Package ‘xQTLbiolinks’

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Title An R Package for Integrative Analysis of Quantitative Trait Locus Data of ‘xQTL’

Version 1.6.2

Description Enables users-customized data retrieval, processing, analysis, and data visualization 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’ and <http://www.ebi.ac.uk/eqtl/api> of ‘eQTL catalogue’.

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

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

License GPL (>= 3)

Encoding UTF-8

LazyData true

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Suggests VariantAnnotation, PupillometryR, coloc, knitr, rtracklayer, usethis, ggridges, CMplot, R.utils

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EBIquery_allTerm

Query supported terms (phenotypes, studies, tissues) in eQTL catalogue

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)

---

eventGeneInfo | Extract gene details from gencodeGeneInfoAllGranges object

**Description**

Extract gene details from gencodeGeneInfoAllGranges object

**Usage**

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

**Arguments**

- `gencodeGeneInfoAllGranges`:
  - from internal data

- `genomeVersion`:
  - "v26" (default) or "v19"

**Value**

A data.table object.

**Examples**

```
gencodeGeneInfo <- eventGeneInfo(gencodeGeneInfoAllGranges)
```

---

retrievelD | Retrieve SNP pairwise LD from locuscompare database

**Description**

SNP pairwise LD 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

```r
d <- retrieveLD('6', 'rs9349379', 'AFR')
```

Description

Retrieve SNP pairwise LD from LDlink database

Usage

```r
retrieveLD_LDproxy(
  targetSnp = "", 
  population = "EUR", 
  windowSize = 50000, 
  method = "download", 
  genomeVersion = "grch38", 
  max_count = 3, 
  token = "9246d2db7917"
)
```

Arguments

- **targetSnp** target SNP, support dbSNP IP.
- **population** Supported population is consistent with the LDlink, which can be listed using function LDlinkR::list_pop()
- **windowSize** Window around the highlighted snp for querying linkage disequilibrium information. Default:500000
- **method** The same as fetchContent function, can be chosen from "download", "curl", "GetWithHeader".
- **genomeVersion** "grch38"(default) or "grch37".
- **max_count** To prevent download failure due to network fluctuations, max number of connection attempts.
- **token** Ldlink token, default: "9246d2db7917"
Value
A data.table object.

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" (must be updated to the latest version from the github to use hyprcoloc). Package coloc or hyprcoloc is required.

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://bioinfo.szbl.ac.cn/xQTL_biolinks/xqtl_data/gwasDFsub_MMP7.txt"
gwasDF <- data.table::fread(url1)
output <- xQTLanalyze_coloc(gwasDF = gwasDF, traitGene= "MMP7", tissueSiteDetail="Prostate")
xQTLanalyze_getSentinelSnp

Detect sentinel SNPs for GWAS using summary statistics data

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. "MAP" (numeric). Allel frequency.
  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.
geneVersion   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

```r
url<"http://bioinfo.szbl.ac.cn/xQTL_biolinks/xqtl_data/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

```r
xQTLanalyze_getTraits(
  sentinelSnpDF, 
  detectRange = 1e+06, 
  tissueSiteDetail = "", 
  genomeVersion = "grch38", 
  grch37To38 = FALSE, 
  overlapWithEGene = TRUE, 
  egeneDF = NULL 
)
```

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.

overlapWithEGene TRUE(default) of FALSE. take the intersection with eGenes, egene data.frame will be automatically download from GTEx, or can be provided by the parameter egeneDF specified the data.frame of one column of genecode ID. Default:TRUE

egeneDF A data.table object of one column of gencode ID. requiring overlapWithEGene is TRUE.

Value
A data.table object

Examples

# without a customized egene file,
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)

# with a egene file:
egeneFile <- "http://bioinfo.szbl.ac.cn/xQTL_biolinks/xqtl_data/egeneDF.txt"
egeneDF <- data.table::fread(egeneFile)
traitsAll <- xQTLanalyze_getTraits(sentinelSnpDF, detectRange=1e4,"Brain - Cerebellum",
genomeVersion="grch37", grch37To38=TRUE, egeneDF=egeneDF)

xQTLanno_calLambda calculate genomic control inflation factor for a QTL/GWAS summary statistics dataset.

Description
calculate genomic control inflation factor for a QTL/GWAS summary statistics dataset.

Usage
xQTLanno_calLambda(summaryDT)
Arguments

**summaryDT**  
A data.frame containing one or two columns: p-value (required) and group (optional)

Value

A data.table object

Examples

```r
url1 <- "http://bioinfo.szbl.ac.cn/xQTL_biolinks/xqtl_data/eqtl/MMP7_qtlDF.txt"
qt1 <- data.table::fread(url1, sep="\t")

