# Package 'gtexr'

September 19, 2024
Title Query the GTEx Portal API
Version 0.1.0
<b>Description</b> A convenient R interface to the Genotype-Tissue Expression (GTEx) Portal API. For more information on the API, see <a href="https://gtexportal.org/api/v2/redoc">https://gtexportal.org/api/v2/redoc</a> .
License MIT + file LICENSE
<pre>URL https://rmgpanw.github.io/gtexr/, https://github.com/rmgpanw/gtexr</pre>
<pre>BugReports https://github.com/rmgpanw/gtexr/issues</pre>
Encoding UTF-8
RoxygenNote 7.3.1
Imports cli, dplyr, httr2 (>= 1.0.0), purrr, rlang, tibble, tidyr
Suggests curl, httptest2, knitr, rmarkdown, testthat (>= 3.0.0)
Config/testthat/edition 3
VignetteBuilder knitr
NeedsCompilation no
Author Alasdair Warwick [aut, cre, cph]
Maintainer Alasdair Warwick <alasdair.warwick.19@ucl.ac.uk></alasdair.warwick.19@ucl.ac.uk>
Repository CRAN
<b>Date/Publication</b> 2024-09-19 15:40:02 UTC
Contents
calculate_expression_quantitative_trait_loci

2 Contents

calculate_splicing_quantitative_trait_loci
download
get_annotation
get_clustered_median_exon_expression
get_clustered_median_gene_expression
get_clustered_median_junction_expression
get_clustered_median_transcript_expression
get_collapsed_gene_model_exon
get_dataset_info
get_downloads_page_data
get_eqtl_genes
get_exons
get_expression_pca
get_file_list
get_fine_mapping
get_full_get_collapsed_gene_model_exon
get_functional_annotation
get_genes
get_gene_expression
get_gene_search
get_genomic_features
get_gwas_catalog_by_location
get_image
get_independent_eqtl
get_linkage_disequilibrium_by_variant_data
get_linkage_disequilibrium_data
get_maintenance_message
get_median_exon_expression
c = -c = 1
$\mathcal{C} = \mathcal{A} = 1$
$\mathcal{E} = -1 - 1$
get_multi_tissue_eqtls
get_neighbor_gene
get_news_item
get_sample_biobank_data
get_sample_datasets_endpoints
get_service_info
get_significant_single_tissue_eqtls
get_significant_single_tissue_eqtls_by_location
get_significant_single_tissue_ieqtls
get_significant_single_tissue_isqtls
get_significant_single_tissue_sqtls
get_single_nucleus_gex
get_single_nucleus_gex_summary
get_sqtl_genes
get_subject
get_tissue_site_detail
get ton expressed genes 64

get_transcripts		ulate expression quan															
get_variant	Index																70
		get_variant	 														67

calculate\_expression\_quantitative\_trait\_loci

Calculate Expression Quantitative Trait Loci

#### **Description**

Calculate your own eQTLs

- This service calculates the gene-variant association for any given pair of gene and variant, which may or may not be significant.
- This requires as input a GENCODE ID, GTEx variant ID, and tissue site detail ID.

By default, the calculation is based on the latest GTEx release.

GTEx Portal API documentation.

# Usage

```
calculate_expression_quantitative_trait_loci(
  tissueSiteDetailId,
  gencodeId,
  variantId,
  datasetId = "gtex_v8"
)
```

## **Arguments**

tissueSiteDetailId

String. The ID of the tissue of interest. Can be a GTEx specific ID (e.g. "Whole\_Blood"; use get\_tissue\_site\_detail() to see valid values) or an Ontology ID.

gencodeId String. A Versioned GENCODE ID of a gene, e.g. "ENSG00000065613.9".

variantId String. A gtex variant ID.

datasetId String. Unique identifier of a dataset. Usually includes a data source and data

release. Options: "gtex\_v8", "gtex\_snrnaseq\_pilot".

## **Details**

Notes on output:

• Beta and standard error are recorded in columns nes and error respectively (see GTEx FAQs)

- variantId contains (in order) chromosome, position, reference allele, alternative allele and human genome build separated by underscores. The reference and alternative alleles for "chr1\_13550\_G\_A\_b38" for example are "G" and "A" respectively.
- See examples for how to calculate minor and alternative allele frequencies.

Notes on input:

• Argument variantId also accepts RSIDs.

