

Package ‘xQTLbiolinks’

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Title Integrative Analysis of Quantitative Trait Locus Data of 'xQTL'

Version 1.2.2

Description User can query, download, and visualize of molecular quantitative trait locus and gene expression data from public resources through the application programming interface <<https://gtexportal.org/home/api-docs/index.html>> of 'GTEx'.

URL <https://github.com/dingruofan/xQTLbiolinks>

BugReports <https://github.com/dingruofan/xQTLbiolinks/issues>

License GPL (>= 3)

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Suggests colocal, knitr, rtracklayer, usethis

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apiAdmin_ping	<i>Heartbeat to check GTEEx API server connectivity.</i>
---------------	--

Description

test GTEEx API server and return download method.

Usage

```
apiAdmin_ping(fetchMethod = "")
```

Arguments

fetchMethod fetchMethod.

Value

A character string of fetchContent method.

apiEbi_ping	<i>Heartbeat to check EBI API server connectivity.</i>
-------------	--

Description

test EBI API server and return download method.

Usage

```
apiEbi_ping()
```

Value

A character string of fetchContent method.

dbsnpQueryRange	<i>retrieve snps from dbSNP using coordinate.</i>
-----------------	---

Description

retrieve snps from dbSNP using coordinate.

Usage

```
dbsnpQueryRange(
  chrom = "",
  startPos = -1,
  endPos = -1,
  genomeBuild = "GRCh38/hg38",
  track = "snp151Common"
)
```

Arguments

chrom	(character) name of chromosome, including chr1-chr22, chrX, chrY.
startPos	A positive integer.
endPos	A positive integer.
genomeBuild	"GRCh38/hg38" or "GRCh38/hg19". Default: "GRCh38/hg38".
track	"snp151Common", "snp150Common" or "snp147Common". Default: "snp151Common".

Value

A data.table object.

EBIquery_allTerm	<i>EBIquery_allTerm</i>
------------------	-------------------------

Description

EBIquery_allTerm

Usage

```
EBIquery_allTerm(term = "genes", termSize = 2000)
```

Arguments

term	"associations", "molecular_phenotypes", "studies", "tissues", "qtl_groups", "genes" or "chromosomes".
termSize	Number of fetched term.

Value

A data.table object.

Examples

```
# Fetch associations:
associations <- data.table::rbindlist(EBIquery_allTerm("associations", termSize=0))

# fetch molecular_phenotypes:
molecular_phenotypes <- EBIquery_allTerm("molecular_phenotypes", termSize=10)

# fetch studies:
studies <- EBIquery_allTerm("studies")

# fetch tissues:
tissues <- EBIquery_allTerm("tissues")

# fetch tissue-study mapping relationships
tissue_S <- EBIquery_allTerm( paste0("tissues/", "UBER_0002046", "/studies" ))

# fetch qtl groups:
qtl_groups <- EBIquery_allTerm("qtl_groups")

# Fetch genes:
geneList <- EBIquery_allTerm("genes", termSize=10)
```

extractGeneInfo	<i>extract gene info of specified genome from gencodeGeneInfoAllGranges</i>
-----------------	---

Description

extract gene info of specified genome from gencodeGeneInfoAllGranges

Usage

```
extractGeneInfo(gencodeGeneInfoAllGranges, genomeVersion = "v26")
```

Arguments

```
gencodeGeneInfoAllGranges
    from internal data
genomeVersion "v26" (default) or "v19"
```

Value

A data.table object.

Examples

```
gencodeGeneInfo <- extractGeneInfo(gencodeGeneInfoAllGranges)
```

fetchContent	<i>Fetch data using url by three methods</i>
--------------	--

Description

Fetch data using url by three methods

Usage

```
fetchContent(url1, method = "curl", downloadMethod = "auto", isJson = TRUE)
```

Arguments

url1	A url string.
method	Can be chosen from "download", "curl", "fromJSON", or "GET".
downloadMethod	The same methods from utils::download.file function.
isJson	Fetches content is a json file or not. Default: TRUE.

Value

A json object.

fetchContentEbi	<i>Fetch records from</i>
-----------------	---------------------------

Description

Fetch records from

Usage

```
fetchContentEbi(
  url1,
  method = "fromJSON",
  downloadMethod = "auto",
  termSize = 1000,
  termStart = 0
)
```

Arguments

url1	A url string.
method	Can be chosen from "download", "curl" or "GET".
downloadMethod	The same methods from utils::download.file function.
termSize	Number of records per request.
termStart	Start position per request.

Value

A data.table object.

is.wholenumber	<i>determine whether is a whole number:</i>
----------------	---

Description

determine whether is a whole number:

Usage

```
is.wholenumber(x, tol = .Machine$double.eps^0.5)
```

Arguments

x	A number
tol	Don't change

Value

TRUE or FALSE

retrieveLD	<i>Retrive SNP pairwise LD from locuscompare database.</i>
------------	--

Description

SNP pairwise ID are calculated based on 1000 Genomes Project Phase 3 version 5. For storage-efficiency, the output will only include SNPs with $r^2 > 0.2$ with the input SNP.

Usage

```
retrieveLD(chr, snp, population)
```

Arguments

chr	(string) Chromosome name. e.g. '22'. Notice that the name should not contain 'chr'.
snp	(string) SNP rsID.
population	(string) One of the 5 populations from 1000 Genomes: 'AFR', 'AMR', 'EAS', 'EUR', and 'SAS'.

Value

A data.frame object.