# calculate lambda value with all variants
xQTLanno_calLambda(qt1[,.(pValue)])

# calculate lambda value for each group:
qt1$groups <- sample(c(0,1), size = nrow(qt1), replace = TRUE)
xQTLanno_calLambda(qt1[,.(pValue, groups)])
```

---

**xQTLanno_chippeak**  
enrichment analysis for GWAS / QTL signals in functional elements, including enhancer, promoter, CPG, and TFs

Description

enrichment analysis for GWAS / QTL signals in functional elements, including enhancer, promoter, CPG, and TFs

Usage

```r
xQTLanno_chippeak(
  snpInfo = "",
  genomeVersion = "hg38",
  enrichElement = NULL,
  distLimit = 1
)
```

Arguments

- **snpInfo**: A data.table/data.frame with three columns: chromosome, position and p-value.
- **genomeVersion**: "hg38" (default) or "hg19". Note: hg19 will be converted to hg38 automatically.
- **enrichElement**: A data.table of data.frame object including 4 columns (consistent with bed4 format): chrom, start, end, name.
- **distLimit**: Defaults: 0 (variants overlap with elements).
Value

A data.table object

Examples

```r
url1 <- "http://bioinfo.szbl.ac.cn/xQTL_biolinks/xqtl_data/gwas/gwasSub.txt.gz"
url2 <- "http://bioinfo.szbl.ac.cn/xQTL_biolinks/xqtl_data/enhancer.txt"
snpInfo <- data.table::fread(url1, sep="\t")
enhancerDT <- data.table::fread(url2, sep="\t")
variants_hit_enhancer <- xQTLanno_chippeak(snpInfo, enrichElement=enhancerDT)
```

---

xQTLanno_genomic

annotate all signals in GWAS / QTL dataset by genome location

Description

annotate all signals in GWAS / QTL dataset by genome location

Usage

```r
xQTLanno_genomic(snpInfo = "", p_cutoff = 5e-08, genomeVersion = "hg38")
```

Arguments

- **snpInfo**: A data.table/data.frame with three columns: chromosome, position, and P-value.
- **p_cutoff**: Cutoff of p-values of significant variants that will be annotated
- **genomeVersion**: "hg38" (default) or "hg19". Note: hg19 will be converted to hg38 automatically.

Value

A data.table object of variants’ genomics distribution

---

xQTLdownload_egene

Download eGenes (eQTL Genes) for a specified gene or a tissue

Description

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

```r
xQTLdownload_egene(
  gene = "", 
  geneType = "auto", 
  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".

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

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

Description

download summary statistics data for eQTLs with a specified gene, variant, tissue or study

source of all eQTL associations is EBI eQTL category.

Usage

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

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(GTEx v7) of variants. Default: FALSE.
data_source "eQTL_catalogue"(default) or "liLab"

Value

A data.table object.

Examples

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

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

# Download eQTL associations of gene ATP11B in Muscle - Skeletal from GTEx_V8:
geneAsso <- xQTLdownload_eqtlAllAsso("ATP11B", tissueLabel="Muscle - Skeletal")

# Download all eQTL associations of SNP rs115688818 in all tissues from all supported studies.
varAsso <- xQTLdownload_eqtlAllAsso(variantName="rs115688818", study="")
# Download eQTL associations of SNP rs115688818 in Muscle - Skeletal from GTEx_V8:
varAsso <- xQTLdownload_eqtlAllAsso(variantName="chr11_102530930_T_C_b38", tissueLabel="Muscle - Skeletal")
xQTLdownload_eqtlAllAssoPos

Download summary statistics data for eQTLs with genome positions.

Description

Download summary statistics data for eQTLs with genome positions.

Usage

```r
xQTLdownload_eqtlAllAssoPos(
  chrom = "",  
pos_lower = numeric(0),  
pos_upper = numeric(0),  
p_lower = 0,  
p_upper = 1.1,  
gene = "",  
geneType = "auto",  
tissueLabel = "",  
study = "gtex_v8",  
recordPerChunk = 1000,  
withB37VariantId = FALSE
)
```

Arguments

- **chrom** (character) name of chromosome, including chr1-chr22, chrX.
- **pos_lower** (integer) lower base pair location threshold, expressed as an integer
- **pos_upper** (integer) upper base pair location threshold, expressed as an integer
- **p_lower** (numeric) lower p-value threshold, can be expressed as a float or using mantissa and exponent annotation (0.001 or 1e-3 or 1E-3)
- **p_upper** (numeric) upper p-value threshold, can be expressed as a float or using mantissa and exponent annotation (0.001 or 1e-3 or 1E-3)
- **gene** (character) gene symbol or gencode id (versioned or unversioned are both supported).
- **geneType** (character) options: "auto","geneSymbol" or "gencodeId". 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".
- **recordPerChunk** (integer) number of records fetched per request (default: 1000).
- **withB37VariantId** a logical value indicating whether to return the genome location(GTEX v7) of variants. Default: FALSE.
xQTLdownload_eqtlExp

Description

Download normalized expression for gene with a variant-gene pair

Usage

xQTLdownload_eqtlExp(
  variantName = "", gene = "", 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".
- **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.

