## Package 'AtlasRDF'

July 12, 2017

Title Gene Expression Atlas query and gene set enrichment package.

Type Package

**Version** 1.12.0 **Date** 2015-01-26

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<b>Depends</b> R (>= 2.10), hash, SPARQL, methods
<b>Description</b> Query the Gene Expression Atlas RDF data at the European Bioinformatics Institute using genes, experimental factors (such as disease, cell type, compound treatments), pathways and proteins. Also contains a function to perform an enrichment of your gene list across Experimental Factor Ontology (EFO) using the Atlas background set.
Note use the R SPARQL package available at http://cran.r-project.org/web/packages/SPARQL/
License Apache License 2.0
<b>biocViews</b> Microarray, DataImport, GeneSetEnrichment, GeneExpression, DifferentialExpression, DataRepresentation
PackageStatus Deprecated
NeedsCompilation no
R topics documented:
AtlasRDF-package
doFishersEnrichment
doFishersEnrichmentForEnsemblIds       5         doFishersEnrichmentForGeneNames       6
drawHeatMapForAtlasExperiment
enrichmentresult-class
excludeSubclasses
factorbackground-class
generef-class
getAllEnsemblGenesForExFactor
getExFactorURIFromLabel
getExperimentIdsForGeneURI
getExperimentsByDescription
getExperimentURIsForGeneId
getGeneListFromPubmedid

2 AtlasRDF-package

Index		31
	vizPvalues	30
	transcription_pathway_enrichment	
	searchForEFOTerms	
	pathwayresult-class	
	orderEnrichmentResults	
	includeOnlySubclasses	
	getTaxonURI	
	getSpeciesSpecificEnsemblGenesForExFactor	
	getRankedPathwaysForGeneIds	
	getPathwayUriFromName	
	getPathwaysFromGenesAndCondition	
	getPathwayForGeneId	
	getOntologyMappings	
	getLabel	
	getGeneUriFromName	20
	getGeneUriFromEnsemblId	
	getGenesForPathwayURI	
	getGenesForExperimentURI	18
	getGenesForExperimentID	17

**Description** 

AtlasRDF-package

Query the Gene Expression Atlas RDF data at the European Bioinformatics Institute using genes, experimental factors (such as disease, cell type, compound treatments), pathways and proteins. Also contains a function to perform an enrichment of your gene list across Experimental Factor Ontology (EFO) using the Atlas background set. Access the backgroun data sets for enrichment from https://github.com/jamesmalone/AtlasRDF-R

Gene Expression Atlas query and gene set enrichment package.

## **Details**

Package: AtlasRDF Type: Package Version: 1.3

Date: 2015-01-26 License: Apache 2.0

#### Note

Linux users may need to perform the following:

sudo apt-get install libxml2-dev from R environment: install.package("XML") sudo apt-get install libcurl4-gnutls-dev

AtlasRDF-package 3

from R environment: install.package("RCurl") from R environment: install.package("hash") from R environment: install.package("SPARQL")

## Author(s)

James Malone, Simon Jupp, Maryam Soleimani

Maintainer: James Malone <malone@ebi.ac.uk>

#### References

Gene Expression Atlas RDF - http://wwwdev.ebi.ac.uk/rdf/services/atlas/ Gene Expression Atlas - http://www.ebi.ac.uk/gxa Experimental Factor Ontology - http://www.ebi.ac.uk/efo Kapushesky M, Adamusiak T, Burdett T, Culhane A, Farne A, Filippov A, Holloway E, Klebanov A, Kryvych N, Kurbatova N, Kurnosov P, Malone J, Melnichuk O, Petryszak R, Pultsin N, Rustici G, Tikhonov A, Travillian RS, Williams E, Zorin A, Parkinson H, Brazma A. (2012) Gene Expression Atlas update—a value-added database of microarray and sequencing-based functional genomics experiments. Nucleic Acids Res. 2012 Jan;40(Database issue):D1077-81.

```
###find experiments where the description contains a given search term
#experimentlist <- getExperimentsByDescription("breast carcinoma")</pre>
###get gene list for given experiment ID
#genelist <- getGenesForExperimentID("E-MEXP-3739")</pre>
###get list of pathways associated with a gene for which the genes are differentially expressed for a giver
pathwayinfo <- getPathwaysFromGenesAndCondition("efo:EFO_0001359")</pre>
pathwayinfo$pathwayname
###perform gene set enrichment using Atlas as background to find factors genes are enriched for
#load the background files for the species of interest (requires downloading)
#find background sets at https://github.com/jamesmalone/AtlasRDF-R
#load("human_gene_list.RData") #human_genelist_bg
#load("human_factor_counts.RData")
                                        #human_factor_counts
###creat or load your genelist of interest
genelist <- c("GSTT1", "PLCE1", "XIST", "ENOSF-1", "AHR", "FSTL1", "YIPF5", "RBM5", "TNP01", "CREB1", "NO)
###get id for species 'human'
taxon <-getTaxonURI("human") #obo:NCBITaxon_9606</pre>
###do enrichment
#results <- doFishersEnrichmentForGeneNames(genelist, taxon, human_genelist_bg, human_factor_counts)</pre>
###visualize results, filtering by p-value
#vizPvalues(results, "0.005")
```

4 doFishersEnrichment

doFishersEnrichment Perform a gene set enrichment using gene list based on gene URIs.

