

# Package ‘loci2path’

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**Type** Package

**Title** Loci2path: regulatory annotation of genomic intervals based on tissue-specific expression QTLs

**Version** 1.29.1

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**Description** loci2path performs statistics-rigorous enrichment analysis of eQTLs in genomic regions of interest.  
Using eQTL collections provided by the Genotype-Tissue Expression (GTEx) project and pathway collections from MSigDB.

**License** Artistic-2.0

**Encoding** UTF-8

**LazyData** true

**Depends** R (>= 3.5)

**Imports** pheatmap, wordcloud, RColorBrewer, data.table, methods,  
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S4Vectors

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**VignetteBuilder** knitr

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**biocViews** FunctionalGenomics, Genetics, GeneSetEnrichment, Software,  
GeneExpression, Sequencing, Coverage, BioCarta

**URL** <https://github.com/StamleyXu/loci2path>

**BugReports** <https://github.com/StamleyXu/loci2path/issues>

**Collate** 'allClasses.R' 'allGenerics.R' 'allAccessor.R'  
'check.geneid.R' 'loci2path.demo-data.R'  
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Contents

|                                 |           |
|---------------------------------|-----------|
| biocarta . . . . .              | 2         |
| check.geneid . . . . .          | 3         |
| eqtlSet-class . . . . .         | 3         |
| eset.list . . . . .             | 4         |
| geneSet-class . . . . .         | 5         |
| getHeatmap . . . . .            | 6         |
| getMat . . . . .                | 7         |
| getPathDescription . . . . .    | 7         |
| getPval . . . . .               | 8         |
| getTissueDegree . . . . .       | 9         |
| getWordcloud . . . . .          | 10        |
| loci2pathResult-class . . . . . | 10        |
| query . . . . .                 | 11        |
| query.gr . . . . .              | 13        |
| <b>Index</b>                    | <b>14</b> |

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|          |                                                |
|----------|------------------------------------------------|
| biocarta | <i>eQTL geneset enrichment query demo data</i> |
|----------|------------------------------------------------|

---

Description

Demo Data to show how to perform eQTL-geneset enrichment query.

Usage

`data(loci2path.demo)`

Format

An object of class `geneSet` of length 1.

Details

- `eqtl.set.list` A list of `eQTLset` objects; eQTL data are collected from GTeX.
- `biocarta` A `Geneset` object; Geneset are from Broad Institute’s MSigDB: curated gene set, category ‘cp’: BIOCARTA
- `query.gr` A `Genomic Region` object; Query regions are from immunoBase, crohn’s disease.

Examples

`data(loci2path.demo)`

---

|              |                                                                              |
|--------------|------------------------------------------------------------------------------|
| check.geneid | <i>check compatibility of gene identifiers between eQTL set and gene set</i> |
|--------------|------------------------------------------------------------------------------|

---

**Description**

This function perform enrichment test between one eQTL set and one gene set

**Usage**

```
check.geneid(e.set, g.set)
```

**Arguments**

|       |                                                       |
|-------|-------------------------------------------------------|
| e.set | an eqtlSet object; the eQTL set to be queried against |
| g.set | an object of geneSet class; the gene set to be tested |

**Value**

a data.frame shows the number of genes from (1) eqtl Set (2) gene Set (3) shared

**Examples**

```
check.geneid(eset.list$Skin, biocarta)
```

---

|               |                      |
|---------------|----------------------|
| eqtlSet-class | <i>eqtlSet Class</i> |
|---------------|----------------------|

---

**Description**

eqtlSet Class contains information for eqtl-gene association, gene identifier, position of SNPs, etc.

**Usage**

```
tissue(x)

eqtlId(x)

eqtlRange(x)

eqtlGene(x)

## S4 method for signature 'eqtlSet'
tissue(x)

## S4 method for signature 'eqtlSet'
eqtlId(x)

## S4 method for signature 'eqtlSet'
eqtlRange(x)

## S4 method for signature 'eqtlSet'
eqtlGene(x)
```

**Arguments**

x                      An eqtlSet object

**Value**

Object of class eqtlSet

**Slots**

tissue character; name of the cell/tissue of the eQTL study  
 eqtlId character; name of the SNPs  
 eqtlRange GenomicRanges; position of the SNPs  
 gene character; gene identifier

**Examples**

```
require(GenomicRanges)
brain.file <- system.file("extdata", "eqtl/brain.gtex.txt",
  package="loci2path")
tab <- read.table(brain.file, stringsAsFactors=FALSE, header=TRUE)
eqtlRange <- GRanges(seqnames=Rle(tab$snp.chr),
  ranges=IRanges(start=tab$snp.pos,
    width=1))
brain.eset <- eqtlSet(tissue="brain",
  eqtlId=tab$snp.id,
  eqtlRange=eqtlRange,
  gene=as.character(tab$gene.entrez.id))
tissue(brain.eset)
head(eqtlId(brain.eset))
eqtlRange(brain.eset)
head(eqtlGene(brain.eset))
```

---

eset.list

*eQTL geneset enrichment query demo data*

---

**Description**

Demo Data to show how to perform eQTL-geneset enrichment query.