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_exp

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

Description

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

Usage

xQTLdownload_exp(
  genes = "",
  geneType = "auto",
  tissueSiteDetail = "Liver",
  toSummarizedExperiment = FALSE,
  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 tissueSiteDetailGTExv8 or tissueSiteDetailGTExv7

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.
xQTLdownload_geneMedExp

Description

Download median expressions for multiple genes in a specified tissue

Usage

xQTLdownload_geneMedExp(
  genes = "",  
geneType = "auto",  
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".
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_hqtl

Download summary statistics data of H3K4me1 and H3K27ac histone QTL (hQTL) using a specified location

Description

Download summary statistics data of H3K4me1 and H3K27ac histone QTL (hQTL) using a specified location

Usage

xQTLdownload_hqtl(
  phenotype_id = "9:99773935-99776816",
  histone_type = "H3K27AC",
  cell_type = "monocyte",
  hqtlmeta = NULL
)

Arguments

phenotype_id phenotype_id that formatted with genome location, like: 9-99773935-99776816, can be obtained using xQTLdownload_hqtlmeta
histone_type (string) One of the histone types: "H3K27AC" or "H3K4ME1".
cell_type (string) One of the cell types: "monocyte", "neutrophil" or "T cell".
hqtlmeta A data.table object obtained via xQTLdownload_hqtlmeta.

Value

A data.table object
xQTLdownload_hqtlmeta  Download metadata for H3K4me1 and H3K27ac histone QTL (hQTL)

Description
Download metadata for H3K4me1 and H3K27ac histone QTL (hQTL)

Usage
xQTLdownload_hqtlmeta(histone_type = "H3K27AC", cell_type = "monocyte")

Arguments
- histone_type (string) One of the histone types: "H3K27AC" or "H3K4ME1".
- cell_type (string) One of the cell types: "monocyte", "neutrophil" or "T cell".

Value
A data.table object including all CpG ID

xQTLdownload_mQTL  Download summary statistics data of DNA methylation QTL (mQTL) using CpG ID

Description
Download summary statistics data of DNA methylation QTL (mQTL) using CpG ID

Usage
xQTLdownload_mQTL(
  cpg_id = "cg00000221",
  tissue_name = "WholeBlood",
  mQTL_meta = NULL
)

Arguments
- cpg_id phenotype_id like: cg00000236, can be obtained using xQTLdownload_mqtlmeta
- tissue_name (String) One of the tissues: BreastMammaryTissue, ColonTransverse, Kidney-Cortex, Lung, MuscleSkeletal, Ovary, Prostate, Testis and WholeBlood
- mQTL_meta meta data of the mQTL that can be accessed by xQTLdownload_mqtlmeta

Value
A data.table object
xQTLdownload_mqtlmeta  Download metadata of DNA methylation QTL (mQTL)

Description

Download metadata of DNA methylation QTL (mQTL)

Usage

xQTLdownload_mqtlmeta(tissue_name = "BreastMammaryTissue")

Arguments

tissue_name  (String) One of the tissues: BreastMammaryTissue, ColonTransverse, Kidney-Cortex, Lung, MuscleSkeletal, Ovary, Prostate, Testis and WholeBlood

Value

A data.table object

xQTLdownload_sc  Download all sc-eQTL associations for a specified gene

Description

Download all sc-eQTL associations for a specified gene

Usage

xQTLdownload_sc(
    gene = "BIN3",
    geneType = "geneSymbol",
    cell_type = "Astrocytes",
    cell_state = "",
    qtl_type = "Cell-type eQTL",
    study_name = "Bryois2022NN"
)

Arguments

gene  (character) gene symbol or gencode id (versioned or unversioned are both supported).
geneType  (character) options: "auto","geneSymbol" or "gencodeId". Default: "geneSymbol".
cell_type  (character) cell types supported in the list of study_info from 'xQTLquery_scInfo'
xQTLdownload_scSig

