carcinoma", "Other histologies/mixed tumor". For BrCa, this parameter corresponds to the variable ‘ca_hist_brca’ and must be one of "Invasive lobular carcinoma", "Invasive ductal carcinoma", "Other histology". The default selection is all histologies. Note that if this parameter is specified, any cases that are missing histology information are automatically excluded from the resulting cohort.

regimen_drugs  Vector with names of drugs in cancer-directed regimen, separated by a comma. For example, to specify a regimen consisting of Carboplatin and Pemetrexed, specify regimen_drugs = "Carboplatin, Pemetrexed". Acceptable values are...
found in the ‘drug_regimen_list’ dataset provided with this package. This parameter corresponds to the variable ‘regimen_drugs’ in the Analytic Data Guide.

regimen_type  Indicates whether the regimen(s) specified in ‘regimen_drugs’ indicates the exact regimen to return, or if regimens containing the drugs listed in ‘regimen_drugs’ should be returned. Must be one of "Exact" or "Containing". The default is "Exact".

regimen_order  Order of cancer-directed regimen. If multiple drugs are specified, ‘regimen_order’ indicates the regimen order for all drugs; different values of ‘regimen_order’ cannot be specified for different drug regimens. If multiple values are specified, e.g. c(1, 2), then drug regimens that met either order criteria are returned.

regimen_order_type  Specifies whether the ‘regimen_order’ parameter refers to the order of receipt of the drug regimen within the cancer diagnosis (across all other drug regimens; "within cancer") or the order of receipt of the drug regimen within the times that that drug regimen was administered (e.g. the first time carboplatin pemetrexed was received, out of all times that the patient received carboplatin pemetrexed; "within regimen"). Acceptable values are "within cancer" and "within regimen".

return_summary  Specifies whether a summary table for the cohort is returned. Default is FALSE. The ‘gtsummary’ package is required to return a summary table.

Details

See the create_analytic_cohort vignette for further documentation and examples.

Value

A list of data frames containing clinical and next generation sequencing information for patients that met the specified criteria. Optionally, if return_summary = TRUE, the list also includes summary tables for the number of records per dataset (‘tbl_overall_summary’) as well as tables of key cancer diagnosis (‘tbl_cohort’), cancer-directed regimen (‘tbl_drugs’) and next generation sequencing (‘tbl_ngs’) variables.

Author(s)

Jessica Lavery

Examples

# Examples using package test data
# Example 1  -------------------------------
# Create a cohort of all patients with stage IV NSCLC adenocarcinoma and
# obtain all of their corresponding clinical and genomic data

ex1 <- create_analytic_cohort(
  data_synapse = genieBPC::nsclc_test_data,
  stage_dx = "Stage IV",
  histology = "Adenocarcinoma"
)
# Example 2 ----------------------------------
# Create a cohort of all NSCLC patients who received Cisplatin, Pemetrexed Disodium or Cisplatin, Etoposide as their first drug regimen for their first index NSCLC

ex2 <- create_analytic_cohort(
  data_synapse = genieBPC::nsclc_test_data,
  regimen_drugs = c(
    "Cisplatin, Pemetrexed Disodium",
    "Cisplatin, Etoposide"
  ),
  regimen_order = 1,
  regimen_order_type = "within cancer"
)

# Example 3 ----------------------------------
# Create a cohort of all NSCLC patients who received Cisplatin, Pemetrexed Disodium at any time throughout the course of treatment for their cancer diagnosis, but in the event that the patient received the drug multiple times, only select the first time.

ex3 <- create_analytic_cohort(
  data_synapse = genieBPC::nsclc_test_data,
  regimen_drugs = c("Cisplatin, Pemetrexed Disodium"),
  regimen_order = 1,
  regimen_order_type = "within regimen"
)

# Example 4 ----------------------------------
# Using create_analytic_cohort with pull_data_synapse

set_synapse_credentials()

nsclc_2_0 <- pull_data_synapse("NSCLC", version = "v2.0-public")

ex4 <- create_analytic_cohort(
  data_synapse = nsclc_2_0$NSCLC_v2.0,
  regimen_drugs = c("Cisplatin, Pemetrexed Disodium"),
  regimen_order = 1,
  regimen_order_type = "within regimen"
)