Examples

```
ld <- retrieveLD('6', 'rs9349379', 'AFR')
```

xQTLanalyze_coloc	<i>Conduct colocalization analysis with trait genes generated from xQTLanalyze_getTraits</i>
-------------------	--

Description

Conduct colocalization analysis with trait genes generated from xQTLanalyze_getTraits

Usage

```
xQTLanalyze_coloc(
  gwasDF,
  traitGene,
  geneType = "auto",
  genomeVersion = "grch38",
  tissueSiteDetail = "",
  study = "gtex_v8",
  mafThreshold = 0.01,
  population = "EUR",
  gwasSampleNum = 50000,
  method = "coloc",
  token = "9246d2db7917",
  bb.alg = FALSE
)
```

Arguments

gwasDF	A data.frame or data.table object of gwas.
traitGene	A gene symbol or a gencode id (versioned).
geneType	(character) options: "auto", "geneSymbol" or "gencodeId". Default: "auto".

genomeVersion	"grch38" (default) or "grch37". Note: grch37 will be converted to grch38 automatically.
tissueSiteDetail	(character) details of tissues in GTEx can be listed using tissueSiteDetailGTExv8 or tissueSiteDetailGTExv7
study	(character) name of studies can be listed using "ebi_study_tissues"
mafThreshold	Cutoff of maf to remove rare variants.
population	Supported population is consistent with the LDlink, which can be listed using function "LDlinkR::list_pop()"
gwasSampleNum	Sample number of GWAS dataset. Default:50000.
method	(character) options: "coloc"(default) or "hyprcoloc" (need a highe version).
token	LDlink provided user token, default = NULL, register for token at https://ldlink.nci.nih.gov/?tab=apiaccess
bb.alg	For hyprcoloc, branch and bound algorithm: TRUE, employ BB algorithm; FALSE, do not. Default: FALSE.

Value

A list of coloc result and details.

Examples

```
url1 <- "http://raw.githubusercontent.com/dingruofan/exampleData/master/gwasDFsub_MMP7.txt"
gwasDF <- data.table::fread(url1)
output <- xQTLanalyze_coloc(gwasDF = gwasDF, traitGene= "MMP7", tissueSiteDetail="Prostate")
```

xQTLanalyze_getSentinelSnp

Detect sentinel SNPs in a given summary statistis dataset.

Description

Return sentinel snps whose pValue < 5e-8(default) and SNP-to-SNP distance > 1e6 bp.

Usage

```
xQTLanalyze_getSentinelSnp(
  gwasDF,
  pValueThreshold = 5e-08,
  centerRange = 1e+06,
  mafThreshold = 0.01,
  genomeVersion = "grch38",
  grch37To38 = FALSE
)
```

Arguments

gwasDF	A data.frame or a data.table object. Five columns are required (arbitrary column names is supported): Col 1. "snps" (character), , using an rsID (e.g. "rs11966562"). Col 2. "chromosome" (character), one of the chromosome from chr1-chr22. Col 3. "position" (integer), genome position of snp. Col 4. "P-value" (numeric). Col 5. "MAF" (numeric). Allel frequency. Col 6. "beta" (numeric). effect size. Col 7. "se" (numeric). standard error.
pValueThreshold	Cutoff of gwas p-value. Default: 5e-8
centerRange	SNP-to-SNP distance. Default:1e6
mafThreshold	Cutoff of maf to remove rare variants.
genomeVersion	Genome version of input file. "grch37" or "grch38" (default).
grch37To38	TRUE or FALSE, we recommend converting grch37 to grch38, or using a input file of grch38 directly. Package rtracklayer is required.

Value

A data.table object.

Examples

```
url<-"http://raw.githubusercontent.com/dingruofan/exampleData/master/GLGC.txt"
gwasDF <- data.table::fread(url)
gwasDF <- gwasDF[, .(rsid, chr, position, P, maf, beta, se)]
sentinelSnpDF <- xQTLanalyze_getSentinelSnp(gwasDF)
```

xQTLanalyze_getTraits *Identify trait genes using sentinel SNPs generated from xQTLanalyze_getSentinelSnp*

Description

Identify trait genes using sentinel SNPs generated from xQTLanalyze_getSentinelSnp

Usage

```
xQTLanalyze_getTraits(
  sentinelSnpDF,
  detectRange = 1e+06,
  tissueSiteDetail = "",
  genomeVersion = "grch38",
  grch37To38 = FALSE
)
```

Arguments

- `sentinelSnpDF` A data.table. Better be the results from the function "xQTLanalyze_getSentinelSnp", seven columns are required, including "rsid", "chr", "position", "pValue", "maf", "beta" and "se".
- `detectRange` A integer value. Trait genes that harbor sentinel SNPs located in the 1kb range upstream and downstream of gene. Default: 1e6 bp
- `tissueSiteDetail` (character) details of tissues in GTEx can be listed using `tissueSiteDetailGTExv8` or `tissueSiteDetailGTExv7`
- `genomeVersion` "grch38" or "grch37". Default: "grch38"
- `grch37To38` TRUE or FALSE, we recommend converting grch37 to grch38, or using a input file of grch38 directly. Package `rtracklayer` is required.

Value

A data.table object

Examples

```
URL1<-"https://gitee.com/stronghoney/exampleData/raw/master/gwas/GLGC_CG0052/sentinelSnpDF.txt"
sentinelSnpDF <- data.table::fread(URL1)
traitsAll <- xQTLanalyze_getTraits(sentinelSnpDF,detectRange=1e4,"Brain - Cerebellum",
                                genomeVersion="grch37", grch37To38=TRUE)
```

xQTLanalyze_propensity

eQTL-specific analysis

Description

eQTL-specific analysis

Usage

```
xQTLanalyze_propensity(
  gene = "",
  geneType = "auto",
  variantName = "",
  variantType = "auto",
  tissueLabels = "",
  study = "",
  population = "EUR"
)
```

Arguments

gene	(character) gene symbol or gencode id (versioned or unversioned are both supported).
geneType	(character) options: "auto", "geneSymbol" or "gencodeId". Default: "auto".
variantName	(character) name of variant, dbsnp ID and variant id is supported, eg. "rs138420351" and "chr17_7796745_C_T_b38".
variantType	(character) options: "auto", "snpId" or "variantId". Default: "auto".
tissueLabels	(a character vector) can be listed with ebi_study_tissues. If is null, use all tissue / cell-types. (Default)
study	(character) Studies can be listed using ebi_study_tissues. If is null, use all studies (Default).
population	(string) One of the 5 populations from 1000 Genomes: 'AFR', 'AMR', 'EAS', 'EUR', and 'SAS'.

Value

A list containing four data.table objects, including: "snpLD" for LD details of the specified SNP; "assoAllLd" for eQTL details of LD-associated SNPs; "lm_R2_logP" for liner regression results; "cor_R2_logP" for correlation outputs;

Examples

```
propensityRes <- xQTLanalyze_propensity( gene="MMP7", variantName="rs11568818", study="TwinsUK")
xQTLvisual_qtlPropensity(propensityRes)
```

xQTLanalyze_TSExp *Perform tissue-specific expression analysis for genes.*

Description

Perform tissue-specific expression analysis for genes.