Description
Download significant sc-eQTL associations for a specified gene

Usage
xQTLdownload_scSig(
  gene = "BIN3",
  geneType = "geneSymbol",
  cell_type = "Astrocytes",
  cell_state = "",
  qtl_type = "Cell-type eQTL",
  study_name = "Bryois2022NN"
)

Arguments

gene (character) gene symbol or gencode id (versioned or unversioned are both supported).
geneType (character) options: "auto", "geneSymbol" or "gencodeId". Default: "geneSymbol".
cell_type (character) cell types supported in the list of study_info from 'xQTLquery_scInfo'
cell_state (character) cell states supported in the list of study_info from 'xQTLquery_scInfo'
qtl_type (character) QTL types supported in the list of study_info from 'xQTLquery_scInfo'
study_name (character) study name supported in the list of study_info from 'xQTLquery_scInfo'

Value
A data.table object
**xQTLdownload_sgene**

Download details of sGenes (sQTL Genes) for a specified gene or a tissue.

**Description**

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

**Usage**

```r
xQTLdownload_sgene(
  gene = "",
  geneType = "auto",
  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".
- **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**

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

Download summary statistics data for sQTLs with a specified gene or a tissue

Description

Download summary statistics data for sQTLs with a specified gene or a tissue

Usage

xQTLdownload_sqtlAllAsso(
  genes = "", 
  geneType = "auto", 
  tissue = "", 
  clu_names = "", 
  clu_geneid_DF = NULL
)

Arguments

<table>
<thead>
<tr>
<th>Argument</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>genes</td>
<td>(character) gene symbol or gencode id (versioned or unversioned are both supported).</td>
</tr>
<tr>
<td>geneType</td>
<td>(character) options: &quot;auto&quot;, &quot;geneSymbol&quot; or &quot;gencodeId&quot;. Default: &quot;gencodeId&quot;.</td>
</tr>
<tr>
<td>tissue</td>
<td>(character) details of tissues in GTEx can be listed using tissueSiteDetailGTExv8</td>
</tr>
<tr>
<td>clu_names</td>
<td>(character) If provided, only the sQTL of clu_names will be downloaded</td>
</tr>
<tr>
<td>clu_geneid_DF</td>
<td>(data.frame) If provided, clu-gencode mapping relationship will be loaded from this data.frame.</td>
</tr>
</tbody>
</table>

Value

A data.table object of sQTL dataset.

xQTLdownload_sqtlExp

Download normalized PSI value of intron for a sQTL pair

Description

Download normalized PSI value of intron for a sQTL pair
Usage

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

Arguments

- **variantName** (character): name of variant, dbSNP ID and variant id is supported, eg. "rs138420351" and "chr17_7796745_C_T_b38".
- **phenotypeId** (character): 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`.

Value

A data.table object.

Examples

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

# Dowload 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_xqtlAllAsso**

*Download summary statistics of xQTL for a specified gene, default:3'aQTL*

---

Description

Download summary statistics of xQTL for a specified gene, default:3’aQTL
Usage

```
xQTLdownload_xqtlAllAsso(
    genes = "",
    geneType = "geneSymbol",
    tissue = "",
    mRNA_refseq = "",
    mRNA_gene_DF = NULL,
    type = "3'\'aQTL"
)
```

Arguments

genes (character) gene symbol or gencode id (versioned or unversioned are both supported).
geneType (character) options: "auto","geneSymbol" or "gencodeId". Default: "geneSymbol".
tissue (character) details of tissues in GTEx can be listed using tissueSiteDetailGTExv8
mRNA_refseq (character) If provided, only the 3’aQTL of mRNA will be downloaded
mRNA_gene_DF (data.frame) If provided, mRNA-gencode mapping relationship will be loaded from this data.frame.
type 3’aQTL(default)

Value

A data.table object of xQTL dataset.

---

`xQTLquery_eqtl`  
*Query multi-tissue eQTL metasoft results*

Description

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

Usage

```
xQTLquery_eqtl(
    variantName = "",
    gene = "",
    variantType = "auto",
    geneType = "auto",
    tissueSiteDetail = "",
    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
- **recordPerChunk** (integer) number of records fetched per request (default: 100).

Value

A data.table object.

Examples

```r
# Query with a gene symbol:
eqtlInfo <- xQTLquery_eqtl(gene="TP53")

# Query with unversioned gencode ID:
eqtl_v8 <- xQTLquery_eqtl(gene="ENSG00000141510")

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

# Query with a variant-gene pair:
xQTLquery_eqtl(variantName="rs1641513",gene="TP53")
xQTLquery_eqtl(variantName="chr1_1667948_A_G_b38", gene="SLC35E2B", tissueSiteDetail="Kidney - Cortex")
```

---

**xQTLquery_eqtlSig**

Query significant eQTL associations for a specified tissue or multiple tissues.