#### Value

A tibble.

#### See Also

 $Other\ Dynamic\ Association\ Endpoints:\ calculate\_ieqtls(),\ calculate\_isqtls(),\ calculate\_splicing\_quantitative and the second of the control of the co$ 

```
## Not run:
# perform request - returns a tibble with a single row
calculate_expression_quantitative_trait_loci(tissueSiteDetailId = "Whole_Blood",
                                             gencodeId = "ENSG00000203782.5",
                                             variantId = "rs79641866")
# unnest list columns with tidyr::unnest()
calculate_expression_quantitative_trait_loci(tissueSiteDetailId = "Whole_Blood",
                                             gencodeId = "ENSG00000203782.5",
                                             variantId = "rs79641866") |>
 tidyr::unnest(c("data", "genotypes"))
# to calculate minor and alternative allele frequencies
calculate_expression_quantitative_trait_loci(
 tissueSiteDetailId = "Liver",
 gencodeId = "ENSG00000237973.1",
 variantId = "rs12119111"
) |>
 dplyr::bind_rows(.id = "rsid") |>
 tidyr::separate(
  col = "variantId",
   into = c(
     "chromosome",
     "position",
     "reference_allele",
     "alternative_allele",
     "genome_build"
  ),
  sep = "_"
) |>
 # ...then ascertain alternative_allele frequency
```

calculate\_ieqtls 5

```
dplyr::mutate(
   alt_allele_count = (2 * homoAltCount) + hetCount,
   total_allele_count = 2 * (homoAltCount + hetCount + homoRefCount),
   alternative_allele_frequency = alt_allele_count / total_allele_count
) |>

dplyr::select(
   rsid,
   beta = nes,
   se = error,
   pValue,
   minor_allele_frequency = maf,
   alternative_allele_frequency,
   chromosome:genome_build,
   tissueSiteDetailId
)
## End(Not run)
```

calculate\_ieqtls

Calculate Ieqtls

# **Description**

Calculate your own Cell Specific eQTLs.

- This service calculates the gene-variant association for any given pair of gene and variant, which may or may not be significant.
- This requires as input a GENCODE ID, GTEx variant ID, and tissue site detail ID.

By default, the calculation is based on the latest GTEx release.

GTEx Portal API documentation.

# Usage

```
calculate_ieqtls(
  cellType,
  tissueSiteDetailId,
  gencodeId,
  variantId,
  datasetId = "gtex_v8"
)
```

# **Arguments**

```
cellType String. "Adipocytes", "Epithelial_cells", "Hepatocytes", "Keratinocytes", "Myocytes", "Neurons", "Neurophils".
```

6 calculate\_isqtls

tissueSiteDetailId

String. The ID of the tissue of interest. Can be a GTEx specific ID (e.g. "Whole\_Blood"; use get\_tissue\_site\_detail() to see valid values) or an Ontology ID.

Ontology ID

gencodeId String. A Versioned GENCODE ID of a gene, e.g. "ENSG00000065613.9".

variantId String. A gtex variant ID.

datasetId String. Unique identifier of a dataset. Usually includes a data source and data

release. Options: "gtex\_v8", "gtex\_snrnaseq\_pilot".

## Value

A tibble.

#### See Also

```
Other Dynamic Association Endpoints: calculate_expression_quantitative_trait_loci(), calculate_isqtls(), calculate_splicing_quantitative_trait_loci()
```

# **Examples**

calculate\_isqtls

Calculate Isqtls

# Description

Calculate your own Cell Specific sQTLs.

- This service calculates the gene-variant association for any given pair of gene and variant, which may or may not be significant.
- This requires as input a GENCODE ID, GTEx variant ID, and tissue site detail ID.

By default, the calculation is based on the latest GTEx release.

GTEx Portal API documentation.

calculate\_isqtls 7

## Usage

```
calculate_isqtls(
  cellType,
  tissueSiteDetailId,
  phenotypeId,
  variantId,
  datasetId = "gtex_v8"
)
```

# Arguments

cellType String. "Adipocytes", "Epithelial\_cells", "Hepatocytes", "Keratinocytes", "Myocytes", "Neurons", "Neutrophils".

tissueSiteDetailId String. The ID of the tissue of interest. Can be a GTEx specific ID (e.g. "Whole\_Blood"; use get\_tissue\_site\_detail() to see valid values) or an Ontology ID.

phenotypeId String. See GTEx portal FAQs for further details.

variantId String. A gtex variant ID.

datasetId String. Unique identifier of a dataset. Usually includes a data source and data

release. Options: "gtex\_v8", "gtex\_snrnaseq\_pilot".