Usage

```
xQTLanalyze_TSExp(
  genes,
  geneType = "auto",
  method = "SPM",
  datasetId = "gtex_v8"
)
```

Arguments

genes	A character vector or a string of gene symbol, gencode id (versioned), or a character string of gene type.
geneType	(character) options: "auto", "geneSymbol" or "gencodeId". Default: "auto".
method	"SPM" or "entropy"
datasetId	(character) options: "gtex_v8" (default), "gtex_v7".

Value

A data.table object.

Examples

```
Tsgene <- xQTLanalyze_TSExp(extractGeneInfo(gencodeGeneInfoAllGranges)$gencodeId[1:5])
```

xQTLdownload_egene	<i>Download details of eGenes (eQTL Genes) for a specified gene or a tissue.</i>
--------------------	--

Description

eGenes are genes that have at least one significant cis-eQTL acting upon them. Results can be filtered by tissue.

Usage

```
xQTLdownload_egene(
  gene = "",
  geneType = "auto",
  datasetId = "gtex_v8",
  tissueSiteDetail = "",
  recordPerChunk = 2000
)
```

Arguments

gene	(character) gene symbol or gencode id (versioned or unversioned are both supported).
geneType	(character) options: "auto", "geneSymbol" or "gencodeId". Default: "auto".
datasetId	(character) options: "gtex_v8" (default), "gtex_v7".
tissueSiteDetail	(character) details of tissues in GTEx can be listed using tissueSiteDetailGTExv8 or tissueSiteDetailGTExv7
recordPerChunk	(integer) number of records fetched per request (default: 2000).

Value

A data.table object.

Examples

```
eGeneInfo <- xQTLdownload_egene("TP53")
eGeneInfo <- xQTLdownload_egene(tissueSiteDetail="Prostate", recordPerChunk=2000)
```

xQTLdownload_eqtl	<i>Download significant or insignificant eQTL associations of a tissue or across all tissues</i>
-------------------	--

Description

can be queried with a gene/variant-gene pair.

Usage

```
xQTLdownload_eqtl(
  variantName = "",
  gene = "",
  variantType = "auto",
  geneType = "auto",
  tissueSiteDetail = "",
  datasetId = "gtex_v8",
  recordPerChunk = 100
)
```

Arguments

variantName	(character) name of variant, dbsnp ID and variant id is supported, eg. "rs138420351" and "chr17_7796745_C_T_b38".
gene	(character) gene symbol or gencode id (versioned or unversioned are both supported). Can not be null.
variantType	(character) options: "auto", "snpId" or "variantId". Default: "auto".
geneType	(character) options: "auto", "geneSymbol" or "gencodeId". Default: "auto".
tissueSiteDetail	(character) details of tissues in GTEx can be listed using tissueSiteDetailGTExv8 or tissueSiteDetailGTExv7
datasetId	(character) options: "gtex_v8" (default), "gtex_v7".
recordPerChunk	(integer) number of records fetched per request (default: 100).

Value

A data.table object.

Examples

```
# Download eQTL info with a gene symbol:
eqtlInfo <- xQTLdownload_eqtl(gene="TP53")

# Use unversioned gencode ID in GTEx V8:
eqtl_v8 <- xQTLdownload_eqtl(gene="ENSG00000141510", datasetId="gtex_v8")

# In a specific tissue:
xQTLdownload_eqtl(gene="ENSG00000141510.16", geneType="gencodeId", tissueSiteDetail="Thyroid" )

# Download eQTL info with a variant-gene pair:
xQTLdownload_eqtl(variantName="rs1641513",gene="TP53", datasetId="gtex_v8")
xQTLdownload_eqtl(variantName="chr1_1667948_A_G_b38", gene="SLC35E2B",
                  tissueSiteDetail="Kidney - Cortex")
```

```
xQTLdownload_eqtlAllAsso
```

Download summary statistics of eQTL of a specified gene, variant, tissue or study.

Description

source of all eQTL associations is EBI eQTL category.

Usage

```
xQTLdownload_eqtlAllAsso(
  gene = "",
  geneType = "auto",
  variantName = "",
  variantType = "auto",
  tissueLabel = "",
  study = "gtex_v8",
  recordPerChunk = 1000,
  withB37VariantId = FALSE
)
```

Arguments

gene	(character) gene symbol or gencode id (versioned or unversioned are both supported).
geneType	(character) options: "auto","geneSymbol" or "gencodeId". Default: "auto".
variantName	(character) name of variant, dbsnp ID and variant id is supported, eg. "rs138420351" and "chr17_7796745_C_T_b38".
variantType	(character) options: "auto", "snpId" or "variantId". Default: "auto".

tissueLabel (character) all supported tissues can be listed using "ebi_study_tissues"
 study (character) name of studies can be listed using "ebi_study_tissues". If the study is null, use all studies (Default).
 recordPerChunk (integer) number of records fetched per request (default: 1000).
 withB37VariantId
 a logical value indicating whether to return the genome location(GTEEx v7) of variants. Default: FALSE.

Value

A data.table object.

Examples

```
# Download all associations of MLH1-rs13315355 pair in all tissues from all studies:
eqtlAsso <- xQTLdownload_eqtlAllAsso(gene="MLH1", variantName = "rs13315355", study="")

# Download associations of gene ATP11B in CD4+ T cell from all supported studies(time-consuming):
geneAsso <- xQTLdownload_eqtlAllAsso(gene="MMP7",tissueLabel = "CD4+ T cell", study="")

# Download associations of gene ATP11B in Muscle - Skeletal from GTEEx_V8:
geneAsso <- xQTLdownload_eqtlAllAsso("ATP11B", tissueLabel="Muscle - Skeletal")

# Download all associations of SNP rs11568818 in all tissues from all supported studies.
varAsso <- xQTLdownload_eqtlAllAsso(variantName="rs11568818", study="")
# Download associations of SNP rs11568818 in Muscle - Skeletal from GTEEx_V8:
varAsso <- xQTLdownload_eqtlAllAsso(variantName="chr11_102530930_T_C_b38",
                                     tissueLabel="Muscle - Skeletal")
```

xQTLdownload_eqtlAllAssoPos

Download summary statistics of eQTL with genome position.

Description

Download summary statistics of eQTL with genome position.