Description

Query significant eQTL associations for a specified tissue or multiple tissues.
Usage

```r
xQTLquery_eqtlSig(
  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 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`.

Value

A data.table object.

Examples

# Query significant eQTL associations with a variant id across all tissues:
xQTLquery_eqtlSig("rs201327123")
xQTLquery_eqtlSig("chr1_14677_G_A_b38")

# Query significant eQTL associations with a variant id in a specified tissue:
xQTLquery_eqtlSig("chr1_14677_G_A_b38",
  tissueSiteDetail="Skin - Sun Exposed (Lower leg)")

# Query eQTL associations for multiple variants:
varInfo <- xQTLquery_varPos(chrom="chr1", pos=c(1102708))
xQTLquery_eqtlSig(variantName=varInfo$snpId)

# Query eQTL associations by genes or tissues:
xQTLquery_eqtlSig(genes="ATAD3B")
xQTLquery_eqtlSig(genes=c("TP53", "SLC35E2B"), tissueSiteDetail="Brain - Cerebellum")
xQTLquery_eqtlSig(genes="ENSG00000141510.16")

# Query eQTL associations with a variant-gene pair:
xQTLquery_eqtlSig(variantName="rs1641513", genes="TP53")
xQTLquery_eqtlSig(variantName="chr1_1667948_A_G_b38",
  genes="SLC35E2B", tissueSiteDetail="Kidney - Cortex")
**xQTLquery_gene**

Query basic information for genes, including name, symbol, position and description

**Description**

Query basic information for genes, including name, symbol, position and description

**Usage**

```r
xQTLquery_gene(genes = "", geneType = "auto", 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" , "ENSG00000078808" .
  
  - **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".

- **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

```r
# query gene of gencode version v26/hg38
geneInfo <- xQTLquery_gene("TP53")
geneInfo <- xQTLquery_gene(c("tp53","na0K","SDF4"))
geneInfo <- xQTLquery_gene(c("ENSG00000210195.2","ENSG00000078808"))
```

### xQTLquery_geneAll

**Query all genes supported in GTEx**

**Description**

Query all genes supported in GTEx

**Usage**

```r
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 details of samples with GTEx IDs**

**Description**

Query details of samples with GTEx IDs

**Usage**

```r
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**

```r
sampleInfo <- xQTLquery_sampleBySampleId(sampleIds)
```

---

**xQTLquery_sampleByTissue**

*Query details of samples by tissue name*

**Description**

Query details of samples by tissue name

**Usage**

```r
xQTLquery_sampleByTissue(
  tissueSiteDetail = "Liver",
  dataType = "RNASEQ",
  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".
- `recordPerChunk`: (integer) number of records fetched per request (default: 200).
- `pathologyNotesCategories`: Default: pathologyNotes info is ignored.

**Value**

A data.table object of samples' information.
xQTLquery_sc

Examples

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

---

**Description**

Query significant sc-eQTLs for a specified gene

**Usage**