## Value

A tibble.

### See Also

Other Dynamic Association Endpoints: calculate\_expression\_quantitative\_trait\_loci(), calculate\_ieqtls(), calculate\_splicing\_quantitative\_trait\_loci()

```
calculate_splicing_quantitative_trait_loci
                         Calculate Splicing Quantitative Trait Loci
```

# **Description**

GTEx Portal API documentation.

#### Usage

```
calculate_splicing_quantitative_trait_loci(
  tissueSiteDetailId,
  phenotypeId,
  variantId,
  datasetId = "gtex_v8"
)
```

#### **Arguments**

tissueSiteDetailId

String. The ID of the tissue of interest. Can be a GTEx specific ID (e.g. "Whole\_Blood"; use get\_tissue\_site\_detail() to see valid values) or an

Ontology ID.

phenotypeId String. See GTEx portal FAQs for further details.

variantId String. A gtex variant ID.

datasetId String. Unique identifier of a dataset. Usually includes a data source and data

release. Options: "gtex\_v8", "gtex\_snrnaseq\_pilot".

## Value

A tibble.

## See Also

```
Other Dynamic Association Endpoints: calculate_expression_quantitative_trait_loci(),
calculate_ieqtls(), calculate_isqtls()
```

```
## Not run:
# perform request - returns a tibble with a single row
calculate_splicing_quantitative_trait_loci(
  tissueSiteDetailId = "Whole_Blood",
  phenotypeId = "chr1:15947:16607:clu_40980:ENSG00000227232.5",
  variantId = "chr1_14677_G_A_b38")
## End(Not run)
```

download 9

download

Download

## **Description**

GTEx Portal API documentation

# Usage

```
download(
  materialTypes = NULL,
  tissueSiteDetailIds = NULL,
  pathCategory = NULL,
  tissueSampleIds = NULL.
  sex = NULL,
  sortBy = NULL,
  sortDirection = NULL,
  searchTerm = NULL,
  sampleIds = NULL,
  subjectIds = NULL,
  ageBrackets = NULL,
  hardyScales = NULL,
  hasExpressionData = NULL,
 hasGenotype = NULL
)
```

# **Arguments**

materialTypes

String, vector. Options: "Cells:Cell Line Viable", "DNA:DNA Genomic", "DNA:DNA Somatic", "RNA:Total RNA", "Tissue:PAXgene Preserved", "Tissue:PAXgene Preserved Paraffin-embedded", "Tissue:Fresh Frozen Tissue".

tissueSiteDetailIds

Character vector of IDs for tissues of interest. Can be GTEx specific IDs (e.g. "Whole\_Blood"; use get\_tissue\_site\_detail() to see valid values) or Ontology IDs.

pathCategory

Character vector. Options: "adenoma", "amylacea", "atelectasis", "atherosclerosis", "atherosis", "atrophy", "calcification", "cirrhosis", "clean\_specimens", "congestion", "corpora\_albicantia", "cyst", "desquamation", "diabetic", "dysplasia", "edema", "emphysema", "esophagitis", "fibrosis", "gastritis", "glomerulosclerosis", "goiter", "gynecomastoid", "hashimoto", "heart\_failure\_cells", "hemorrhage", "hepatitis", "hyalinization", "hypereosinophilia", "hyperplasia", "hypertrophy", "hypoxic", "infarction", "inflammation", "ischemic\_changes", "macrophages", "mastopathy", "metaplasia", "monckeberg", "necrosis", "nephritis", "nephrosclerosis", "no\_abnormalities", "nodularity", "pancreatitis", "pigment", "pneumonia", "post\_menopausal", "prostatitis", "saponification", "scarring", "sclerotic", "solar\_elastosis", "spermatogenesis", "steatosis", "sweat\_glands", "tma".