Usage

```
xQTLdownload_eqtlAllAssoPos(
  chrom = "",
  pos_lower = numeric(0),
  pos_upper = numeric(0),
  p_lower = 0,
  p_upper = 1.1,
  gene = "",
```

xQTLdownload_eqtlExp *Download normalized expression of gene for a eQTL pair.*

Description

Download normalized expression of gene for a eQTL pair.

Usage

```
xQTLdownload_eqtlExp(
  variantName = "",
  gene = "",
  variantType = "auto",
  geneType = "auto",
  tissueSiteDetail = "",
  datasetId = "gtex_v8"
)
```

Arguments

variantName	(character) name of variant, dbsnp ID and variant id is supported, eg. "rs138420351" and "chr17_7796745_C_T_b38".
gene	(character) gene symbol or gencode id (versioned or unversioned are both supported).
variantType	(character) options: "auto", "snpId" or "variantId". Default: "auto".
geneType	(character) options: "auto", "geneSymbol" or "gencodeId". Default: "auto".
tissueSiteDetail	(character) details of tissues in GTEx can be listed using tissueSiteDetailGTExv8 or tissueSiteDetailGTExv7
datasetId	(character) options: "gtex_v8" (default), "gtex_v7".

Value

A data.table object.

Examples

```
# Download exp with variant-gene pair in different tissues:
xQTLdownload_eqtlExp(variantName="rs1641513",gene="TP53", tissueSiteDetail="Liver")

# Download expression using variant ID and gencode ID.
xQTLdownload_eqtlExp(variantName="chr1_14677_G_A_b38",gene="ENSG00000228463.9",
  tissueSiteDetail="Stomach")
```

xQTLdownload_eqtlSig *Download significant eQTL associations of a specified tissue or across all tissues.*

Description

Download significant eQTL associations of a specified tissue or across all tissues.

Usage

```
xQTLdownload_eqtlSig(
  variantName = "",
  genes = "",
  variantType = "auto",
  geneType = "auto",
  tissueSiteDetail = "",
  datasetId = "gtex_v8"
)
```

Arguments

variantName	(character) name of variant, dbsnp ID and variant id is supported, eg. "rs138420351" and "chr17_7796745_C_T_b38".
genes	(character string or a character vector) gene symbols or gencode ids (versioned or unversioned are both supported).
variantType	(character) options: "auto", "snpId" or "variantId". Default: "auto".
geneType	(character) options: "auto", "geneSymbol" or "gencodeId". Default: "auto".
tissueSiteDetail	(character) details of tissues in GTEx can be listed using tissueSiteDetailGTExv8 or tissueSiteDetailGTExv7
datasetId	(character) options: "gtex_v8" (default), "gtex_v7".

Value

A data.table object.

Examples

```
# Download eQTL info for a variant:
xQTLdownload_eqtlSig("rs201327123")
xQTLdownload_eqtlSig("chr1_14677_G_A_b38")
xQTLdownload_eqtlSig("11_66328719_T_C_b37", datasetId="gtex_v7")
xQTLdownload_eqtlSig("11_66328719_T_C_b37", datasetId="gtex_v7",
  tissueSiteDetail="Skin - Sun Exposed (Lower leg)")

# Download eQTL association according to all tissues with genome location:
```

```

varInfo <- xQTLquery_varPos(chrom="chr1", pos=c(1102708),"gtex_v8")
xQTLdownload_eqtlSig(variantName=varInfo$snpId)

# Download eQTL info for gene:
xQTLdownload_eqtlSig(genes="ATAD3B")
xQTLdownload_eqtlSig(genes=c("TP53", "SLC35E2B"), tissueSiteDetail= "Brain - Cerebellum")
xQTLdownload_eqtlSig(genes="ENSG00000141510.16", datasetId="gtex_v8")

# Download eQTL info for a variant-gene pair:
xQTLdownload_eqtlSig(variantName="rs1641513", genes="TP53", datasetId="gtex_v8")
xQTLdownload_eqtlSig(variantName="rs1641513", genes="TP53", datasetId="gtex_v7")
xQTLdownload_eqtlSig(variantName="chr1_1667948_A_G_b38",
                     genes="SLC35E2B", tissueSiteDetail="Kidney - Cortex")

```

xQTLdownload_exp	<i>Download normalized gene expression at the sample level in a specified tissue.</i>
------------------	---

Description

Download normalized gene expression at the sample level in a specified tissue.

Usage

```

xQTLdownload_exp(
  genes = "",
  geneType = "auto",
  tissueSiteDetail = "Liver",
  datasetId = "gtex_v8",
  toSummarizedExperiment = TRUE,
  recordPerChunk = 80,
  pathologyNotesCategories = FALSE
)

```

Arguments

genes	(character string or a character vector) gene symbols or gencode ids (versioned or unversioned are both supported).
geneType	(character) options: "auto", "geneSymbol" or "gencodeId". Default: "auto".
tissueSiteDetail	(character) details of tissues in GTEx can be listed using <code>tissueSiteDetailGTExv8</code> or <code>tissueSiteDetailGTExv7</code>
datasetId	(character) options: "gtex_v8" (default), "gtex_v7".
toSummarizedExperiment	a logical value indicating whether to return a data.frame or a summarizedExperiment object. Default: TRUE, return a toSummarizedExperiment object.

recordPerChunk (integer) number of records fetched per request (default: 80).
 pathologyNotesCategories
 a logical value indicating whether to return pathologyNotes. Default: FALSE,
 the pathologyNotes is ignored.

Value

return a SummarizedExperiment or a data.table object harboring gene expression profiles and samples' information.

Examples

```
# Download gene expression with a genecode ID:
expProfiles <- xQTLdownload_exp("ENSG00000210195.2", tissueSiteDetail="Liver")

# extract expression profile from SummarizedExperiment object:
expDT <- SummarizedExperiment::assay(expProfiles)

# extract samples' detail from SummarizedExperiment object:
sampleDT <- SummarizedExperiment::colData(expProfiles)

# Download gene expression profiles of multiple genes:
expProfiles <- xQTLdownload_exp(c("tp53", "naDK", "SDF4"),
                               tissueSiteDetail="Artery - Coronary",
                               pathologyNotesCategories=TRUE,
                               toSummarizedExperiment=FALSE)

# Download with versioned and unversioned genecode Id.
expProfiles <- xQTLdownload_exp(c("ENSG00000141510.16", "ENSG00000008130.15", "ENSG00000078808"),
                               tissueSiteDetail="Artery - Coronary",
                               toSummarizedExperiment=FALSE)
```

xQTLdownload_geneMedExp

Download median expression of all samples for specified genes across tissues.