## **Description**

Perform an enrichment of an input gene list using the Atlas as a background set for a specified species. The genes are given as a set of gene URIs (e.g. <a href="http://identifiers.org/ensembl/ENSG00000229807">http://identifiers.org/ensembl/ENSG00000229807</a>). The results are a list of experimental factors for which these genes are enriched. Access the background data sets for enrichment from <a href="https://github.com/jamesmalone/AtlasRDF-R">https://github.com/jamesmalone/AtlasRDF-R</a>

## Usage

doFishersEnrichment(genelist, genelist\_bg, genecounts)

#### **Arguments**

genelist Variable genelist a vector of genes using gene URIs

genelist\_bg genelist\_bg background list appropriate for the species of interest should be

obtained from svn

genecounts genecounts counts list appropriate for the species of interest should be obtained

from svn

#### Value

Returns a list of enrichmentresult objects which are described as follows:

factoruri the URI of the experimental factor

label the human readable label for the experimental factor

p. value the p. value for the Fisher's exact test for the gene list with this factor. This can

be seen as avidence for this factor being highly enriched (low p value) or not

(high p value) for the input gene list

estimate an estimate of the odds ratio. Note that the conditional Maximum Likelihood

Estimate (MLE) rather than the unconditional MLE (the sample odds ratio) is

used. Only present in the 2 by 2 case.

alternative a character string describing the alternative hypothesis.

null.value the odds ratio under the null, or. Only present in the 2 by 2 case.

method the character string "Fisher's Exact Test for Count Data".

enrichedgenes list of the genes from the input gene list that were enriched for this factor as a

list of Gene URIs. To get the gene names use the function getClassLabel

#### Author(s)

James Malone, European Bioinformatics Institute Simon Jupp, European Bioinformatics Institute Maryam Soleimani, European Bioinformatics Institute

## References

Agresti, A. (1990) Categorical data analysis. New York: Wiley. Pages 59-66.

#### **Examples**

doFishersEnrichmentForEnsemblIds

Perform a gene set enrichment using gene list based on common EN-SEMBL gene IDs.

#### **Description**

Perform an enrichment of an input gene list using the Atlas as a background set for a specified species. The genes are given as a set of ENSEMBL gene IDs (e.g. "ENSG00000188554"). The results are a list of experimental factors for which these genes are enriched. Access the backgroun data sets for enrichment from https://github.com/jamesmalone/AtlasRDF-R

#### Usage

doFishersEnrichmentForEnsemblIds(geneids, genelist\_bg, genecounts)

#### **Arguments**

geneids Variable geneids a vector of genes using ENSEMBL gene IDs

genelist\_bg genelist\_bg background list appropriate for the species of interest should be

obtained from svn

genecounts genecounts counts list appropriate for the species of interest should be obtained

from svn

#### Value

Returns a list of enrichmentresult objects which are described as follows:

factoruri the URI of the experimental factor

label the human readable label for the experimental factor

p.value the p.value for the Fisher's exact test for the gene list with this factor. This can

be seen as avidence for this factor being highly enriched (low p value) or not

(high p value) for the input gene list

estimate an estimate of the odds ratio. Note that the conditional Maximum Likelihood

Estimate (MLE) rather than the unconditional MLE (the sample odds ratio) is

used. Only present in the 2 by 2 case.

alternative a character string describing the alternative hypothesis.

null.value the odds ratio under the null, or. Only present in the 2 by 2 case.

method the character string "Fisher's Exact Test for Count Data".

enrichedgenes list of the genes from the input gene list that were enriched for this factor as a

list of Gene URIs. To get the gene names use the function getClassLabel

#### Author(s)

James Malone, European Bioinformatics Institute Simon Jupp, European Bioinformatics Institute Maryam Soleimani, European Bioinformatics Institute

#### References

Agresti, A. (1990) Categorical data analysis. New York: Wiley. Pages 59-66.

#### **Examples**

 ${\tt doFishersEnrichmentForGeneNames}$ 

Perform a gene set enrichment using gene list based on common gene names.

## Description

Perform an enrichment of an input gene list using the Atlas as a background set for a specified species. The genes are given as a set of common gene names (e.g. Brca1). The results are a list of experimental factors for which these genes are enriched. Access the backgroun data sets for enrichment from https://github.com/jamesmalone/AtlasRDF-R

#### Usage

doFishersEnrichmentForGeneNames(genenames, taxon, genelist\_bg, genecounts, endpoint="http://w

#### **Arguments**

yariable genenames a vector of genes using common gene names

taxon The taxon is an ontology URI which can be found using the function getTaxonURI()

genelist\_bg genelist\_bg background list appropriate for the species of interest should be obtained from svn

genecounts genecounts counts list appropriate for the species of interest should be obtained from svn

endpoint SPARQL endpoint to query data from. Default value is the live Atlas SPARQL

endpoint but this can be overidden. Required to find Gene URIs from gene

names

#### Value

Returns a list of enrichmentresult objects which are described as follows:

factoruri the URI of the experimental factor label the human readable label for the experimental factor p.value the p.value for the Fisher's exact test for the gene list with this factor. This can be seen as avidence for this factor being highly enriched (low p value) or not (high p value) for the input gene list estimate an estimate of the odds ratio. Note that the conditional Maximum Likelihood Estimate (MLE) rather than the unconditional MLE (the sample odds ratio) is used. Only present in the 2 by 2 case. alternative a character string describing the alternative hypothesis. null.value the odds ratio under the null, or. Only present in the 2 by 2 case. method the character string "Fisher's Exact Test for Count Data". enrichedgenes list of the genes from the input gene list that were enriched for this factor as a