---

drug_regimen_list  List of Drug Regimen Names by Cohort

Description

A dataset containing the cancer-directed drug names and their synonyms.
Usage

drug_regimen_list

Format

A table for cancer-directed drug names associated with each data release:

- **cohort** GENIE BPC Project cancer. One of "NSCLC" (non-small cell lung cancer), "CRC" (colorectal cancer), "BrCa" (breast cancer), "PANC" (pancreatic cancer), "Prostate" (prostate cancer), and "BLADDER" (bladder cancer).
- **cohort_data_release** GENIE BPC data release. Occasionally, drug names were updated across releases to include additional drug name synonyms.
- **drug_name** Name of generic/ingredient cancer-directed drug
- **drug_name_full** Name of generic/ingredient cancer-directed drug with associated synonyms ...

---

**drug_regimen_sunburst** Visualize drug regimen sequences in a sunburst plot

Description

This function allows the user to visualize the complete treatment course for selected cancer diagnoses.

Usage

drug_regimen_sunburst(data_synapse, data_cohort, max_n_regimens = NULL, ...)

Arguments

- **data_synapse** The item from the nested list returned from ‘pull_data_synapse()’
- **data_cohort** The list returned from the ‘create_analytic_cohort()’ function call
- **max_n_regimens** The maximum number of regimens displayed in the sunburst plot
- ... Additional parameters passed to ‘sunburstR::sunburst()’

Details

See the **drug_regimen_sunburst vignette** for additional details and examples.

Value

Returns data frame ‘treatment_history’ and interactive plot ‘sunburst_plot’
Examples

# Example 1 ----------------------------------
# Example using package test data
# get clinico-genomic files for a specific cohort
nsclc_sub <- create_analytic_cohort(
  data_synapse = genieBPC::nsclc_test_data,
  stage_dx = c("Stage III", "Stage IV")
)

# create sunburst plot
ex1 <- drug_regimen_sunburst(
  data_synapse = nsclc_test_data,
  data_cohort = nsclc_sub,
  max_n_regimens = 3
)

# Example 2 ----------------------------------
# using pull_data_synapse
nsclc_2_0 <- pull_data_synapse("NSCLC", version = "v2.0-public")

nsclc_stg_iv <- create_analytic_cohort(
  data_synapse = nsclc_2_0$NSCLC_v2.0,
  stage = "Stage IV"
)

ex2 <- drug_regimen_sunburst(
  data_synapse = nsclc_2_0$NSCLC_v2.0,
  data_cohort = nsclc_stg_iv,
  max_n_regimens = 3
)

---

genie_panels

*Genomic Panels Included in GENIE BPC Data*

Description

A dataset containing the name, assay identifier, and number of genes in each next-generation sequencing targeted panel included in GENIE BPC.

Usage

genie_panels

Format

A data frame with 12 rows and 3 variables:

**Sequence.Assay.ID**  Next-generation sequencing targeted panel assay identifier
nsclc_test_data

Panel  Panel name
Genes  Number of genes included ...

---

nsclc_test_data  Simulated fake GENIE BPC data for function examples and tests

Description

A named list of simulated NSCLC clinical and genomic data

Usage

nsclc_test_data

Format

A list of data frames

- pt_char  Patient characteristic data.frame
- ca_dx_index  Index cancer diagnosis data.frame
- ca_dx_non_index  Non-index cancer diagnosis data.frame
- ca_drugs  Cancer directed-regimen data.frame
- prissmm_imaging  PRISSMM Imaging report data.frame
- prissmm_pathology  PRISSMM Pathology report data.frame
- prissmm_md  PRISSMM medical oncologist report data.frame
- cpt  Cancer Panel Test (CPT)/Next Generation Sequencing (NGS) data.frame
- mutations_extended  Mutations data.frame
- fusions  Fusions data.frame
- cpt  Copy Number Alteration (CNA) data.frame