Description

Download median expression of all samples for specified genes across tissues.

Usage

```
xQTLdownload_geneMedExp(
  genes = "",
  geneType = "auto",
  datasetId = "gtex_v8",
```

```
tissueSiteDetail = "",
recordPerChunk = 150
)
```

Arguments

genes (character string or a character vector) gene symbols or gencode ids (versioned or unversioned are both supported).

geneType (character) options: "auto", "geneSymbol" or "gencodeId". Default: "auto".

datasetId (character) options: "gtex_v8" (default), "gtex_v7".

tissueSiteDetail (character) details of tissues in GTEx can be listed using `tissueSiteDetailGTExv8` or `tissueSiteDetailGTExv7`

recordPerChunk (integer) number of records fetched per request (default: 150).

Value

A data.table object.

Examples

```
geneMedExp <- xQTLdownload_geneMedExp(genes="LYNX1")
geneMedExp <- xQTLdownload_geneMedExp(genes=c("TP53", "IRF5"))
```

xQTLdownload_sgene	<i>Download details of sGenes (sQTL Genes) for a specified gene or a tissue.</i>
--------------------	--

Description

sGenes are genes that have at least one significant sQTL acting upon them. Results may be filtered by tissue.

Usage

```
xQTLdownload_sgene(
  gene = "",
  geneType = "auto",
  datasetId = "gtex_v8",
  tissueSiteDetail = "",
  recordPerChunk = 2000
)
```

Arguments

gene (character) gene symbol or gencode id (versioned or unversioned are both supported). Can be null.

geneType (character) options: "auto", "geneSymbol" or "gencodeId". Default: "auto".

datasetId (character) options: "gtex_v8" (default), "gtex_v7".

tissueSiteDetail (character) details of tissues in GTEx can be listed using `tissueSiteDetailGTExv8` or `tissueSiteDetailGTExv7`

recordPerChunk (integer) number of records fetched per request (default: 2000).

Value

A data.table object.

Examples

```
sGeneInfo <- xQTLdownload_sgene(tissueSiteDetail="Liver")
sGeneInfo <- xQTLdownload_sgene(gene="DDX11", tissueSiteDetail="Liver" )
```

xQTLdownload_sqtlExp *Download normalized expression of intron for a sQTL pair.*

Description

Download normalized expression of intron for a sQTL pair.

Usage

```
xQTLdownload_sqtlExp(
  variantName = "",
  phenotypeId = "",
  variantType = "auto",
  tissueSiteDetail = "",
  datasetId = "gtex_v8"
)
```

Arguments

variantName (character) name of variant, dbsnp ID and variant id is supported, eg. "rs138420351" and "chr17_7796745_C_T_b38".

phenotypeId A character string. Format like: "chr1:497299:498399:clu_54863:ENSG00000239906.1"

variantType (character) options: "auto", "snpId" or "variantId". Default: "auto".

tissueSiteDetail (character) details of tissues in GTEx can be listed using `tissueSiteDetailGTExv8` or `tissueSiteDetailGTExv7`

datasetId (character) options: "gtex_v8" (default), "gtex_v7".

Value

A data.table object.

Examples

```
# Download sQTL expression in different tissues:
xQTLdownload_sqtlExp(variantName="rs1450891501",
                    phenotypeId="chr1:497299:498399:clu_54863:ENSG00000239906.1",
                    tissueSiteDetail="Lung")

# Download sQTL expression using variant ID.
xQTLdownload_sqtlExp(variantName="chr1_1259424_T_C_b38",
                    phenotypeId="chr1:1487914:1489204:clu_52051:ENSG00000160072.19",
                    tissueSiteDetail="Adipose - Subcutaneous")
```

xQTLdownload_sqtlSig *Download significant sQTL associations of a tissue or across all tissues*

Description

Only GTEx v8 is supported.

Usage

```
xQTLdownload_sqtlSig(
  variantName = "",
  genes = "",
  variantType = "auto",
  geneType = "auto",
  tissueSiteDetail = ""
)
```

Arguments

variantName (character) name of variant, dbsnp ID and variant id is supported, eg. "rs138420351" and "chr17_7796745_C_T_b38".

genes (character string or a character vector) gene symbol or gencode id (versioned or unversioned are both supported).

variantType (character) options: "auto", "snpId" or "variantId". Default: "auto".

geneType (character) options: "auto", "geneSymbol" or "gencodeId". Default: "auto".

tissueSiteDetail (character) details of tissues in GTEx can be listed using tissueSiteDetailGTExv8 or tissueSiteDetailGTExv7

Value

A data.table object.

Examples

```
# Download sQTL details with rsid:
xQTLdownload_sqtlSig(variantName="rs201327123")
xQTLdownload_sqtlSig(variantName="chr1_14677_G_A_b38", tissueSiteDetail="Whole Blood")

# Download sQTL details with gene symbol and gencode ID:
xQTLdownload_sqtlSig(genes="ENSG00000141510.16", tissueSiteDetail="Lung" )
xQTLdownload_sqtlSig(genes=c("ATAD3B", "MLH1"))

# Download sQTL details with the variant-genes pair:
xQTLdownload_sqtlSig(variantName="rs201327123", genes=c("WASH7P", "RP11-206L10.2"))
xQTLdownload_sqtlSig(variantName="chr17_7465085_A_G_b38", genes="TP53", tissueSiteDetail="Lung")
```

xQTLquery_gene	<i>Query basic information (including name, symbol, position and description, etc.) of genes.</i>
----------------	--

Description

Query basic information (including name, symbol, position and description, etc.) of genes.

Usage

```
xQTLquery_gene(
  genes = "",
  geneType = "auto",
  gencodeVersion = "v26",
  recordPerChunk = 150
)
```

Arguments

genes A character vector or a string of gene symbol, gencode id (versioned or unversioned), or a character string of gene type.