## Author(s)

James Malone, European Bioinformatics Institute Simon Jupp, European Bioinformatics Institute Maryam Soleimani, European Bioinformatics Institute

list of Gene URIs. To get the gene names use the function  $\mathtt{getClassLabel}$ 

#### References

Agresti, A. (1990) Categorical data analysis. New York: Wiley. Pages 59-66.

```
###perform gene set enrichment using Atlas as background to find factors genes are enriched for
#load the background files for the species of interest (requires downloading)
#load("human_gene_list.RData") #human_genelist_bg
#load("human_factor_counts.RData") #human_factor_counts

###creat or load your genelist of interest
genelist <- c("GSTT1", "PLCE1", "XIST", "ENOSF-1", "AHR", "FSTL1", "YIPF5", "RBM5", "TNP01", "CREB1", "NO)
###get id for species 'human'</pre>
```

```
taxon <-getTaxonURI("human") #obo:NCBITaxon_9606

###do enrichment
#results <- doFishersEnrichmentForGeneNames(genelist, taxon, human_genelist_bg, human_factor_counts)</pre>
```

drawHeatMapForAtlasExperiment

Produce a heatmap of differentially expressed genes vs conditions for an Atlas experiment.

## **Description**

Draws a heatmap of genes that are differentially expressed beyond a given t stat signficance against conditions they are diff expressed for for a given Atlas experiment.

## Usage

draw Heat Map For Atlas Experiment (experiment id, tstat significance = 5, numoff actors diff expressed and taken to the following of the control of the following properties of the control of the con

#### **Arguments**

experimentid Atlas experiment ID which is to be visualized tstatsignificance

The significance beyond which a gene should be included for visualisation. A significance value of '5' will only include genes with a t-stat of  $\geq 5$  or  $\leq -5$ .

 $numoff actors \\ diff expressed across$ 

Draw only genes which are differentially expressed across this number of spec-

ified factors (default is 1)

endpoint SPARQL endpoint to query data from. Default value is the live Atlas SPARQL

endpoint but this can be overidden.

#### Value

matrix of gene names with t-statistics for level of differential expression for each factor (one factor per column)

#### Author(s)

Simon Jupp, James Malone

```
#data <- drawHeatMapForAtlasExperiment("E-MTAB-1670", 12)</pre>
```

enrichmentresult-class 9

```
enrichmentresult-class
```

Class "enrichmentresult"

#### **Description**

Class to store the enrichment for a single experimental factor for a given gene set.

## Usage

```
#enrichmentresult()
```

## **Objects from the Class**

Objects can be created by calls of the form enrichmentresult().

#### **Fields**

```
factoruri: Object of class "character" the URI of the experimental factor
label: Object of class "character" human readable label of the experimental factor
p.value: Object of class "numeric" p.value for enrichment for this factor
estimate: Object of class "numeric" see Fishers exact test
alternative: Object of class "character" see Fishers exact test
null.value: Object of class "numeric" see Fishers exact test
method: Object of class "character" Fishers exact test
enrichedgenes: Object of class "vector" Vector of the genes enriched for this factor from the
gene list
```

## Methods

No methods defined with class "enrichmentresult" in the signature.

## Author(s)

James Malone

```
showClass("enrichmentresult")
```

excludeSubclasses

Exclude subclasses of factors from enrichment results

#### **Description**

Removes all factors which are subclasses of a specified type from Atlas gene set enrichment results. For instance, if all chemical compounds were to be excluded form the enrichment results or all subclasses of cancer use this function.

## Usage

excludeSubclasses(filterparentclass, resultset, endpoint="http://www.ebi.ac.uk/rdf/services/atla

## **Arguments**

filterparentclass

The class from which all subclasses will be excluded - including this class.

resultset Result set from an Atlas gene set enrichment.

endpoint SPARQL endpoint to query data from. Default value is the live Atlas SPARQL

endpoint but this can be overidden.

#### Value

new enrichment result set with the specified class and subclasses excluded.

## Author(s)

James Malone, Simon Jupp

## **Examples**

####filter out all subclasses of chemical compounds from the result set
#####enrichment results has been produced using the enrichment functions in AtlasRDF package
#filteredresults <- excludeSubclasses("obo:CHEBI\_37577", enrichmentresults)
#vizPvalues(filteredresults, 0.01)</pre>

factorbackground-class

Class "factorbackground"

## Description

Used in enrichment to represent an experimental factor and any associated genes for that factor as well as additional ontological information about the factor.