**Usage**

```
data(loci2path.demo)
```

**Format**

An object of class list of length 3.

**Details**

eset.list A list of eQTLset objects; eQTL data are collected from GTeX.

biocarta A Geneset object; Geneset are from Broad Institute's MSigDB: curated gene set, category 'cp': BIOCARTEA

query.gr A Genomic Region object; Query regions are from immunoBase, Psoriasis disease.

**Examples**

```
data(loci2path.demo)
```

---

|               |                      |
|---------------|----------------------|
| geneSet-class | <i>geneSet Class</i> |
|---------------|----------------------|

---

**Description**

geneSet Class contains information for names of gene sets and a list of gene sets

**Usage**

```
numGene(x)

description(x)

geneSetList(x)

## S4 method for signature 'geneSet'
numGene(x)

## S4 method for signature 'geneSet'
description(x)

## S4 method for signature 'geneSet'
geneSetList(x)
```

**Arguments**

x                      An geneSet object

**Value**

Object of class geneSet

**Slots**

numGene   numeric; the total number of all genes; This number is used in enrichment tests

description   vector of character; additional information for gene sets, such as names, URLs, a short description, etc.

geneSetList   list; a list of gene sets; each member is a vector containing a group of gene identifiers

**Examples**

```
biocarta.link.file <- system.file("extdata",
  "geneSet/biocarta.txt", package="loci2path")
biocarta.link <- read.delim(biocarta.link.file, header=FALSE,
  stringsAsFactors=FALSE)
biocarta.set.file <- system.file("extdata", "geneSet/biocarta.set.txt",
  package="loci2path")
set.geneid <- read.table(biocarta.set.file, stringsAsFactors=FALSE)
```

```

set.geneid <- strsplit(set.geneid[,1], split=",")
names(set.geneid) <- biocarta.link[,1]
biocarta <- geneSet(
  geneSetList=set.geneid,
  description=biocarta.link[,2],
  numGene=31847)
numGene(biocarta)
head(description(biocarta))
head(geneSetList(biocarta))

```

---

getHeatmap

*Generate heatmap of enrichment matrix from query result*


---

## Description

This function generate the enrichment heatmap using pheatmap package.

## Usage

```

getHeatmap(res, ...)

## S4 method for signature 'loci2pathResult'
getHeatmap(res, main = "",
  test.method = c("gene", "eqtl", "glm"), filter.quantile = 0.5,
  max.ptw.gene = 5000)

```

## Arguments

|                 |                                                                                                                                                                                                       |
|-----------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| res             | query result from function query.egset.list()                                                                                                                                                         |
| ...             | additional params                                                                                                                                                                                     |
| main            | title of the heatmap, default is ""                                                                                                                                                                   |
| test.method     | Choose which enrichment test should be used to retrieve p-values from. Options include:"gene"(default, gene-based fisher's exact test),"eqtl" (eqtl based fisher's exact test), "glm" (ordered query) |
| filter.quantile | Filter option; choose the max quantile of all p-values being kept in the matrix; default is 0.5, which means p-values larger than median p-values are discarded                                       |
| max.ptw.gene    | Filter option; minimum number of genes in a pathway; default is 5000 (pathway with >5000 genes are not included in the matrix)                                                                        |

## Value

|          |                   |
|----------|-------------------|
| pathways | frequent pathways |
| tissues  | frequent tissues  |

## Examples

```

result <- query(query.gr=query.gr,
  loci=eset.list, path=biocarta)
getHeatmap(result)

```

---

|        |                                                                   |
|--------|-------------------------------------------------------------------|
| getMat | <i>Extract tissue/geneset enrichment matrix from query result</i> |
|--------|-------------------------------------------------------------------|

---

## Description

This function extracts the enrichment matrix from eQTL list query result. The rows of the matrix are pathways; and the columns of the matrix are tissues/cell lines of the eQTL sets. P-Values from enrichment tests are summarized in this matrix

