

Package ‘genieBPC’

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Type Package

Title Project GENIE BioPharma Collaborative Data Processing Pipeline

Version 2.2.0

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.

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BugReports <https://github.com/GENIE-BPC/genieBPC/issues>

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check_genie_access	<i>Check Access to GENIE Data</i>
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Description

Check Access to GENIE Data

Usage

```
check_genie_access(pat = NULL, check_consortium_access = FALSE)
```

Arguments

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)
```

cohort_institution *List of Institutions by Cohort*

Description

A dataset containing the institutions participating in curation of each cancer cohort

Usage

```
cohort_institution
```

Format

A lookup table for institutions participating in curation by cancer cohort

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), "BLADDER" (bladder cancer), and "RENAL" (renal cancer).

institution GENIE BPC participating institution. ...

`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](#).

Usage

```
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

<code>data_synapse</code>	The item from the nested list returned from <code>pull_data_synapse()</code> that corresponds to the cancer cohort of interest.
<code>index_ca_seq</code>	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 PRISMM phenomic data curation. Specifying multiple index cancer sequences, e.g. <code>index_ca_seq = c(1, 2)</code> will return index cancers to patients with 1 index cancer and will return the first AND second index cancers to patients with multiple.
<code>institution</code>	GENIE BPC participating institution. See lookup table 'cohort_institution' for a list of institutions participating in curation of each cancer cohort. 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::nslc_test_data,
  stage_dx = "Stage IV",
  histology = "Adenocarcinoma"
)

names(ex1)

# 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::nslc_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::nslc_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), "BLADDER" (bladder cancer), and "RENAL" (renal 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 in parentheses ...

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	<i>Genomic Panels Included in GENIE BPC Data</i>
--------------	--

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

Panel Panel name

Genes Number of genes included ...

nslc_test_data	<i>Simulated fake GENIE BPC data for function examples and tests</i>
----------------	--

Description

A named list of simulated NSCLC clinical and genomic data

Usage

```
nslc_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.

Usage

```
pull_data_synapse(
  cohort = NULL,
  version = NULL,
  download_location = 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), "BLADDER" (Bladder Cancer), and "RENAL" (Renal 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.
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' Personal Access Token (PAT) during each session, call: `'set_synapse_credentials(pat = "your_pat")'`.

If your credentials are stored as an environmental variable, 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_PAT = <your-pat>'`

Alternatively, you can pass your PAT to each individual data pull function if preferred, although it is recommended that you manage your PATs 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](#)
- [CRC v3.1-Consortium Analytic Data Guide](#)
- [BrCa v1.1-Consortium Analytic Data Guide](#)
- [BrCa v1.2-Consortium Analytic Data Guide](#)
- [BrCa v1.0-Public 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](#)
- [PANC v1.0-Public Analytic Data Guide](#)
- [Prostate v1.1-Consortium Analytic Data Guide](#)
- [Prostate v1.2-Consortium Analytic Data Guide](#)
- [Prostate v1.0-Public Analytic Data Guide](#)
- [RENAL v1.1-Consortium Analytic Data Guide](#)

Author(s)

Karissa Whiting, Michael Curry

Examples

```
# 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

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.

Usage

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

Arguments

<code>data_cohort</code>	CPT (NGS) dataframe returned from the <code>create_analytic_cohort</code> function
<code>oncotree_code</code>	character vector specifying which sample OncoTree codes to keep. See "cpt_oncotree_code" column of <code>data_cohort</code> argument above to get options.
<code>sample_type</code>	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.
<code>min_max_time</code>	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()

nslc_2_0 <- pull_data_synapse("NSCLC", version = "v2.0-public")

ex1 <- create_analytic_cohort(
  data_synapse = nslc_2_0$NSCLC_v2.0,
  stage_dx = c("Stage IV"),
  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 = nslc_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

```
set_synapse_credentials(pat = NULL)
```

Arguments

`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' Personal Access Token (PAT) during each session, call: `'set_synapse_credentials(pat = "your_pat")'`.

If your credentials are stored as an environmental variable, 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_PAT = <your-pat>'`

Alternatively, you can pass your PAT to each individual data pull function if preferred, although it is recommended that you manage your PATs 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

```
## Not run:  
set_synapse_credentials(  
  pat = "your-personal-access-token"  
)  
  
## End(Not run)
```

synapse_tables	<i>'Synapse' table IDs</i>
----------------	----------------------------

Description

A dataset containing the 'Synapse' table IDs for each dataset in GENIE BPC.

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

<https://www.synapse.org/#!/Synapse:syn21226493/wiki/599164>

synapse_version	<i>Return list of available GENIE BPC data releases</i>
-----------------	---

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.

Usage

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

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), "BLADDER" (Bladder Cancer), and "RENAL" (Renal Cancer).
most_recent	Indicates whether the function will return only the most recent version number for each cohort (<code>most_recent = TRUE</code>) or all available version numbers for each cohort (<code>most_recent = FALSE</code>)

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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