#### **Extends**

All reference classes extend and inherit methods from "envRefClass".

factorbackground-class

11

#### **Fields**

uri: Object of class character URI of the experimental factor.
label: Object of class character name of the experimental factor.
species: Object of class character Species ID of the experimental factor.
geneuris: Object of class vector URIs of the genes for this experimental factor.
numgenesexpressed: Object of class integer Count of the genes expressed for this factor.
numgenesnotexpressed: Object of class integer Count of the genes not expressed for this factor from the total genes for this species.
subclasses: Object of class vector Subclass of this factor from ontology

superclasses: Object of class vector Superclasses of this factor from ontology

#### Methods

```
mergegeneuris(value): Merge gene URIs into the current vector
getsuperclasses(): Get superclasses for this factor from the ontology
setsuperclasses(value): Set superclasses for this factor from the ontology
getsubclasses(): Get subclasses for this factor from the ontology
setsubclasses(value): Set ontology subclasses for this factor
getnumgenesnotexpressed(): Get the number of genes not expressed for this factor as a total
     from total number of genes for this species
setnumgenesnotexpressed(value): Set the number of genes not expressed for this factor as a
     total from total number of genes for this species
getnumgenesexpressed(): Get the number of genes expressed for this factor.
setnumgenesexpressed(value): Set the number of genes expressed for this factor.
getgeneuris(): Get URIs for genes differentially expressed for this factor.
setgeneuris(value): Set URIs for genes differentially expressed for this factor.
getspecies(): Get species ID for this factor.
setspecies(value): Set species ID for this factor.
getlabel(): Get name for this factor.
setlabel(value): Set name for this factor.
geturi(): Get the URI for this factor.
seturi(value): Set the URI for this factor.
```

## Author(s)

James Malone, Simon Jupp

```
showClass("factorbackground")
```

12 generef-class

generef-class

Class "generef"

#### **Description**

Class to store gene references, including the URI, name and experimental factors associated with the gene.

#### **Extends**

All reference classes extend and inherit methods from "envRefClass".

#### **Fields**

```
geneuri: Object of class character the URI of the gene.
genelabel: Object of class characterGene name.
geneensemblid: Object of class character ENSEMBL ID of the gene, if available.
species: Object of class character Species ID of the gene.
exfactoruris: Object of class vector Exerimental factors associated to this gene.
```

#### Methods

```
mergeexfactoruris(value): merge an experimental factor(s) into the list getexfactoruris(): get the list of experimental factors for this gene getspecies(): Get the species ID for this gene setspecies(value): Set the species ID for this gene getensemblid(): Get the ENSEMBL ID for this gene. setensemblid(value): Set the ENSEMBL ID for this gene. getgenelabel(): Get gene name. setgenelabel(value): Set gene name. getgeneuri(): Get the URI for the gene. setgeneuri(value): Set the URI for the gene.
```

#### Author(s)

```
James Malone, Simon Jupp
```

```
showClass("generef")
```

getAllEnsemblGenesForExFactor

Get all ENSEMBL genes reported as differentially expressed for a given experimental factor.

## Description

Get all ENSEMBL genes reported as differentially expressed for a given experimental factor. Returns the gene URI, ENSEMBL Id and common gene name.

## Usage

```
getAllEnsemblGenesForExFactor(exfactor, limit, endpoint)
```

#### **Arguments**

exfactor The URI of the experimental factor e.g. "efo:EFO\_0001420"

limit Limit the number of genes returned by the query to this, no limit specified or

value of 0 returns all.

endpoint SPARQL endpoint to query data from. Default value is the live Atlas SPARQL

endpoint but this can be overidden.

#### Value

Data frame describing genes for the specified experimental factor with the following slots:

dbXref the URI of the gene

geneName the common name of the gene ensemblid the ENSEMBL id of the gene

propertyValue the property value for this gene corresponding to the experimental factor in the

query

#### Author(s)

James Malone, Simon Jupp

```
###get the genes for the experimental factor 'sepsis' (efo:EFO_0001420)
#results <- getAllEnsemblGenesForExFactor("efo:EFO_0001420")
#results[1,] #first gene result</pre>
```

getExFactorURIFromLabel

Get the EFO URI from a given label

## **Description**

Find the EFO ontology class URI for a given search label.

## Usage

getExFactorURIFromLabel(label, endpoint="http://www.ebi.ac.uk/rdf/services/atlas/sparql")

## **Arguments**

label Search string to find the EFO class URI by

endpoint SPARQL endpoint to query data from. Default value is the live Atlas SPARQL

endpoint but this can be overidden.

#### Value

EFO class URI for the given search term

#### Author(s)

James Malone, Simon Jupp

## **Examples**

```
####find the efo class uri for "asthma"
getExFactorURIFromLabel("asthma") #<http://www.ebi.ac.uk/efo/EFO_0000270>
```

getExperimentIdsForGeneURI

Get ArrayExpress experiment ID which contain a gene specified by URI.

## Description

Get all of the ArrayExpress experiment IDs which contain a gene, specified by a URI.

## Usage

getExperimentIdsForGeneURI(geneuri, endpoint = "http://www.ebi.ac.uk/rdf/services/atlas/sparq

## **Arguments**

geneuri Gene URI

endpoint The location of the SPARQL endpoint - default value is live Atlas endpoint

#### Value

List of ArrayExpress experiment IDs for the specified gene.

#### Author(s)

James Malone, Simon Jupp

## **Examples**

```
###get the experiment IDs for a gene ENSG00000142556 (ENSEMBL ID)
#getExperimentIdsForGeneURI("http://identifiers.org/ensembl/ENSG00000142556")
```

 ${\tt getExperimentsByDescription}$ 

Search for experiments in Atlas based on a string, e.g. liver, cancer etc.

## Description

Find experiments in Atlas based on a search string such as a "liver" or "cancer". Returns the experiment IDs plus the text in which the search string was found.