10 download

tissueSampleIds

Array of strings. A list of Tissue Sample ID(s).

sex String. Options: "male", "female".

sortBy String. Options: "sampleId", "ischemicTime", "aliquotId", "tissueSampleId",

"hardyScale", "pathologyNotes", "ageBracket", "tissueSiteDetailId", "sex".

sortDirection String. Options: "asc", "desc". Default = "asc".

searchTerm String.

sampleIds Character vector. GTEx sample ID.

subjectIds Character vector. GTEx subject ID.

ageBrackets The age bracket(s) of the donors of interest. Options: "20-29", "30-39", "40-49",

"50-59", "60-69", "70-79".

hardyScales Character vector. A list of Hardy Scale(s) of interest. Options: "Ventilator case",

"Fast death - violent", "Fast death - natural causes", "Intermediate death", "Slow

death".

hasExpressionData

Logical.

hasGenotype Logical.

#### **Details**

Note: running this request with no filters (i.e. download()) raises an error.

# Value

A tibble

#### See Also

Other Biobank Data Endpoints: get\_sample\_biobank\_data()

```
## Not run:
download(
   materialTypes = "RNA:Total RNA",
   tissueSiteDetailIds = "Thyroid",
   pathCategory = "clean_specimens",
   sex = "male",
   ageBrackets = "50-59"
)
## End(Not run)
```

get\_annotation 11

on Get Annotation
-------------------

# Description

This service returns the list of annotations and allowed values by which a particular dataset can be subsetted. Results may be filtered by dataset.

GTEx Portal API documentation

# Usage

```
get_annotation(datasetId = "gtex_v8", page = 0, itemsPerPage = 250)
```

# **Arguments**

datasetId String. Unique identifier of a dataset. Usually includes a data source and data

release. Options: "gtex\_v8", "gtex\_snrnaseq\_pilot".

page Integer (default = 0). itemsPerPage Integer (default = 250).

#### **Details**

Note: the output for this function appears to be incomplete currently.

## Value

A tibble

## See Also

```
Other Datasets Endpoints: get_collapsed_gene_model_exon(), get_downloads_page_data(), get_file_list(), get_full_get_collapsed_gene_model_exon(), get_functional_annotation(), get_linkage_disequilibrium_by_variant_data(), get_linkage_disequilibrium_data(), get_sample_datasets_get_subject(), get_tissue_site_detail(), get_variant(), get_variant_by_location()
```

```
## Not run:
  get_annotation()
## End(Not run)
```

#### **Description**

Find median transcript expression data along with hierarchical clusters.

- Returns median normalized transcript expression in tissues of all known transcripts of a given gene along with the hierarchical clustering results of tissues and transcripts, based on exon expression, in Newick format.
- The hierarchical clustering is performed by calculating Euclidean distances and using the average linkage method.
- This endpoint is not paginated.

By default, this endpoint queries the latest GTEx release.

GTEx Portal API documentation

# Usage

```
get_clustered_median_exon_expression(
  gencodeIds,
  datasetId = "gtex_v8",
  tissueSiteDetailIds = NULL
)
```

# **Arguments**

gencodeIds A character vector of Versioned GENCODE IDs, e.g. c("ENSG00000132693.12", 
"ENSG00000203782.5").

datasetId String. Unique identifier of a dataset. Usually includes a data source and data 
release. Options: "gtex\_v8", "gtex\_snrnaseq\_pilot".

tissueSiteDetailIds Character vector of IDs for tissues of interest. Can be GTEx specific IDs (e.g. 
"Whole\_Blood"; use get\_tissue\_site\_detail() to see valid values) or Ontology IDs.

# Value

A tibble, with clustering data stored as an attribute, "clusters".

#### See Also

```
Other Expression Data Endpoints: get_clustered_median_gene_expression(), get_clustered_median_junction_expget_clustered_median_transcript_expression(), get_expression_pca(), get_gene_expression(), get_median_exon_expression(), get_median_gene_expression(), get_median_junction_expression(), get_median_transcript_expression(), get_single_nucleus_gex(), get_single_nucleus_gex_summary(), get_top_expressed_genes()
```

## **Examples**

```
get_clustered_median_gene_expression
Get Clustered Median Gene Expression
```

## **Description**

Find median gene expression data along with hierarchical clusters.

- Returns median gene expression in tissues along with The hierarchical clustering results of tissues and genes, based on gene expression, in Newick format.
- Results may be filtered by dataset, gene or tissue, but at least one gene must be provided
- The hierarchical clustering is performed by calculating Euclidean distances and using the average linkage method.
- This endpoint is not paginated.