## Usage

```
getMat(res, ...)

## S4 method for signature 'loci2pathResult'
getMat(res, test.method = c("gene", "eqtl",
  "glm"), filter.quantile = 0.5, max.ptw.gene = 5000)
```

## Arguments

|                 |                                                                                                                                                                                                          |
|-----------------|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| res             | query result from function query.egset.list()                                                                                                                                                            |
| ...             | additional params                                                                                                                                                                                        |
| test.method     | Choose which enrichment test should be used to retrieve p-values from. Options include: "gene" (default, gene-based fisher's exact test), "eqtl" (eqtl based fisher's exact test), "glm" (ordered query) |
| filter.quantile | Filter option; choose the max quantile of all p-values being kept in the matrix; default is 0.5, which means p-values larger than median p-values are discarded                                          |
| max.ptw.gene    | Filter option; minimum number of genes in a pathway; default is 5000 (pathway with >5000 genes are not included in the matrix)                                                                           |

## Value

p-value matrix collected from enrichment result table

## Examples

```
result <- query(query.gr=query.gr,
  loci=eset.list, path=biocarta)
mat <- getMat(result, test.method="gene")
```

---

|                    |                                                                                        |
|--------------------|----------------------------------------------------------------------------------------|
| getPathDescription | <i>Extract description for enriched pathways from query result and gene-Set object</i> |
|--------------------|----------------------------------------------------------------------------------------|

---

## Description

This function extracts the pathway description from geneSet object.

**Usage**

```
getPathDescription(res, ...)

## S4 method for signature 'loci2pathResult'
getPathDescription(res, geneset)
```

**Arguments**

```
res          query result from function query.egset.list()
...          additional params
geneset      A geneSet object
```

**Value**

a vector of gene set description from geneSet description slot

**Examples**

```
result <- query(query.gr=query.gr,
  loci=eset.list, path=biocarta)
path.des <- getPathDescription(result, biocarta)
```

---

|         |                                                                                 |
|---------|---------------------------------------------------------------------------------|
| getPval | <i>Extract tissue/geneset enrichment p-value distribution from query result</i> |
|---------|---------------------------------------------------------------------------------|

---

**Description**

This function extracts the enrichment p-value distribution from eQTL list query result. P-values from different tissues/cell types are organized, and QQ-plot is generated against uniform distribution

**Usage**

```
getPval(res, ...)

## S4 method for signature 'loci2pathResult'
getPval(res, test.method = c("gene", "eqtl",
  "glm"))
```

**Arguments**

```
res          query result from function query.egset.list()
...          additional params
test.method  Choose which enrichment test should be used to retrieve p-values from. Options
              include: "gene" (default, gene-based fisher's exact test), "eqtl" (eqtl based fisher's
              exact test), "glm" (ordered query)
```

**Value**

generate pval distribution plot

**Examples**

```
result <- query(query.gr=query.gr,
  loci=eset.list, path=biocarta)
getPval(result, test.method="gene")
```

---

|                 |                                                |
|-----------------|------------------------------------------------|
| getTissueDegree | <i>Extract tissue degree from query result</i> |
|-----------------|------------------------------------------------|

---

**Description**

This function extracts the tissue degree from eQTL list query result for each pathway.

**Usage**

```
getTissueDegree(res, ...)

## S4 method for signature 'loci2pathResult'
getTissueDegree(res, loci)
```

**Arguments**

|      |                                                            |
|------|------------------------------------------------------------|
| res  | query result from function query.egset.list()              |
| ...  | additional params                                          |
| loci | a list of eqtlSet; each member should be an eqtlSet object |

**Value**

|                    |                                                                      |
|--------------------|----------------------------------------------------------------------|
| gene.tissue.map    | shows mapping:gene<->tissue                                          |
| gene.tissue.degree | shows tissue degree for each gene                                    |
| mean.tissue.degree | shows the average tissue degree for each pathway in the result table |

**Examples**

```
result <- query(query.gr=query.gr,
  loci=eset.list, path=biocarta)
tissue.degree=getTissueDegree(result, eset.list)
head(tissue.degree$gene.tissue.map)
head(tissue.degree$gene.tissue.degree)
head(tissue.degree$mean.tissue.degree)
```

---

|              |                                                                   |
|--------------|-------------------------------------------------------------------|
| getWordcloud | <i>Plot word cloud using frequent terms of pathways and genes</i> |
|--------------|-------------------------------------------------------------------|

---

### Description

This function draw the enrichment heatmap using wordcloud package.