## Usage

getExperimentsByDescription(searchterm, limit = 0, endpoint = "http://www.ebi.ac.uk/rdf/services/

## Arguments

searchterm The search term.

limit Optional limit on number of returned results (default is everything)

endpoint The location of the SPARQL endpoint - default value is live Atlas endpoint

#### Value

\$experiment Atlas Experiment ID of matched experiment to search term'

\$description Description of experiment which matched term

## Author(s)

James Malone, Simon Jupp

## **Examples**

###get experiments which contain the word liver in the description
getExperimentsByDescription("liver")

getExperimentURIsForGeneId

Get experiment URIs which contain a gene specified by ENSEMBL ID.

## Description

Get all of the experiment URIs which contain a gene, specified by an ENSEMBL ID, which has been reported as differentially expressed with said experiment.

## Usage

getExperimentURIsForGeneId(geneid, endpoint = "http://www.ebi.ac.uk/rdf/services/atlas/sparql

## **Arguments**

geneid ENSEMBL gene ID

endpoint The location of the SPARQL endpoint - default value is live Atlas endpoint

#### Value

List of experiment URIs for the specified gene. Note, the last part of the URI is the accession number used in Arrayexpress to identify an experiment.

## Author(s)

James Malone, Simon Jupp

## **Examples**

```
###get the experiment IDs for a gene ENSMUSG00000031314 (ENSEMBL ID) #getExperimentURIsForGeneId("ENSMUSG00000031314")
```

getGeneListFromPubmedid

Get gene lists in Atlas for a given pubmed id (assuming the data is in Atlas)

## **Description**

Get differentially expressed genes reported in an experiment in Atlas for which a given pubmed id is linked (assuming the data is in Atlas)

## Usage

```
getGeneListFromPubmedid(searchid, endpoint = "http://www.ebi.ac.uk/rdf/services/atlas/sparql")
```

#### **Arguments**

searchid PubMed ID to find data for

endpoint SPARQL endpoint to query data from. Default value is the live Atlas SPARQL

endpoint but this can be overidden.

#### Value

List of genes for experiment (via PubMed ID)

#### Author(s)

James Malone, Simon Jupp

#### **Examples**

```
###get gene list for the publication with pubmed id 19668377
#getGeneListFromPubmedid("19668377")
```

getGenesForExperimentID

Get all of the genes reported in an experiment, speicifed by the Atlas experiment ID.

## Description

Get all of the gene as reported within an experiment. The experiment is specified as an Atlas experiment ID e.g. E-GEOD-3865

#### Usage

getGenesForExperimentID(experiment, endpoint = "http://www.ebi.ac.uk/rdf/services/atlas/sparql")

## **Arguments**

experiment Atlas experiment ID.

endpoint The location of the SPARQL endpoint - default value is live Atlas endpoint

## Value

data frame containg following elements

expressionValue

The summarised expression value for this gene

dbXref The URI identifier for this gene

pvalue The p-value representing differentially expression for this gene

propertyValue The property value for which this gene was reported as differentially expressed

#### Author(s)

James Malone, Simon Jupp

#### **Examples**

```
###get genes for experiment ID E-MEXP-3739
#Can take several minutes to complete
#res <- getGenesForExperimentID("E-MEXP-3739")
###list first gene in the experiment
#res[1,]</pre>
```

getGenesForExperimentURI

Get all of the genes reported in an experiment, speicifed by the Atlas experiment URI.

## Description

Get all of the gene as reported within an experiment. The experiment is specified as an Atlas experiment URI e.g. <a href="http://rdf.ebi.ac.uk/resource/atlas/E-GEOD-13396">http://rdf.ebi.ac.uk/resource/atlas/E-GEOD-13396</a>>

### Usage

getGenesForExperimentURI(experiment, endpoint = "http://www.ebi.ac.uk/rdf/services/atlas/sparql"

#### **Arguments**

experiment Atlas experiment URI.

endpoint The location of the SPARQL endpoint - default value is live Atlas endpoint

#### Value

data frame containg following elements

expressionValue

The summarised expression value for this gene

dbXref The URI identifier for this gene

pvalue The p-value representing differentially expression for this gene

propertyValue The property value for which this gene was reported as differentially expressed

## Author(s)

James Malone, Simon Jupp

```
###get genes for experiment <a href="http://rdf.ebi.ac.uk/resource/atlas/E-GEOD-13396">#Can take several minutes to complete
#res <- getGenesForExperimentURI("<a href="http://rdf.ebi.ac.uk/resource/atlas/E-MEXP-3739">| ###list first gene in the experiment
#res[1,]
```

getGenesForPathwayURI Get genes associated with a signalling pathway from Reactome.

#### **Description**

Get genes that are associated with proteins which are in turn associated to a specific signalling pathway in Reactome.

## Usage

```
getGenesForPathwayURI(pathwayuri, endpoint = "http://www.ebi.ac.uk/rdf/services/atlas/sparql")
```

## **Arguments**

pathwayuri The URI of a reactome pathway.

endpoint SPARQL endpoint to query data from. Default value is the live Atlas SPARQL

endpoint but this can be overidden.