- **gene symbol (Default).**
A character string or a character vector (case ignored). like: "tp53", "naDK", "SDF4".
- **gencode/ensemble id** (versioned or unversioned).
A character string or a character vector (case ignored). like: "ENSG00000210195.2", "ENSG000000007".
- **gene classification.**
when "geneType" is "geneCategory", supported "genes" can be listed using function `gencodeGenetype$V26` or `gencodeGenetype$V19`

geneType (character) options: "auto", "geneSymbol" or "gencodeId". Default: "auto".

gencodeVersion (character) options: "v26"(default, matched with `gtex_v8`) or "v19"

recordPerChunk (integer) number of records fetched per request (default: 150).

Value

A data.table object of queried gene information. including following columns:

- **genes.** Input genes
- **geneSymbol.** Gene symbol.
- **gencodeId.** Gencode/ensemble id (versioned).
- **entrezGeneId.** Entrez gene ID.
- **geneType.** Gene type.
- **chromosome.** Note: "chr" is added in gencode v26,
- **start.**
- **end.**
- **strand.**
- **tss.** Transcriptional start site.
- **gencodeVersion.** Gencode Version.
- **genomeBuild.** Genome version.
- **description.**

Examples

```
# query gene of gencode version v26/hg38
geneInfo <- xQTLquery_gene("TP53")
geneInfo <- xQTLquery_gene(c("tp53", "naDK", "SDF4") )
geneInfo <- xQTLquery_gene(c("ENSG00000210195.2", "ENSG0000078808"))
```

xQTLquery_geneAll *Fetch details of all genes supported in GTEx.*

Description

Fetch details of all genes supported in GTEx.

Usage

```
xQTLquery_geneAll(gencodeVersion = "v26", recordPerChunk = 2000)
```

Arguments

gencodeVersion (character) options: "v26"(default, matched with gtex_v8) or "v19"
recordPerChunk (integer) number of records fetched per request (default: 2000).

Value

A data.table object of all genes' information.

`xQTLquery_sampleBySampleId`*Query sample's details with samples' IDs.*

Description

Query sample's details with samples' IDs.

Usage

```
xQTLquery_sampleBySampleId(  
  sampleIds,  
  recordPerChunk = 150,  
  pathologyNotesCategories = FALSE  
)
```

Arguments

`sampleIds` A character vector or a string of sample ID.
`recordPerChunk` (integer) number of records fetched per request (default: 200).
`pathologyNotesCategories`
Default: pathologyNotes info is ignored.

Value

a data.table object of samples' information.

Examples

```
sampleIds <- c("GTEX-11NUK-0011-R4a-SM-D012B", "GTEX-110NC-0011-R4b-SM-D093H",  
             "GTEX-11DXY-0526-SM-5EGGQ", "GTEX-130VJ-1026-SM-5IFGI")  
sampleInfo <- xQTLquery_sampleBySampleId(sampleIds)
```

`xQTLquery_sampleByTissue`*Query sample's details with tissue name*

Description

Query sample's details with tissue name

Usage

```
xQTLquery_sampleByTissue(
  tissueSiteDetail = "Liver",
  dataType = "RNASEQ",
  datasetId = "gtex_v8",
  recordPerChunk = 200,
  pathologyNotesCategories = FALSE
)
```

Arguments

tissueSiteDetail (character) details of tissues in GTEx can be listed using `tissueSiteDetailGTExv8` or `tissueSiteDetailGTExv7`

dataType A character string. Options: "RNASEQ" (default), "WGS", "WES", "OMNI".

datasetId (character) options: "gtex_v8" (default), "gtex_v7".

recordPerChunk (integer) number of records fetched per request (default: 200).

pathologyNotesCategories Default: pathologyNotes info is ignored.

Value

return a data.table object of samples' information

Examples

```
sampleInfo <- xQTLquery_sampleByTissue("Brain - Amygdala" )
sampleInfo <- xQTLquery_sampleByTissue(tissueSiteDetail="Liver", pathologyNotesCategories=TRUE)
```

xQTLquery_tissue *Fetch all details of a specified tissue or all tissues*

Description

Information includes tissue IDs, number of RNA-Seq samples, number of RNA-Seq samples with genotype, number of expressed genes, number of eGenes. Also includes tissueSiteDetail ID, name, abbreviation, iberon ID, and standard tissue colors. TissueSiteDetails are grouped by TissueSites. By default, this service reports from the latest GTEx release.

Usage

```
xQTLquery_tissue(tissueName = "", datasetId = "gtex_v8")
```

Arguments

tissueName Tissue name, tissue ID or tissue site name. Default return all tissues' information. Can be choose from `tissueSiteDetailGTExv8` or `tissueSiteDetailGTExv7`

datasetId (character) options: "gtex_v8" (default), "gtex_v7".

Value

A data.table object.

Examples

```
tissueAll <- xQTLquery_tissue(datasetId="gtex_v7")
BrainInfo <- xQTLquery_tissue("Brain", datasetId="gtex_v7")
```

xQTLquery_varId	<i>Query variant in GTEx with variant ID or dbSNP ID</i>
-----------------	--

Description

Query variant in GTEx with variant ID or dbSNP ID

Usage

```
xQTLquery_varId(variantName = "", variantType = "auto", datasetId = "gtex_v8")
```

Arguments

variantName (character) name of variant, dbsnp ID and variant id is supported, eg. "rs138420351" and "chr17_7796745_C_T_b38".

variantType (character) options: "auto", "snpId" or "variantId". Default: "auto".

datasetId (character) options: "gtex_v8" (default), "gtex_v7".

Value

A data.table object.

Examples

```
xQTLquery_varId("rs12596338")
xQTLquery_varId("rs12596338", datasetId="gtex_v7")
xQTLquery_varId("chr11_66561248_T_C_b38")
xQTLquery_varId("11_66328719_T_C_b37", variantType="variantId", datasetId="gtex_v7")
```

xQTLquery_varPos	<i>Query variants in GTEx using genome position.</i>
------------------	--

Description

Query variants in GTEx using genome position.

Usage

```
xQTLquery_varPos(
  chrom = "",
  pos = numeric(0),
  datasetId = "gtex_v8",
  recordPerChunk = 200
)
```

Arguments

chrom (character) name of chromosome, including chr1-chr22, chrX, chrY.
 pos An integer array.
 datasetId (character) options: "gtex_v8" (default), "gtex_v7".
 recordPerChunk (integer) number of records fetched per request (default: 200).

Value

A data.table object.