### Usage

```
getWordcloud(res, ...)

## S4 method for signature 'loci2pathResult'
getWordcloud(res, min.freq.tissue = 5,
  min.freq.gset = 5, max.words = 50)
```

### Arguments

|                 |                                                                  |
|-----------------|------------------------------------------------------------------|
| res             | query result from function query.egset.list()                    |
| ...             | additional params                                                |
| min.freq.tissue | minimum frequency of tissue/cell to be plotted in the word cloud |
| min.freq.gset   | minimum frequency of geneset to be plotted in the word cloud     |
| max.words       | maximum words to be generated                                    |

### Value

empty

### Examples

```
result <- query(query.gr=query.gr,
  loci=eset.list, path=biocarta)
getWordcloud(result, min.freq.tissue=2, min.freq.gset=1)
```

---

loci2pathResult-class *loci2pathResult Class*

---

### Description

loci2pathResult Class contains information for the query result from query function query. Result object contains a ranked pathway table, and a vector of gene names that are associated with loci covered by query regions

**Usage**

```
resultTable(x)

coveredGene(x)

## S4 method for signature 'loci2pathResult'
resultTable(x)

## S4 method for signature 'loci2pathResult'
coveredGene(x)
```

**Arguments**

x                      An geneSet object

**Value**

Object of CClass loci2pathResult

**Slots**

resultTable data.frame; contains enrichment statistics, summary of eQTL and gene numbers, pathway names and gene names, etc.

coveredGene list; each member is a vector of genes associated with one tissue, whose associating loci are covered by query regions

**Examples**

```
result <- query(query.gr=query.gr,
  loci=eset.list, path=biocarta)
result
resultTable(result) # a data.frame for enriched pathways
coveredGene(result)
```

---

query

---

*Query enrichment in geneset through multiple eQTL sets.*


---

**Description**

This is the main function for loci2path query. Query can be made on either pathway enrichment or tissue-specificity, depending on the input Class. See **Details** for more.

**Usage**

```
query(query.gr, loci, path, ...)
```

## S4 method for signature 'GenomicRanges,list,ANY'

```
query(query.gr, loci, N = 2897310462)
```

## S4 method for signature 'GenomicRanges,eqlSet,geneSet'

```
query(query.gr, loci, path,
```

```

query.score = NULL, verbose = FALSE)

## S4 method for signature 'GenomicRanges,list,geneSet'
query(query.gr, loci, path,
      query.score = NULL, parallel = FALSE, verbose = FALSE)

```

### Arguments

|                          |                                                                                                                                                                                      |
|--------------------------|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <code>query.gr</code>    | a <code>GenomicRange</code> object, representing query regions                                                                                                                       |
| <code>loci</code>        | a list of <code>eQTLSet</code> ; each member should be an <code>eQTLSet</code> ; Or it can be a single <code>eQTLSet</code> .                                                        |
| <code>path</code>        | Pathways or <code>geneSets</code> to be tested for enrichment                                                                                                                        |
| <code>...</code>         | additional params                                                                                                                                                                    |
| <code>N</code>           | the total number of non-N nucleotides in the genome; default <code>N=2897310462</code> is for hg19                                                                                   |
| <code>query.score</code> | optional, set to <code>NULL</code> if the regions are not ordered. If the query regions are ordered, <code>query.score</code> is the quantity based on which the regions are ordered |
| <code>verbose</code>     | bool; whether to show <code>eQTLSet/geneSet</code> summary information; default is <code>FALSE</code>                                                                                |
| <code>parallel</code>    | bool; whether to enable parallel computing; default is <code>FALSE</code>                                                                                                            |

### Details

The user need to specify

1. Query region;
2. `loci`; one or more `eQTL` set; this is usually more than one `eQTL` set. Only multiple `eQTL` set derived from different cells/tissues will show cell/tissue specificity.
3. `path`; pre-defined Pathways, or gene sets. the gene sets that enrichment tests would be performed to.

`loci` must be provided; `path` is optional. When `path` is missing, the tissue-specificity query for the regions is performed.

The most common case for `loci` is an `eQTL` set list. This function perform enrichment test between one `eQTL` set and a group of gene sets. Usually query are based on `eQTL` set list, rather than only one `eQTL` set. Several result exploring functions (`getMat`, `getHeatmap`, `getPval`, etc...) are designed for query result from `eQTL` set list and gene sets. The class `loci2pathResult` is also designed for `eQTL` set list query result only. The result returns a `loci2pathResult` only the class of `loci` is a list of `eQTLSet`.