---

pull_data_synapse  Obtain clinical & genomic data files for GENIE BPC Project

Description

Function to access specified versions of clinical and genomic GENIE BPC data from Synapse and read them into the R environment. See the pull_data_synapse vignette for further documentation and examples.
pull_data_synapse

Usage

```r
pull_data_synapse(
  cohort = NULL,
  version = NULL,
  download_location = NULL,
  username = NULL,
  password = NULL,
  pat = NULL
)
```

Arguments

- **cohort**: Vector or list specifying the cohort(s) of interest. Must be one of "NSCLC" (Non-Small Cell Lung Cancer), "CRC" (Colorectal Cancer), or "BrCa" (Breast Cancer), "PANC" (Pancreatic Cancer), "Prostate" (Prostate Cancer), and "BLADDER" (Bladder Cancer). This is not case sensitive.
- **version**: Vector specifying the version of the cohort. Must match one of the release versions available for the specified 'cohort' (see `synapse_version()` for available cohort versions). When entering multiple cohorts, it is inferred that the order of the version numbers passed corresponds to the order of the cohorts passed. Therefore, 'cohort' and 'version' must be in the same order to ensure the correct data versions are pulled. See examples below for details.
- **download_location**: if 'NULL' (default), data will be returned as a list of dataframes with requested data as list items. Otherwise, specify a folder path to have data automatically downloaded there. When a path is specified, data are not read into the R environment.
- **username**: 'Synapse' username
- **password**: 'Synapse' password
- **pat**: 'Synapse' personal access token

Value

Returns a nested list of clinical and genomic data corresponding to the specified cohort(s).

Authentication

To access data, users must have a valid 'Synapse' account with permission to access the data set and they must have accepted any necessary 'Terms of Use'. Users must always authenticate themselves in their current R session. (see README: Data Access and Authentication for details). To set your 'Synapse' credentials during each session, call:

`set_synapse_credentials(username = "your_username", password = "your_password")`

In addition to passing your 'Synapse' username and password, you may choose to set your 'Synapse' Personal Access Token (PAT) by calling: `set_synapse_credentials(pat = "your_pat")`.

If your credentials are stored as environmental variables, you do not need to call `set_synapse_credentials()` explicitly each session. To store authentication information in your environmental variables, add the
following to your .Renviron file, then restart your R session' (tip: you can use `usethis::edit_r_environ()` to easily open/edit this file):

- `SYNAPSE_USERNAME = <your-username>`
- `SYNAPSE_PASSWORD = <your-password>`
- `SYNAPSE_PAT = <your-pat>`

Alternatively, you can pass your username and password or your PAT to each individual data pull function if preferred, although it is recommended that you manage your passwords outside of your scripts for security purposes.

**Analytic Data Guides**

Documentation corresponding to the clinical data files can be found on `Synapse` in the Analytic Data Guides:

- NSCLC v1.1-Consortium Analytic Data Guide
- NSCLC v2.2-Consortium Analytic Data Guide
- NSCLC v2.0-Public Analytic Data Guide
- NSCLC v3.1-Consortium Analytic Data Guide
- CRC v1.3-Consortium Analytic Data Guide
- CRC v2.0-Public Analytic Data Guide
- BrCa v1.1-Consortium Analytic Data Guide
- BrCa v1.2-Consortium Analytic Data Guide
- BLADDER v1.1-Consortium Analytic Data Guide
- BLADDER v1.2-Consortium Analytic Data Guide
- PANC v1.1-Consortium Analytic Data Guide
- PANC v1.2-Consortium Analytic Data Guide
- Prostate v1.1-Consortium Analytic Data Guide
- Prostate v1.2-Consortium Analytic Data Guide

**Author(s)**

Karissa Whiting, Michael Curry

**Examples**

```r
# Example 1 ----------------------------------
# Set up 'Synapse' credentials
set_synapse_credentials()

# Print available versions of the data
synapse_version(most_recent = TRUE)

# Pull version 2.0-public for non-small cell lung cancer
# and version 2.0-public for colorectal cancer data
```
```r
ex1 <- pull_data_synapse(
  cohort = c("NSCLC", "CRC"),
  version = c("v2.0-public", "v2.0-public")
)

names(ex1)
```

---

**regimen_abbreviations**  *List of Drug Regimen Abbreviations*

**Description**
A dataset containing the cancer-directed drug regimens and their common abbreviations

**Usage**
`regimen_abbreviations`

**Format**
A table for cancer-directed drug regimens and their common abbreviations

- **regimen_drugs**  List of all drugs in the regimen
- **abbreviation**  Common name of drug regimen, e.g. FOLFOX ...