```r
xQTLquery_sc(
  gene = "BIN3",
  geneType = "geneSymbol",
  cell_type = "Astrocytes",
  cell_state = "",
  qtl_type = "Cell-type eQTL",
  study_name = "Bryois2022NN"
)
```

**Arguments**

- `gene` (character) gene symbol or gencode id (versioned or unversioned are both supported).
- `geneType` (character) options: "auto", "geneSymbol" or "gencodeId". Default: "geneSymbol".
- `cell_type` (character) cell types supported in the list of study_info from `xQTLquery_scInfo`
- `cell_state` (character) cell states supported in the list of study_info from `xQTLquery_scInfo`
- `qtl_type` (character) QTL types supported in the list of study_info from `xQTLquery_scInfo`
- `study_name` (character) study name supported in the list of study_info from `xQTLquery_scInfo`

**Value**

A data.table object
xQTLquery_scInfo  

Query metadata of sc-eQTLs

Description

Query metadata of sc-eQTLs

Usage

xQTLquery_scInfo()

Value

A data.table object

xQTLquery_sqtlSig  

Query significant sQTL associations for a tissue or multiple tissues

Description

Only GTEx v8 is supported.

Usage

xQTLquery_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
xQTLquery_tissue

Value
A data.table object.

Examples

# Query sQTL associations with rsid:
xQTLquery_sqtlSig(variantName="rs201327123")
xQTLquery_sqtlSig(variantName="chr1_14677_G_A_b38", tissueSiteDetail="Whole Blood")

# Query sQTL associations with gene symbol and gencode ID:
xQTLquery_sqtlSig(genes="ENSG00000141510.16", tissueSiteDetail="Lung")
xQTLquery_sqtlSig(genes=c("ATAD3B", "MLH1"))

# Query sQTL associations with the variant-genes pair:
xQTLquery_sqtlSig(variantName="rs201327123", genes=c("WASH7P", "RP11-206L10.2"))
xQTLquery_sqtlSig(variantName="chr17_7465085_A_G_b38", genes="TP53", tissueSiteDetail="Lung")

xQTLquery_tissue  Query details for a specified tissue

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, uberon ID, and standard tissue colors. TissueSiteDetails are grouped by TissueSites. By default, this service reports from the latest GTEx release.

Usage
xQTLquery_tissue(tissueName = "")

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

Value
A data.table object.

Examples
tissueAll <- xQTLquery_tissue()  # fetch all tissues
Brain <- xQTLquery_tissue("Brain")
xQTLquery_varId

Query variant with variant ID or dbSNP ID

Description
Query variant with variant ID or dbSNP ID

Usage
xQTLquery_varId(variantName = "", variantType = "auto")

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".

Value
A data.table object.

Examples
xQTLquery_varId("rs12596338")
xQTLquery_varId("chr11_66561248_T_C_b38")

xQTLquery_varPos
Query variants using genome position.

Description
Query variants using genome position.

Usage
xQTLquery_varPos(chrom = "", pos = numeric(0), recordPerChunk = 200)

Arguments
chrom (character) name of chromosome, including chr1-chr22, chrX, chrY.

pos An integer array.

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

Value
A data.table object.
**Examples**

xQTLquery_varPos(chrom="chr1", pos=c(1102708,1105739))

---

**xQTLvisual_anno**

*Visualize enrichment of variants derived from xQTLanno_genomic*

**Description**

Visualize enrichment of variants derived from xQTLanno_genomic

**Usage**

xQTLvisual_anno(snpHits, pValueBy = 5, annoType = "enrichment")

**Arguments**

- **snpHits**: A data.table object from result of xQTLanno_genomic
- **pValueBy**: Cut step of pvalue. Defaults: 5
- **annoType**: "enrichment" or "overlapping"

**Value**

A ggplot object

---

**xQTLvisual_coloc**

*Heatmap plot of the LD-p-value relationship of the eQTL*

**Description**

Heatmap plot of the LD-p-value relationship of the eQTL

**Usage**

xQTLvisual_coloc(
    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 a data.table object and a ggplot object

Examples

heatmapQTL <- xQTLvisual_coloc(gene="MMP7", variantName="rs11568818", study="TwinsUK")

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

Description

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

Usage

xQTLvisual_eqtl(gene, geneType = "auto")

Arguments

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

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

Value

A ggplot object.
xQTLvisual_eqtlExp

Examples

xQTLvisual_eqtl("KIF15")

xQTLvisual_eqtlExp  Boxplot of normalized expression stratified by genotypes for eQTL.

Description

Boxplot of normalized expression stratified by genotypes for eQTL.

Usage

xQTLvisual_eqtlExp(
  variantName = "",  
gene = "",  
variantType = "auto",  
genType = "auto",  
tissueSiteDetail = "",  
axis_text_size = 1.3,  
axis_title_size = 1.3,  
title_size = 1.4,  
xlab_text = "Genotypes",  
ylab_text = "Normalized expression",  
ylim_v = NULL,  
title_text = "",  
jitter_color = NULL
)

Arguments

variantName  (character) name of variant, dbsnp ID and variant id is supported, eg. "rs138420351" and "chr17_7796745_C_T_b38".
gegene  (character) gene symbol or gencode id (versioned or unversioned are both supported).
variantType  (character) options: "auto", "snpId" or "variantId". Default: "auto".
genType  (character) options: "auto", "geneSymbol" or "gencodeId". Default: "auto".
tissueSiteDetail  (character) details of tissues in GTEx can be listed using tissueSiteDetailGTExv8 or tissueSiteDetailGTExv7
axis_text_size  (numeric) text size of the axis labels
axis_title_size  (numeric) text size of the axis title
title_size  (numeric) text size of the title of the plot
xQTLvisual_geneExpTissues

- **xlab_text**: Label for x-axis
- **ylab_text**: for y-axis
- **ylim_v**: Set scale limits
- **title_text**: Title of the plot
- **jitter_color**: (A character vector) Set the point color.

**Value**

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

**Examples**

```r
expEqtl<-xQTLvisual_eqtlExp(variantName="rs3778754", gene ="IRF5",
tissueSiteDetail="Whole Blood", xlab_text="Genotypes", ylab_text="Expression", ylim_v=c(-2,2),
axis_text_size=1.3, axis_title_size=1.3, title_size=1.4,
title_text="Genotype-expression", jitter_color=c("#83bea5", "#e09069","8f9dc6") )
```

**Description**

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

**Usage**

```r
xQTLvisual_geneExpTissues(
    gene = "",
    geneType = "auto",
    tissues = "All",
    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.
- **log10y**: Display values of expression in log scale. Default: FALSE.
- **toTissueSite**: TRUE or FALSE, display all subtissues or tissue Site. Default: TRUE.
Value

A list containing expression profile and a ggplot object.