#### Value

List of gene URIs for the pathway

#### Author(s)

James Malone, Simon Jupp

## **Examples**

```
###get gene list for the pathway AKT Signaling in Cancer
#getGenesForPathwayURI("<a href="http://identifiers.org/reactome/REACT_147727.2>")
```

```
{\tt getGeneUriFromEnsemblId}
```

Get gene URI from an ENSEMBL ID.

## **Description**

Get the URI for an ENSEMBL gene based on the ENSEMBL ID.

## Usage

```
getGeneUriFromEnsemblId(id, endpoint = "http://www.ebi.ac.uk/rdf/services/atlas/sparql")
```

## **Arguments**

id ENSEMBL gene ID

endpoint The location of the SPARQL endpoint - default value is live Atlas endpoint

#### Value

List of URIs for a given ENSEMBL ID - this is almost always one but it is feasible there are two so be aware.

#### Author(s)

James Malone, Simon Jupp

## **Examples**

```
###get the URI identifier for a gene based on the ENSEMBL ID
#
getGeneUriFromEnsemblId("ENSMUSG00000031314")
```

getGeneUriFromName

Get URI of a gene based on the common gene name.

#### **Description**

Get the URI of an ENSEMBL gene based on a search by the common gene name.

## Usage

getGeneUriFromName(genename, taxon, endpoint = "http://www.ebi.ac.uk/rdf/services/atlas/sparq")

## **Arguments**

genename The common gene name by which to search

taxon The URI of the taxon to search genes for, use function getTaxonURI to get the

URI

endpoint The location of the SPARQL endpoint - default value is live Atlas endpoint

## Value

list of gene URIs matching the query: note this can be more than one

#### Author(s)

James Malone, Simon Jupp

```
###get the gene URI identifier based on the common gene name for "SRGN" in human
#getGeneUriFromName("SRGN", "obo:NCBITaxon_9606")
```

getLabel 21

getL	ahal
gett	aber

Get label of an entity based on the URI.

## **Description**

Every ontology class has a human readable label associated with it, thhis function will enable you to get that readable label based on the URI (the identifier) for that entity. For instance, the URI http://www.ebi.ac.uk/efo/EFO\_0001185 has label HeLa.

## Usage

```
getLabel(uri, endpoint = "http://www.ebi.ac.uk/rdf/services/atlas/sparql")
```

## **Arguments**

uri URI of the class for which the label is required.

endpoint The location of the SPARQL endpoint - default value is live Atlas endpoint

#### Value

List of labels for the class URI

#### Author(s)

James Malone, Simon Jupp

#### **Examples**

```
###get the label for the class efo:EFO_0001185 (HeLa)
getLabel("efo:EFO_0001185")
```

getOntologyMappings

Get mappings for a given ontology class URI to EFO using the NCBO BioPortal

## Description

Get mappings for a given class URI from an ontology other than EFO using the NCBO BioPortal. Function is useful for querying the Atlas RDF using your own ontology. For instance, if you are using ICD-10 or SnoMed this function will enable you to extract any mappings to EFO, used to annotate the Atlas data, thereby enabling querying by your ontology of interest. The returned EFO class should then be used to query Atlas using the other query functions.

## Usage

```
getOntologyMappings(searchuri, endpoint = "http://www.ebi.ac.uk/rdf/services/atlas/sparql")
```

## **Arguments**

searchuri The ontology class URI for which mappings to EFO are required.

endpoint SPARQL endpoint to query data from. Default value is the live Atlas SPARQL

endpoint but this can be overidden.

#### Value

List of EFO URI(s) to which the class maps

#### Author(s)

James Malone, Simon Jupp

#### References

NCBO BioPortal - http://bioportal.bioontology.org/

#### **Examples**

###find mappings in EFO from <a href="http://purl.bioontology.org/ontology/SNOMEDCT/87163000">http://purl.bioontology.org/ontology/SNOMEDCT/87163000</a> (leukemia in snomed)
#getOntologyMappings("<a href="http://purl.bioontology.org/ontology/SNOMEDCT/87163000")</pre>

getPathwayForGeneId

Get pathways associated to a gene.

## **Description**

Get signalling pathways from Reactome that are associated to a gene, via associated protein(s) which are in turn connected to said signalling pathway(s) in Reactome.

#### Usage

getPathwayForGeneId(geneid, endpoint = "http://www.ebi.ac.uk/rdf/services/atlas/sparql")

#### **Arguments**

geneid The ENSEMBL ID for a gene.

endpoint SPARQL endpoint to query data from. Default value is the live Atlas SPARQL

endpoint but this can be overidden.

#### Value

List of pathway URIs.

### Author(s)

James Malone, Simon Jupp

```
###get pathways for the gene SNHG5
#getPathwayForGeneId("http://identifiers.org/ensembl/ENSG00000203875")
```

getPathwaysFromGenesAndCondition

Get pathways connected to genes which are differentially expressed for a user specified condition.

## **Description**

Get list of pathway names that are connected to genes (via proteins which are encoded for by those genes) and which are reported as differentially expressed under a specified condition. The condition is specified as an ontology URI.