---

**select_unique_ngs**  *Selecting corresponding unique next generation sequencing reports*

**Description**
For patients with multiple associated next generation (NGS) sequencing reports, select one unique NGS report per patient for the purpose of creating an analytic dataset based on user-defined criterion, including OncoTree code, primary vs. metastatic tumor sample, and earliest vs. most recent sample. If multiple reports for a patient remain available after the user-defined specifications, or if no specifications are provided, the panel with the largest number of genes is selected by default. Sample optimization is performed in the order that the arguments are specified in the function, regardless of the arguments’ order provided by the user. Namely, the OncoTree code is prioritized first, sample type is prioritized second and finally the time is prioritized last. For patients with exactly one genomic sample, that unique genomic sample will be returned regardless of whether it meets the user-specified parameters. Running the `select_unique_ngs()` function will ensure that the resulting dataset returned by merging the next generation sequencing report data onto the `cohort_ca_dx` dataset returned by `create_analytic_cohort()` will maintain the structure of `cohort_ca_dx` (either one record per patient or one record per diagnosis). Currently, if multiple diagnoses per patient are returned from `create_analytic_cohort()`, using `select_unique_ngs()` will select a single NGS report per patient. In future iterations, this will be updated so that one NGS report per diagnosis can be selected.
select_unique_ngs

Usage

select_unique_ngs(
  data_cohort,
  oncotree_code = NULL,
  sample_type = NULL,
  min_max_time = NULL
)

Arguments

data_cohort CPT (NGS) dataframe returned from the create_analytic_cohort function
oncotree_code character vector specifying which sample OncoTree codes to keep. See "cpt_oncotree_code" column of data_cohort argument above to get options.
sample_type character specifying which type of genomic sample to prioritize, options are "Primary", "Local" and "Metastasis". Default is to not select a NGS sample based on the sample type.
min_max_time character specifying if the first or last genomic sample recorded should be kept. Options are "min" (first) and "max" (last).

Details

Note that the NGS dataset serves as the link between the clinical and genomic data, where the NGS dataset includes one record per NGS report per patient, including the NGS sample ID that is used to link to the genomic data files. Merging data from the NGS report onto the analytic cohort returned from create_analytic_cohort() therefore allows users to utilize all clinical and genomic data available.

See the select_unique_ngs vignette for further documentation and examples.

Value

returns the 'cohort_ngs' object of the create_analytic_cohort with unique genomic samples taken from each patients.

Author(s)

Karissa Whiting

Examples

# Example 1 ----------------------------------
# Create a cohort of all patients with stage IV NSCLC of
# histology adenocarcinoma
set_synapse_credentials()

nsclc_2.0 <- pull_data_synapse("NSCLC", version = "v2.0-public")

ex1 <- create_analytic_cohort(
  data_synapse = nsclc_2.0$NSCLC_v2.0,
  stage_dx = c("Stage IV"),
  sample_type = "Local",
histology = "Adenocarcinoma"
)

# select unique next generation sequencing reports for those patients
samples_data1 <- select_unique_ngs(
  data_cohort = ex1$cohort_ngs,
  sample_type = "Primary"
)

# Example 2 ----------------------------------
# Create a cohort of all NSCLC patients who
# received Cisplatin, Pemetrexed Disodium or Cisplatin,
# Etoposide as their first drug regimen
ex2 <- create_analytic_cohort(
  data_synapse = nsclc_2_0$NSCLC_v2.0,
  regimen_drugs = c(
    "Cisplatin, Pemetrexed Disodium",
    "Cisplatin, Etoposide"
  ),
  regimen_order = 1,
  regimen_order_type = "within regimen"
)

samples_data2 <- select_unique_ngs(
  data_cohort = ex2$cohort_ngs,
  oncotree_code = "NSCLCPD",
  sample_type = "Metastasis",
  min_max_time = "max"
)

---

**set_synapse_credentials**

*Connect to 'Synapse' API*

**Description**

This function sets 'Synapse' credentials for the user's current session.