Examples

```
xQTLquery_varPos(chrom="chr1", pos=c(1102708,1105739),"gtex_v8")
xQTLquery_varPos(chrom="1", pos=c(1038088,1041119),"gtex_v7")
xQTLquery_varPos("1", c(1246438, 1211944, 1148100),"gtex_v7")
```

xQTLvisual_eqtl	<i>Box plot with jittered points for showing number and significance of eQTL associations</i>
-----------------	---

Description

Box plot with jittered points for showing number and significance of eQTL associations

Usage

```
xQTLvisual_eqtl(gene, geneType = "auto", datasetId = "gtex_v8")
```

Arguments

gene	(character) gene symbol or gencode id (versioned or unversioned are both supported).
geneType	(character) options: "auto", "geneSymbol" or "gencodeId". Default: "auto".
datasetId	(character) options: "gtex_v8" (default), "gtex_v7".

Value

A ggplot object.

Examples

```
xQTLvisual_eqtl("KIF15")
```

xQTLvisual_eqtlExp *Boxplot of normalized expression among genotypes for eQTL.*

Description

Boxplot of normalized expression among genotypes for eQTL.

Usage

```
xQTLvisual_eqtlExp(
  variantName = "",
  gene = "",
  variantType = "auto",
  geneType = "auto",
  tissueSiteDetail = "",
  datasetId = "gtex_v8"
)
```

Arguments

variantName	(character) name of variant, dbsnp ID and variant id is supported, eg. "rs138420351" and "chr17_7796745_C_T_b38".
gene	(character) gene symbol or gencode id (versioned or unversioned are both supported).
variantType	(character) options: "auto", "snpId" or "variantId". Default: "auto".
geneType	(character) options: "auto", "geneSymbol" or "gencodeId". Default: "auto".
tissueSiteDetail	(character) details of tissues in GTEx can be listed using tissueSiteDetailGTExv8 or tissueSiteDetailGTExv7
datasetId	(character) options: "gtex_v8" (default), "gtex_v7".

Value

A list containing eQTL detail, expression profile and a ggplot object.

Examples

```
expEqtl<-xQTLvisual_eqtlExp(variantName="rs3778754",gene ="IRF5",tissueSiteDetail="Whole Blood")
```

```
xQTLvisual_geneExpTissues
```

Violin plot of distribution of the gene expression profiles among multiple tissues.

Description

Violin plot of distribution of the gene expression profiles among multiple tissues.

Usage

```
xQTLvisual_geneExpTissues(  
  gene = "",  
  geneType = "auto",  
  tissues = "All",  
  datasetId = "gtex_v8",  
  log10y = FALSE,  
  toTissueSite = FALSE  
)
```

Arguments

gene	(character) gene symbol or gencode id (versioned or unversioned are both supported).
geneType	(character) options: "auto", "geneSymbol" or "gencodeId". Default: "auto".
tissues	A character string or a vector. "All" (default) means that all tissues is included.
datasetId	(character) options: "gtex_v8" (default), "gtex_v7".
log10y	Display values of expression in log scale. Default: FALSE.
toTissueSite	TRUE or FALSE, display all subtissues or tissue Site. Default: TURE.

Value

A list containing expression profile and a ggplot object.

Examples

```
# Display gene expression in specified tissues.
geneExpTissues <- xQTLvisual_geneExpTissues("TP53", tissues=c("Lung", "Brain", "Ovary"))
```

xQTLvisual_genesExp *Density plot of expression profiles of the gene*

Description

Density plot of expression profiles of the gene

Usage

```
xQTLvisual_genesExp(
  genes,
  geneType = "auto",
  tissueSiteDetail = "",
  datasetId = "gtex_v8"
)
```

Arguments

genes	(character string or a character vector) gene symbol or gencode id (versioned or unversioned are both supported).
geneType	(character) options: "auto", "geneSymbol" or "gencodeId". Default: "auto".
tissueSiteDetail	(character) details of tissues in GTEx can be listed using tissueSiteDetailGTExv8 or tissueSiteDetailGTExv7
datasetId	(character) options: "gtex_v8" (default), "gtex_v7".

Value

A ggplot object.

Examples

```
genes <- c("FNDC8", "S100Z", "AQP6", "AMOT", "C3orf38", "FOXL1", "COX11",
          "FCN3", "DDX58", "CFI", "MS4A18", "NUDT13", "HOXA4", "VSX1")
xQTLvisual_genesExp(genes, tissueSiteDetail="Lung")

genes <- c("ENSG00000073598.5", "ENSG00000171643.13", "ENSG00000086159.12", "ENSG00000126016.15",
          "ENSG00000179021.9", "ENSG00000176678.5", "ENSG00000166260.10", "ENSG00000142748.12",
          "ENSG00000107201.9", "ENSG00000205403.12", "ENSG00000214782.7", "ENSG00000166321.13",
          "ENSG00000197576.13", "ENSG00000100987.14")
xQTLvisual_genesExp(genes, geneType="gencodeId", tissueSiteDetail="Liver")
```

xQTLvisual_locusCombine

Generate a combined figure including locuszoom and locuscompare plot object.

Description

This function is rebuilt from locuscompare.R (<https://github.com/boxiangliu/locuscomparer/blob/master/R/locuscompare.R>).

Usage

```
xQTLvisual_locusCombine(
  gwasEqtldata,
  posRange = "",
  population = "EUR",
  highlightSnp = "",
  legend_position = "bottomright",
  snpLD = NULL
)
```

Arguments

gwasEqtldata	A data.frame or a data.table that including signals from both GWAS and eQTL. Five columns are required (arbitrary column names is supported): Col 1. "snps" (character), using an rsID (e.g. "rs11966562"). Col 2. "chromosome" (character), one of the chromosome from chr1-chr22. Col 3. "postion" (integer), genome position of snp. Col 4. "P-value" (numeric) of GWAS signals. Col 5. "P-value" (numeric) of eQTL signals.
posRange	Genome range that you want to visualize (e.g. "chr6:3e7-7e7"). Default is the region that covers all snps.
population	One of the 5 popuations from 1000 Genomes: 'AFR', 'AMR', 'EAS', 'EUR', and 'SAS'.
highlightSnp	Default is the snp that with lowest p-value.
legend_position	(string, optional) Either 'bottomright', 'topright', or 'topleft'. Default: 'bottom-right'.
snpLD	A data.frame object of LD matrix. Default is null.

Value

A ggplot object.

