Package: rigvf (via r-universe)

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Title R interface to the IGVF Catalog

Version 0.99.6

Description The IGVF Catalog provides data on the impact of genomic variants on function. The `rigvf` package provides an interface to the IGVF Catalog, allowing easy integration with Bioconductor resources.

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Depends R (>= 4.1.0)

Imports methods, httr2, rjsoncons, dplyr, tidyr, rlang, memoise, cachem, whisker, jsonlite, GenomicRanges, IRanges, GenomeInfoDb

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BugReports https://github.com/IGVF/rigvf/issues

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VignetteBuilder knitr

Config/testthat/edition 3

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Contents

`arango` *Internal interface to ArangoDB REST API*

Description

Objects documented on this page are for developer use. `arango_request()` formulates 'GET' or 'POST' to the ArangoDB API.

`arango_auth()` uses username and password to authenticate against the database.

`arango_collections()` implements the `_api/collection` endpoint for available collections in the IGVF database.

`arango_cursor()` implements the `_api/cursor` endpoint to allow a user-specified query of the IGVF database.

Usage

```
arango_request(path, ..., body = NULL, jwt_token = NULL)

arango_auth(
  username = rigvf_config$get("username"),
  password = rigvf_config$get("password")
)

arango_collections(
  username = rigvf_config$get("username"),
  password = rigvf_config$get("password")
)

arango_cursor(
  query,
  ...,
  username = rigvf_config$get("username"),
  password = rigvf_config$get("password")
)
```

Arguments

`path`	character(1) path to the API end point. Note that database-specfic paths are prefixed with `/_db/igvf`.

`...`	for `arango_request()`, named arguments to be used a query parameters. for `arango_cursor()`, named arguments to be interpreted as bind variables in the query.
`body`	if not NULL, formulate a POST request with JSON body.
`jwt_token`	character(1) JWT token obtained via `arango_auth()`.
`username`	character(1) ArangoDB user name. Default: "guest".
`password`	character(1) ArangoDB password. Default: "guestigvfcatalog". A better practice is to use an environment variable to record the password, rather than encoding in a script, so `password = Sys.getenv("RIGVF_ARANGODB_PASSWORD")`.
`query`	character(1) the FILE NAME (without extension `.aql` of the query template.

Details

`arango_auth()` is 'memoized', so invoked only once per hour for a particular user and password. The memoised result can be cleared with `memoise::forget(arango_auth)`.

`arango_cursor()` expects queries to be written in package system files in the `inst/aql` directory. This allows rapid iteration during query development (the package does not need to re-loaded when the query is updated) and some opportunity for language-specific highlighting if supported by the developer's text editor.

Value

`arango_request()` returns the JSON response as a character(1) vector.

`arango_auth()` returns a JWT token to be used for authentication in subsequent calls.

`arango_collections()` returns a tibble with columns `name`, `type` (either 'node' or 'edge'), and `count`.

`arango_cursor()` returns the JSON character(1) 'result' of the query.

Examples

```
## available queries
templates <- system.file(package = "rigvf", "aql")
dir(templates)
```

`catalog_queries` *Query the IGVF Catalog via REST API*

Description

This page documents functions using the IGVF REST API, documented at https://api.catalog.igvf.org/#.

Note that functions will only return a limited number of responses, see `limit` and `page` arguments below for control over number of responses.

`gene_variants()` locates variants associated with a gene. Only one of `gene_id`, `hgnc`, `gene_name`, or `alias` should be specified.

`variant_genes()` locates genes associated with a variant. Only one of `spdi`, `hgvs`, `rsid`, `variant_id`, or `chr` + `position` should be specified.

`gene_elements()` locates elements associated with a gene.

`elements()` locates genomic elements based on a genomic range query.

`element_genes()` locates genomic elements and associated genes based on a genomic range query.

Usage

```
gene_variants(
  gene_id = NULL,
  hgnc = NULL,
  gene_name = NULL,
  alias = NULL,
  organism = "Homo sapiens",
  log10pvalue = NULL,
  effect_size = NULL,
  page = 0L,
  limit = 25L,
  verbose = FALSE
)

variant_genes(
  spdi = NULL,
  hgvs = NULL,
  rsid = NULL,
  variant_id = NULL,
  chr = NULL,
  position = NULL,
  organism = "Homo sapiens",
  log10pvalue = NULL,
  effect_size = NULL,
  page = 0L,
  limit = 25L,
  verbose = FALSE
)

gene_elements(gene_id = NULL, page = 0L, limit = 25L, verbose = FALSE)

elements(range = NULL, page = 0L, limit = 25L)

element_genes(range = NULL, page = 0L, limit = 25L, verbose = FALSE)
```