**Usage**

```r
set_synapse_credentials(username = NULL, password = NULL, pat = NULL)
```

**Arguments**

- `username` 'Synapse' username. If NULL, package will search environmental variables for 'SYNAPSE_USERNAME'.
- `password` 'Synapse' password. If NULL, package will search environmental variables for 'SYNAPSE_PASSWORD'.

---
set_synapse_credentials

pat 'Synapse' Personal Access Token. If NULL, package will search environmental variables for 'SYNAPSE_PAT'.

Details

To access data, users must have a valid 'Synapse' account with permission to access the data set and they must have accepted any necessary 'Terms of Use'. Users must authenticate themselves in their current R session. (See README 'Data Access and Authentication' at https://genie-bpc.github.io/genieBPC/ for details). To set your 'Synapse' credentials during each session, call: 'set_synapse_credentials(username = "your_username", password = "your_password")'.

In addition to passing your 'Synapse' username and password, you may choose to set your 'Synapse' Personal Access Token (PAT) by calling: 'set_synapse_credentials(pat = "your_pat")'.

If your credentials are stored as environmental variables, you do not need to call 'set_synapse_credentials()' explicitly each session. To store authentication information in your environmental variables, add the following to your .Renviron file, then restart your R session (tip: you can use 'usethis::edit_r_environ()' to easily open/edit this file):

- 'SYNAPSE_USERNAME = <your-username>'
- 'SYNAPSE_PASSWORD = <your-password>'
- 'SYNAPSE_PAT = <your-pat>'

Alternatively, you can pass your username and password or your PAT to each individual data pull function if preferred, although it is recommended that you manage your passwords outside of your scripts for security purposes.

Value

A success message if you credentials are valid for 'Synapse' platform; otherwise an error

Author(s)

Karissa Whiting

Examples

```r
## Not run:
set_synapse_credentials(
    username = "your-username",
    password = "your-password"
)
set_synapse_credentials(
    pat = "your-personal-access-token"
)
```

## End(Not run)
**synapse_tables**

<table>
<thead>
<tr>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>A dataset containing the 'Synapse' table IDs for each dataset in GENIE BPC.</td>
</tr>
</tbody>
</table>

**Usage**

`synapse_tables`

**Format**

A lookup table for 'Synapse' data table IDs:

- **cohort** GENIE BPC Project Cohort
- **version** Release version
- **release_date** Month and year of data release
- **df** Clinical, Genomic, or Metadata Dataset
- **synapse_id** 'Synapse' table ID for each dataset ...

**Source**


---

**synapse_version**

<table>
<thead>
<tr>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Return list of available GENIE BPC data releases</td>
</tr>
</tbody>
</table>

**Usage**

`synapse_version(cohort = NULL, most_recent = FALSE)`

**Description**

GENIE BPC data are updated periodically to add variables and reflect additional data cleaning. Each time the data are updated the data release version number is incremented. The `synapse_version()` function will get available version numbers for each cohort to help the user determine what is the most recent version for each cohort.
**Arguments**

- **cohort**: Vector specifying the cohort(s) of interest. Cohorts must be one of "NSCLC" (Non-Small Cell Lung Cancer), "CRC" (Colorectal Cancer), or "BrCa" (Breast Cancer), "PANC" (Pancreatic Cancer), "Prostate" (Prostate Cancer), and "BLADDER" (Bladder Cancer).

- **most_recent**: Indicates whether the function will return only the most recent version number for each cohort ('most_recent' = TRUE) or all available version numbers for each cohort ('most_recent' = FALSE).

**Details**

Specifies the version numbers available for each cancer cohort. Version numbers are specified as part of the call to `pull_data_synapse()`.

**Value**

Returns a table containing the available versions for each cohort. Consortium releases are restricted to GENIE BPC consortium members.

**Examples**

```
synapse_version()
synapse_version(most_recent = TRUE)
```
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