Examples

```
# load data:
u1 <- "http://raw.githubusercontent.com/dingruofan/exampleData/master/gwas/AD/gwasEqtldata.txt"
gwasEqtldata <- data.table::fread(u1)
xQTLvisual_locusCombine(gwasEqtldata, highlightSnp="rs13120565")
```

```
xQTLvisual_locusCompare
```

Dotplot of comparing regional signals between GWAS and xQTL.

Description

This function is rebuilt from locuscompare.R (<https://github.com/boxiangliu/locuscomparer/blob/master/R/locuscompare.R>).

Usage

```
xQTLvisual_locusCompare(
  eqtlDF,
  gwasDF,
  highlightSnp = "",
  population = "EUR",
  legend = TRUE,
  legend_position = c("topright", "bottomright", "topleft"),
  snpLD = NULL
)
```

Arguments

eqtlDF	A data.frame or data.table with two columns: dbSNP id and p-value.
gwasDF	A data.frame or data.table with two columns: dbSNP id and p-value.
highlightSnp	Default is the snp that is farthest from the origin of the coordinates.
population	One of the 5 populations from 1000 Genomes: 'AFR', 'AMR', 'EAS', 'EUR', and 'SAS'.# @param token LDlink provided user token, default = NULL, register for token at https://ldlink.nci.nih.gov/?tab=apiaccess
legend	(boolean, optional) Should the legend be shown? Default: TRUE.
legend_position	(string, optional) Either 'bottomright', 'topright', or 'topleft'. Default: 'bottomright'.
snpLD	A data.frame object of LD matrix. Default is null.

Value

A ggplot object.

Examples

```
library(data.table)
# load data:
eqtlDF <- fread("https://gitee.com/stronghoney/exampleData/raw/master/eqtl/eqtlAsso1.txt")
gwasDF <- fread("https://gitee.com/stronghoney/exampleData/raw/master/gwas/AD/gwasChr6Sub3.txt")
# visualize:
xQTLvisual_locusCompare( eqtlDF, gwasDF, legend_position="topleft")
```

xQTLvisual_locusZoom *Locuszoom plot for visualizing regional signals relative to genomic position with a file of summary statistics*

Description

This function is rebuilt from locuscompare.R (<https://github.com/boxiangliu/locuscomparer/blob/master/R/locuscompare.R>).

Usage

```
xQTLvisual_locusZoom(
  DF,
  highlightSnp = "",
  population = "EUR",
  posRange = "",
  legend = TRUE,
  legend_position = c("topright", "bottomright", "topleft"),
  snpLD = NULL
)
```

Arguments

DF	A data.frame or a data.table object. Four columns are required (arbitrary column names is supported): Col 1. "snps" (character), , using an rsID (e.g. "rs11966562"); Col 2. "chromosome" (character), one of the chromosome from chr1-chr22; Col 3. "postion" (integer), genome position of snp. Col 4. "P-value" (numeric).
highlightSnp	Default is the snp that with lowest p-value.
population	One of the 5 popuations from 1000 Genomes: 'AFR', 'AMR', 'EAS', 'EUR', and 'SAS'.
posRange	Genome range that you want to visualize (e.g. "chr6:3e7-7e7"). Default is the region that covers all snps.
legend	(boolean, optional) Should the legend be shown? Default: TRUE.
legend_position	(string, optional) Either 'bottomright', 'topright', or 'topleft'. Default: 'bottom-right'.
snpLD	A data.frame of LD matirx. Default is null.

Value

A list containing data.table and ggplot object.

Examples

```
library(data.table)
# For GWAS dataset:
gwasDF <- fread("https://gitee.com/stronghoney/exampleData/raw/master/gwasChr6Sub4.txt")
xQTLvisual_locusZoom(gwasDF)
# Zoom in:
xQTLvisual_locusZoom(gwasDF, posRange="chr6:4.7e7-4.8e7", population = "EUR")

# For eQTL of a gene of interest (time-consuming):
eqtlAsso <- xQTLdownload_eqtlAllAsso("RP11-385F7.1", tissueLabel = "Brain - Cortex",
                                     withB37VariantId=FALSE)
xQTLvisual_locusZoom(eqtlAsso[,c("snpId", "chrom", "pos", "pValue")], highlightSnp="rs4711878")
# Zoom in:
xQTLvisual_locusZoom(eqtlAsso[,c("snpId", "chrom", "pos", "pValue")], highlightSnp="rs4711878",
                     posRange="chr6:47.3e6-47.9e6")
```

xQTLvisual_qlPropensity

Visualization of QTL specificity among multiple cells/tissues.

Description

Visualization of QTL specificity among multiple cells/tissues.

Usage

```
xQTLvisual_qlPropensity(propensityRes, P_cutoff = 1)
```

Arguments

propensityRes A data.table object from the function xQTLanalyze_propensity
P_cutoff (numeric) cutoff of the p-value of tissue propensity. Default: 1

Value

A ggplot object

Examples

```
propensityRes <- xQTLanalyze_propensity( gene="MMP7", variantName="rs11568818", study="TwinsUK")
xQTLvisual_qlPropensity(propensityRes)
```

xQTLvisual_sqt1Exp *Boxplot of normalized expression among genotypes for sQTL.*

Description

Boxplot of normalized expression among genotypes for sQTL.

Usage

```
xQTLvisual_sqt1Exp(
  variantName = "",
  phenotypeId = "",
  variantType = "auto",
  tissueSiteDetail = "",
  datasetId = "gtex_v8"
)
```

Arguments

variantName (character) name of variant, dbsnp ID and variant id is supported, eg. "rs138420351" and "chr17_7796745_C_T_b38".

phenotypeId A character string. Format like: "chr1:497299:498399:clu_54863:ENSG00000239906.1"

variantType (character) options: "auto", "snpId" or "variantId". Default: "auto".

tissueSiteDetail (character) details of tissues in GTEx can be listed using `tissueSiteDetailGTExv8` or `tissueSiteDetailGTExv7`

datasetId (character) options: "gtex_v8" (default), "gtex_v7".

Value

A list containing variant detail, expression profile and a ggplot object.

Examples

```
expSqt1 <-xQTLvisual_sqt1Exp(variantName="chr11_66561248_T_C_b38",
  phenotypeId ="chr11:66348070:66353455:clu_8500:ENSG00000255468.6",
  tissueSiteDetail="Skin - Sun Exposed (Lower leg)")
```

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