Arguments

`gene_id`	character(1) Ensembl gene identifier, e.g., "ENSG00000106633"

hgnc	character(1) HGNC identifier
gene_name	character(1) Gene symbol, e.g., "GCK"
alias	character(1) Gene alias
organism	character(1) Either 'Homo sapiens' (default) or 'Mus musculus'
log10pvalue	character(1) The following can be used to set thresholds on the negative log10pvalue: gt (>), gte (>=), lt (<), lte (<=), with a ":" following and a value, e.g., "gt:5.0"
effect_size	character(1) Optional string used for thresholding on the effect size of the variant on the gene. See 'log10pvalue'. E.g., "gt:0.5"
page	integer(1) when there are more response items than limit, offers pagination. starts on page 0L, next is 1L, ...
limit	integer(1) the limit parameter controls the page size and can not exceed 1000
verbose	logical(1) return additional information about variants and genes
spdi	character(1) SPDI of variant
hgvs	character(1) HGVS of variant
rsid	character(1) RSID of variant
variant_id	character(1) IGVF variant ID
chr	character(1) UCSC-style chromosome name of variant, e.g. "chr1"
position	character(1) 0-based position of variant
range	the query GRanges (expects 1-based start position)

Value

`gene_variants()` returns a tibble describing variants associated with the gene; use `verbose = TRUE` to retrieve more extensive information.

`variant_genes()` returns a tibble describing genes associated with a variant; use `verbose = TRUE` to retrieve more extensive information.

`gene_elements()` returns a tibble describing elements associated with the gene; use `verbose = TRUE` to retrieve more extensive information.

`elements()` returns a GRanges object describing elements.

`element_genes()` returns a tibble describing genomic element and gene pairs.

Examples

```
rigvf::gene_variants(gene_name = "GCK")

rigvf::gene_variants(gene_name = "GCK", effect_size="gt:0.5")

rigvf::gene_variants(gene_name = "GCK", verbose = TRUE)

rigvf::variant_genes(spdi = "NC_000001.11:920568:G:A")

res <- rigvf::gene_elements(gene_id = "ENSG00000187961")
res
res |>
```

```
    dplyr::select(regions) |>
    tidyr::unnest_wider(regions)

rng <- GenomicRanges::GRanges("chr1", IRanges::IRanges(1157520,1158189))

rigvf::elements(range = rng)

rigvf::element_genes(range = rng)
```

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db_queries *Query the IGVF Catalog via ArangoDB*

Description

edges() and nodes() identify edges or nodes in the data base.

db_gene_variants() locates variants associated with a (Ensembl) gene identifier.

db_gene_elements() locates genomic elements associated with a (Ensembl) gene identifier.

Usage

```
db_edges(
  username = rigvf_config$get("username"),
  password = rigvf_config$get("password")
)

db_nodes(
  username = rigvf_config$get("username"),
  password = rigvf_config$get("password")
)

db_gene_variants(
  gene_id,
  threshold,
  username = rigvf_config$get("username"),
  password = rigvf_config$get("password")
)

db_gene_elements(
  gene_id,
  threshold,
  username = rigvf_config$get("username"),
  password = rigvf_config$get("password")
)
```

Arguments

username	character(1) ArangoDB user name. Default: "guest".
password	character(1) ArangoDB password. Default: "guestigvfcatalog". A better practice is to use an environment variable to record the password, rather than encoding in a script, so password = Sys.getenv("RIGVF_ARANGODB_PASSWORD").
gene_id	character(1) Ensembl gene identifier
threshold	numeric(1) minimum association statistic, minus log10 p-value for variants, and score for elements

Value

edges() and nodes() return a tibble with the edge or node name and count of occurrences in the database.

db_gene_variants() returns a tibble summarizing variants associated with the gene.

db_gene_elements() returns a tibble summarizing genomic elements associated with the gene.

Examples

```
db_edges()

db_nodes()

db_gene_variants("ENSG00000106633", threshold = 4.0)

db_gene_elements("ENSG00000106633", threshold = 0.5)
```

---

rigvf *Package configuration global variable management*

Description

Objects documented on this page are for developer use.

rigvf_config provides a simple interface to manage 'package global' variables via rigvf_config$get(), rigvf_config$set(), etc. The default username and password provide guest access.

Usage

```
rigvf_config
```

Format

rigvf_config is a list of functions for listing (ls())and manipulating (get(), set(), unset()) package-global variables.

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