If user input one `eQTL` set as argument `loci`, a simple list object will be returned for specific research purpose.

### Value

a `data.frame` showing the tissue enrichment of the query regions by binomial test.  
 a list; `result.table` is the major result table showing enrichment assessment; `cover.gene` is the vector showing the genes from the `eQTL` Sets covered by the query region(s)  
 a `loci2pathResult` class object

### See Also

`loci2pathResult`

**Examples**

```

gr.tissue <- query(query.gr, eset.list)
#build one eqtlset
skin.eset <- eset.list$Skin
#query one egset
res.one <- query(query.gr, skin.eset, biocarta)
#enrichment result table
res.one$result.table
#all the genes associated with eQTLs covered by the query region
res.one$cover.gene
result <- query(query.gr=query.gr,
               loci=eset.list, path=biocarta)
#enrichment result table
resultTable(result)
#all the genes associated with eQTLs covered by the query region
coveredGene(result)

```

---

query.gr

*eQTL geneset enrichment query demo data*


---

**Description**

Demo Data to show how to perform eQTL-geneset enrichment query.

**Usage**

```
data(loci2path.demo)
```

**Format**

An object of class GRanges of length 47.

**Details**

eqtl.set.list A list of eQTLset objects; eQTL data are collected from GTeX.

biocarta A Geneset object; Geneset are from Broad Institute's MSigDB: curated gene set, category 'cp': BIOCARTE

query.gr A Genomic Region object; Query regions are from immunoBase, crohn's disease.

**Examples**

```
data(loci2path.demo)
```

# Index

- \* **cloud**
  - getWordcloud, [10](#)
- \* **datasets**
  - biocarta, [2](#)
  - eset.list, [4](#)
  - query.gr, [13](#)
- \* **heatmap**
  - getHeatmap, [6](#)
- \* **result**
  - getMat, [7](#)
  - getPval, [8](#)
  - getTissueDegree, [9](#)
- \* **word**
  - getWordcloud, [10](#)
- biocarta, [2](#)
- check.geneid, [3](#)
- coveredGene (loci2pathResult-class), [10](#)
- coveredGene, loci2pathResult-method (loci2pathResult-class), [10](#)
- description (geneSet-class), [5](#)
- description, geneSet-method (geneSet-class), [5](#)
- eqtlGene (eqtlSet-class), [3](#)
- eqtlGene, eqtlSet-method (eqtlSet-class), [3](#)
- eqtlId (eqtlSet-class), [3](#)
- eqtlId, eqtlSet-method (eqtlSet-class), [3](#)
- eqtlRange (eqtlSet-class), [3](#)
- eqtlRange, eqtlSet-method (eqtlSet-class), [3](#)
- eqtlSet (eqtlSet-class), [3](#)
- eqtlSet-class, [3](#)
- eset.list, [4](#)
- geneSet (geneSet-class), [5](#)
- geneSet-class, [5](#)
- geneSetList (geneSet-class), [5](#)
- geneSetList, geneSet-method (geneSet-class), [5](#)
- getHeatmap, [6](#)
- getHeatmap, loci2pathResult-method (getHeatmap), [6](#)
- getMat, [7](#)
- getMat, loci2pathResult-method (getMat), [7](#)
- getPathDescription, [7](#)
- getPathDescription, loci2pathResult-method (getPathDescription), [7](#)
- getPval, [8](#)
- getPval, loci2pathResult-method (getPval), [8](#)
- getTissueDegree, [9](#)
- getTissueDegree, loci2pathResult-method (getTissueDegree), [9](#)
- getWordcloud, [10](#)
- getWordcloud, loci2pathResult-method (getWordcloud), [10](#)
- loci2pathResult (loci2pathResult-class), [10](#)
- loci2pathResult-class, [10](#)
- numGene (geneSet-class), [5](#)
- numGene, geneSet-method (geneSet-class), [5](#)
- query, [11](#)
- query, eqtlSet, geneSet-method (query), [11](#)
- query, GenomicRanges, eqtlSet, geneSet-method (query), [11](#)
- query, GenomicRanges, list, ANY-method (query), [11](#)
- query, GenomicRanges, list, geneSet-method (query), [11](#)
- query, list, geneSet-method (query), [11](#)
- query, list-method (query), [11](#)
- query.gr, [13](#)
- resultTable (loci2pathResult-class), [10](#)
- resultTable, loci2pathResult-method (loci2pathResult-class), [10](#)
- tissue (eqtlSet-class), [3](#)
- tissue, eqtlSet-method (eqtlSet-class), [3](#)