Examples

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

xQTLvisual_genoBox  \hspace{1cm} Boxplot of values stratified by genotypes with customized data

Description

Boxplot of values stratified by genotypes with customized data

Usage

```r
xQTLvisual_genoBox(
  genoDT, 
  axis_text_size = 1.3, 
  axis_title_size = 1.3, 
  title_size = 1.4, 
  xlab_text = "Genotypes", 
  ylab_text = "Normalized expression", 
  ylim_v = NULL, 
  title_text = "", 
  jitter_color = NULL
)
```

Arguments

<table>
<thead>
<tr>
<th>Argument</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>genoDT</td>
<td>(Data.frame) including two columns, &quot;value&quot; and &quot;genotypes&quot;</td>
</tr>
<tr>
<td>axis_text_size</td>
<td>(numeric) text size of the axis labels</td>
</tr>
<tr>
<td>axis_title_size</td>
<td>(numeric) text size of the axis title</td>
</tr>
<tr>
<td>title_size</td>
<td>(numeric) text size of the title of the plot</td>
</tr>
<tr>
<td>xlab_text</td>
<td>(character) Label for x-axis</td>
</tr>
<tr>
<td>ylab_text</td>
<td>(character) Label for x-axis</td>
</tr>
<tr>
<td>ylim_v</td>
<td>(numeric vector) Set scale limits</td>
</tr>
<tr>
<td>title_text</td>
<td>(character) Title of the plot</td>
</tr>
<tr>
<td>jitter_color</td>
<td>(A character vector) Set the point color.</td>
</tr>
</tbody>
</table>
Value

A ggplot object.

Examples

url1 <- "http://bioinfo.szbl.ac.cn/xqTL_biolinks/xqtl_data/eqtl/eqtlExpLabel.txt"
genodT <- data.table::fread(url1)
box_plot <- xQTLvisual_genoBox(genodT, title_size=1.6, title_text="Geno-Exp association")

xQTLvisual_locusCombine

Generate a combined figure including locusZoom and locuscompare plot

Description


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. "position" (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.
xQTLvisual_locusCompare

$xQTLvisual_locusCompare$

Dotplot of comparing regional signals between GWAS and xQTL

Description

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'.
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 object of LD matrix. Default is null.

Value
A ggplot object.
Examples

```r
library(data.table)
# load data:
eqt1DF <- 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 summary statistics data*

Description

This function is rebuilt from `locuscompare.R` ([https://github.com/boxiangliu/locuscomparer/blob/master/R/locuscompare.R](https://github.com/boxiangliu/locuscomparer/blob/master/R/locuscompare.R)).

Usage

```r
xQTLvisual_locusZoom(
  DF,
  highlightSnp = "",
  population = "EUR",
  posRange = "",
  legend = TRUE,
  ylim = NULL,
  legend_position = c("topright", "bottomright", "topleft"),
  point_fill = NULL,
  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. "position" (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 populations 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.
ylim set the minimum and maximum values for the y-axis. By default, the function will automatically determine the y-axis limits based on the data being plotted.

legend_position (character, optional) Either 'bottomright', 'topright', or 'topleft'. Default: 'bottomright'.

point_fill (character, optional) Customized color vectors (5 kinds of colors).

snpLD A data.frame of LD matrix. Default is null.

Value A list containing data.table and ggplot object.