## Usage

getPathwaysFromGenesAndCondition(condition, endpoint = "http://www.ebi.ac.uk/rdf/services/atl

#### **Arguments**

condition URI of a condition based on the EFO ontology class.

endpoint The location of the SPARQL endpoint - default value is live Atlas endpoint

## Value

Returns a data frame with the following:

pathwayname The name of the pathway

pathway The URI identifier of the pathway

expressionvalue

The gene and expression for the pathway

p-value p-value of the differential expression of the gene for this pathway in this condi-

tion

## Author(s)

James Malone, Simon Jupp

## **Examples**

###get pathways connected to genes which are differentially expressed for HeLa (efo:EFO\_00011185)
#getPathwaysFromGenesAndCondition("efo:EFO\_0001185")

getPathwayUriFromName Get pathway URI given a pathway name

## Description

Given the name of a pathway find URI(s) that match this name

## Usage

getPathwayUriFromName(name, endpoint = "http://www.ebi.ac.uk/rdf/services/atlas/sparql")

## **Arguments**

name The textual name of the pathway to search for

endpoint The location of the SPARQL endpoint - default value is live Atlas endpoint

#### Value

data frame containing slots

pathwayuri The URI of the corresponding pathway

label The textual human readable name of the pathway

#### Author(s)

James Malone, Simon Jupp

## **Examples**

 $\label{thm:phrase} \begin{tabular}{ll} \tt\#get\ pathways\ which\ have\ the\ phrase\ 'ligand\ binding'\ included\ in\ their\ name \\ \tt\#getPathwayUriFromName("ligand\ binding") \\ \end{tabular}$ 

getRankedPathwaysForGeneIds

Get pathway names and URIs for given gene list, sorted into list with most common pathways first.

## **Description**

Given a list of ENSEMBL gene IDs, the function will find pathways which these genes are involved in. These pathways will be grouped and ranked according to those with highest number of genes in common.

## Usage

getRankedPathwaysForGeneIds(genelist, endpoint = "http://www.ebi.ac.uk/rdf/services/atlas/sparql

## **Arguments**

genelist The list of ENSEMBL gene IDs to find pathways for.

endpoint The location of the SPARQL endpoint - default value is live Atlas endpoint

#### Value

A ranked list (pathway with greatest number of genes first) of pathwayresult classes with the following slots:

pathwayuri The URI of the corresponding pathway

label The textual human readable name of the pathway

numgenes A count of the number of genes from the gene list associated with this pathway

genes Vector of genes from the gene list that are associated with this pathway

#### Author(s)

James Malone, Simon Jupp

## **Examples**

getSpeciesSpecificEnsemblGenesForExFactor

Get ENSEMBL genes which have been differentially expressed for a given condition and species.

#### **Description**

Get the ENSEMBL gene IDs and names which are reported as differentially expressed in Atlas for a specified conditions and species.

#### Usage

getSpeciesSpecificEnsemblGenesForExFactor(exfactor, taxon, limit = 0, endpoint="http://www.ebi.ac

## Arguments

exfactor URI of the experimental factor search term

taxon URI of the species to limit search to

limit Optional limit to number of genes returned (default is all)

endpoint The location of the SPARQL endpoint - default value is live Atlas endpoint

#### Value

data frame of results with the following:

dbXref URI identifier of the gene genename Common gene name ensemblid ENSEMBL ID of gene

#### Author(s)

James Malone, Simon Jupp

#### **Examples**

###get genes differentially expressed for HeLa cells (efo:EFO\_0001185) in human (obo:NCBITaxon\_9606) #getSpeciesSpecificEnsemblGenesForExFactor("efo:EFO\_0001185", "obo:NCBITaxon\_9606")

getTaxonURI

Get URI of a taxon class based on the common or taxonomic name.

## Description

Convenience method to the ontology URI, used in other queries, based on an input common or taxonomic name such as "human" or "Homo sapiens"

#### Usage

```
getTaxonURI(taxonName)
```

#### **Arguments**

taxonName

Common or taxonomic name

#### Value

URI of the taxon

## Author(s)

James Malone, Simon Jupp

## **Examples**

```
###get URI for human
getTaxonURI("human")
###or for arabidopsis
getTaxonURI("arabidopsis")
```

includeOnlySubclasses Filter enrichment results to only include experimental factors of a given parent class (e.g. all cancers)

## **Description**

Function which allows filtering of enrichment results to remove experimental factors apart from those factors which are a subclasses of a given parent class. For instance, include only disease factors or only cell types.

#### Usage

includeOnlySubclasses(includeparentclass, resultset, endpoint = "http://www.ebi.ac.uk/rdf/ser

orderEnrichmentResults 27

#### **Arguments**

includeparentclass

The URI of the parent class for which all experimental factor subclasses will be

included (removing all others).

resultset The full enrichment result set

endpoint The location of the SPARQL endpoint - default value is live Atlas endpoint

## Value

New Atlas enrichment result set, filtered to include only experimental factors specified in the function call.

#### Author(s)

James Malone, Simon Jupp

#### **Examples**

```
###include only experimental factors which are diseases in the enrichment results
#filteredgenes <- includeOnlySubclasses("efo:EFO_0000408", mouseresults, endpoint="http://wwwdev.ebi.ac.
###view results
#vizPvalues(filteredgenes)</pre>
```

orderEnrichmentResults

Order the results of a gene set enrichment by p-value.

## **Description**

Following a gene set enrichment, order the results so that the most significantly enriched factors are first, in decreasing order of significane (i.e. where lowest p-value is most significant)

#### Usage

```
orderEnrichmentResults(resultset)
```

## Arguments

resultset A set of results following an AtlasRDF gene set enrichment.