Examples

```r
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_manhattan**  
*Manhattan plot for a GWAS summary statistics dataset*

**Description**
Manhattan plot for a GWAS summary statistics dataset

**Usage**
xQTLvisual_manhattan(gwasDF, pvalue_cutoff = 1e-04, num_snp_selected = 2000)

**Arguments**
gwasDF A data.frame of summary statistics data, including four cols arranged in the following order: SNP name, chromosomes, position, p-value.
The Manhattan plot is a helpful tool for visualizing genome-wide association study results. However, when there are a large number of SNPs, the plot can become difficult to render and generate a large file size. This is due to the stacking of non-significant SNPs at the bottom of the plot. To address this issue, we can choose to filter out some of the non-significant SNPs or randomly select a subset of them to plot. This will improve the readability of the plot and reduce the file size.

Default: 2000. Number of SNPs randomly selected for each chromosome.

Value

A pdf format figure.

Examples

gwasDF <- data.table::fread(
    "http://bioinfo.szbl.ac.cn/xQTL_biolinks/xqtl_data/gwas/AD/gwasChr6Sub.txt")
xQTLvisual_manhattan(gwasDF[,.(rsid, chr, position,P)])

xQTLvisual_PZPlot

Compare P-values reported to P-values calculated from Z statistics derived from the reported beta and standard error.

Description

Compare P-values reported to P-values calculated from Z statistics derived from the reported beta and standard error.

Usage

xQTLvisual_PZPlot(
    summaryDT,  
    binCutLogP = 4, 
    binNumber = 2000, 
    distribution_func = "pnorm"
)

Arguments

<table>
<thead>
<tr>
<th>Argument</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>summaryDT</td>
<td>A data.frame with three cols: pval, beta, se.</td>
</tr>
<tr>
<td>binCutLogP</td>
<td>To speed up the rendering process of the plot for tens of millions of GWAS variants, variants with a p-value below a specified threshold (binCutLogP) are randomly sampled for display.</td>
</tr>
<tr>
<td>binNumber</td>
<td>The number of points randomly selected for plotting.</td>
</tr>
<tr>
<td>distribution_func</td>
<td>&quot;pnorm&quot;(default) or &quot;pchiwq&quot;</td>
</tr>
</tbody>
</table>
The function `xQTLvisual_qqPlot` creates a Quantile-quantile plot with p-values from GWAS summary statistics data. It is used to visualize the distribution of p-values against the expected quantiles. The function takes a data frame `summaryDT` with the column `pval` containing the p-values. Additional arguments include:

- `legend_p`: TRUE or FALSE, or legend position, including: top, bottom, left and right.
- `binCutLogP`: SNPs whose logP greater than this will be binned, other than not binned.
- `binNumber`: Number of bins.

The function returns a ggplot2 object.

**Examples**

```r
url1 <- "http://bioinfo.szbl.ac.cn/xQTL_biolinks/xqtl_data/gwasDFsub_MMP7.txt"
sumDT <- data.table::fread(url1, sep="\t")
xQTLvisual_PZPlot(sumDT[,.(pValue, beta, se)], distribution_func="pchisq")

# Sample usage
url1 <- "http://bioinfo.szbl.ac.cn/xQTL_biolinks/xqtl_data/gwasSub.txt.gz"
snpInfo <- data.table::fread(url1, sep="\t")
xQTLvisual_qqPlot(snpInfo[,.(pValue)],binCutLogP=5, binNumber=10000)
```
**xQTLvisual_sqtl**

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

**Description**

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

**Usage**

```r
xQTLvisual_sqtl(gene, geneType = "auto")
```

**Arguments**

- `gene` (character) gene symbol or gencode id (versioned or unversioned are both supported).
- `geneType` (character) options: "auto","geneSymbol" or "gencodeId". Default: "auto".

**Value**

A ggplot object.

**Examples**

```r
xQTLvisual_sqtl("KIF15")
```

**xQTLvisual_sqtlExp**

*Boxplot of normalized PSI stratified by genotypes for sQTL.*

**Description**

Boxplot of normalized PSI stratified by genotypes for sQTL.

**Usage**

```r
xQTLvisual_sqtlExp(
    variantName = "",
    phenotypeId = "",
    variantType = "auto",
    tissueSiteDetail = "",
    axis_text_size = 1.3,
    axis_title_size = 1.3,
    title_size = 1.4,
    xlab_text = "Genotypes",
```
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

axis_text_size (numeric) text size of the axis labels

axis_title_size (numeric) text size of the axis title

title_size (numeric) text size of the title of the plot

xlab_text (character) Lable for x-axis

ylab_text (character) Lable for x-axis

ylim_v (numeric vector) Set scale limits

title_text (character) Title of the plot

jitter_color (A character vector) Set the point color.

Value

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

---

**xQTL_export**

Export expression object to a specified format

**Description**

Export expression object to a specified format

**Usage**

xQTL_export(exp_object, out_format = "to_clusterP")

**Arguments**

exp_object expression object derived from xQTLdownload_exp

out_format "to_clusterP", "to_wgcna" and to "to_deseq"
Value

A data.frame/data.table object

Examples

```r
expProfiles <- xQTLdownload_exp(c("tp53","naDK","SDF4"),
                              tissueSiteDetail="Artery - Coronary",
                              pathologyNotesCategories=TRUE, toSummarizedExperiment = TRUE)
```
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