#### Value

Ordered set of results with most signficant factor first, least signficant last.

## Author(s)

James Malone

28 pathwayresult-class

## **Examples**

```
###order the results of an enrichment
#orderedresults <- orderEnrichmentResults(enrichmentresults)
###view the top 10 most significant
#vizPValues(orderedresults[1:20], cutoff = "1")</pre>
```

pathwayresult-class

Class "pathwayresult"

## **Description**

Class to store information about a pathway and associated genes.

## Usage

```
#pathwayresult()
```

## **Objects from the Class**

Objects can be created by calls of the form pathwayresult().

## **Fields**

```
pathwayuri: Object of class "character" the URI of the pathway from Reactome.

label: Object of class "character" The human readable label of the pathway.

numgenes: Object of class "numeric" A count of the number of genes for this pathway.

genes: Object of class "vector" List of the genes associated with this pathway.
```

## Methods

No methods defined with class "pathwayresult" in the signature.

#### Author(s)

James Malone

```
showClass("pathwayresult")
```

searchForEFOTerms 29

		erms

Search for EFO URIs from a given label text

## **Description**

Finds any EFO ontology class whose label matches the given search label or part of it.

#### Usage

searchForEFOTerms(label, endpoint="http://www.ebi.ac.uk/rdf/services/atlas/sparql")

## **Arguments**

label Search string to find the EFO class URI by

endpoint SPARQL endpoint to query data from. Default value is the live Atlas SPARQL

endpoint but this can be overidden.

#### Value

Data frame of EFO class URIs and their corresponding labels

#### Author(s)

James Malone, Simon Jupp

#### **Examples**

 $\verb|###find|$  the efo classes which contain the word 'cancer' in their label  $\verb|#searchForEFOTerms("cancer")|$ 

transcription\_pathway\_enrichment

Expression Gene Set Enrichment result for

## Description

A result set example from running the Atlas Expression gene set enrichment function. The gene list was originally extracted form a transcription pathways in Reactome and enrichment performed to see which experimental factors were enriched for this pathway.

## Usage

transcription\_pathway\_enrichment

## **Format**

A list of enrichmentresult objects

#### **Source**

Gene Expression Atlas RDF at European Bioinformatics Institute

30 vizPvalues

vizPvalues

Visualize the results of an enrichment filtering for a specificed p-value.

## **Description**

Produce a plot of the most enriched experimental factors following an Atlas gene set enrichment. The cutoff allows a filter to be placed by p-value.

## Usage

```
vizPvalues(resultset, cutoff = "0.05")
```

## Arguments

resultset A set of results following an AtlasRDF gene set enrichment.

cutoff The p-value threshold at which to include a gene for visualization

## Value

Plot of results

#### Author(s)

James Malone

## **Examples**

###visualize the results from a gene set enrichment performed using AtlasRDF package #vizPValues(enrichmentresults, cutoff = "0.02")

# Index

*Topic <b>classes</b>	<pre>getGenesForExperimentID, 17</pre>
enrichmentresult-class, 9	getGenesForExperimentURI, 18
factorbackground-class, 10	getGenesForPathwayURI, 19
generef-class, 12	getGeneUriFromEnsemblId, 19
pathwayresult-class, 28	getGeneUriFromName, 20
*Topic datasets	getLabel, 21
transcription_pathway_enrichment,	getOntologyMappings, 21
29	getPathwayForGeneId, 22
*Topic <b>package</b>	${\tt getPathwaysFromGenesAndCondition}, {\tt 23}$
AtlasRDF-package, 2	getPathwayUriFromName, 24
	<pre>getRankedPathwaysForGeneIds, 24</pre>
AtlasRDF (AtlasRDF-package), 2	${\tt getSpeciesSpecificEnsemblGenesForExFactor},$
AtlasRDF-package, 2	25
class:enrichmentresult	getTaxonURI, 26
(enrichmentresult-class), 9	includeOnlySubclasses, 26
class: factorbackground	and an Francish mount Decoults 27
(factorbackground-class), 10	orderEnrichmentResults, 27
<pre>class:generef (generef-class), 12 class:pathwayresult</pre>	pathwayresult (pathwayresult-class), 28
(pathwayresult-class), 28	pathwayresult-class, 28
(pathwayresurt-crass), 28	pathwayi csait class, 20
doFishersEnrichment, 4	searchForEF0Terms, 29
<pre>doFishersEnrichmentForEnsemblIds, 5</pre>	
doFishersEnrichmentForGeneNames, 6	transcription_pathway_enrichment, 29
<pre>drawHeatMapForAtlasExperiment, 8</pre>	vi=Dvalvaa 20
	vizPvalues, 30
enrichmentresult	
(enrichmentresult-class), 9	
enrichmentresult-class, 9	
envRefClass, 10, 12	
excludeSubclasses, 10	
factorbackground	
(factorbackground-class), 10	
factorbackground-class, 10	
Tactor background Class, 10	
generef (generef-class), 12	
generef-class, 12	
<pre>getAllEnsemblGenesForExFactor, 13</pre>	
getExFactorURIFromLabel, 14	
<pre>getExperimentIdsForGeneURI, 14</pre>	
<pre>getExperimentsByDescription, 15</pre>	
<pre>getExperimentURIsForGeneId, 16</pre>	
<pre>getGeneListFromPubmedid, 16</pre>	