By default, this service queries the latest GTEx release.

GTEx Portal API documentation

```
get_clustered_median_gene_expression(
  gencodeIds,
  datasetId = "gtex_v8",
  tissueSiteDetailIds = NULL
)
```

### **Arguments**

gencodeIds A character vector of Versioned GENCODE IDs, e.g. c("ENSG00000132693.12", "ENSG00000203782.5").

datasetId String. Unique identifier of a dataset. Usually includes a data source and data release. Options: "gtex\_v8", "gtex\_snrnaseq\_pilot".

tissueSiteDetailIds Character vector of IDs for tissues of interest. Can be GTEx specific IDs (e.g. "Whole\_Blood"; use get\_tissue\_site\_detail() to see valid values) or Ontology IDs.

#### Value

A tibble, with clustering data stored as an attribute, "clusters".

#### See Also

```
Other Expression Data Endpoints: get_clustered_median_exon_expression(), get_clustered_median_junction_expression(), get_clustered_median_transcript_expression(), get_expression_pca(), get_gene_expression(), get_median_exon_expression(), get_median_gene_expression(), get_median_junction_expression(), get_median_transcript_expression(), get_single_nucleus_gex(), get_single_nucleus_gex_summary(), get_top_expressed_genes()
```

```
get_clustered_median_junction_expression
Get Clustered Median Junction Expression
```

# **Description**

Find median junction expression data along with hierarchical clusters.

- Returns median junction read counts in tissues of a given gene from all known transcripts
  along with the hierarchical clustering results of tissues and genes, based on junction expression, in Newick format.
- Results may be filtered by dataset, gene or tissue, but at least one gene must be provided.
- The hierarchical clustering is performed by calculating Euclidean distances and using the average linkage method.
- This endpoint is not paginated.

By default, this service queries the latest GTEx release.

GTEx Portal API documentation

#### Usage

```
get_clustered_median_junction_expression(
  gencodeIds,
  datasetId = "gtex_v8",
  tissueSiteDetailIds = NULL
)
```

## **Arguments**

```
gencodeIds A character vector of Versioned GENCODE IDs, e.g. c("ENSG00000132693.12", 
"ENSG00000203782.5").

datasetId String. Unique identifier of a dataset. Usually includes a data source and data 
release. Options: "gtex_v8", "gtex_snrnaseq_pilot".

tissueSiteDetailIds Character vector of IDs for tissues of interest. Can be GTEx specific IDs (e.g. 
"Whole_Blood"; use get_tissue_site_detail() to see valid values) or On-
tology IDs.
```

#### Value

A tibble, with clustering data stored as an attribute, "clusters".

## See Also

```
Other Expression Data Endpoints: get_clustered_median_exon_expression(), get_clustered_median_gene_expression(), get_clustered_median_transcript_expression(), get_expression_pca(), get_gene_expression(), get_median_exon_expression(), get_median_gene_expression(), get_median_junction_expression(), get_median_transcript_expression(), get_single_nucleus_gex(), get_single_nucleus_gex_summary(), get_top_expressed_genes()
```

# **Examples**

# **Description**

Find median transcript expression data of all known transcripts of a gene along with hierarchical clusters.

- Returns median normalized expression in tissues of all known transcripts of a given gene along
  with the hierarchical clustering results of tissues and genes, based on expression, in Newick
  format.
- Results may be filtered by dataset, gene or tissue, but at least one gene must be provided.
- The hierarchical clustering is performed by calculating Euclidean distances and using the average linkage method.
- This endpoint is not paginated.

By default, this service queries the latest GTEx release.

GTEx Portal API documentation

```
get_clustered_median_transcript_expression(
  gencodeIds,
  datasetId = "gtex_v8",
  tissueSiteDetailIds = NULL
)
```

### **Arguments**

gencodeIds A character vector of Versioned GENCODE IDs, e.g. c("ENSG00000132693.12", "ENSG00000203782.5").

datasetId String. Unique identifier of a dataset. Usually includes a data source and data release. Options: "gtex\_v8", "gtex\_snrnaseq\_pilot".

tissueSiteDetailIds Character vector of IDs for tissues of interest. Can be GTEx specific IDs (e.g. "Whole\_Blood"; use get\_tissue\_site\_detail() to see valid values) or Ontology IDs.

## Value

A tibble, with clustering data stored as an attribute, "clusters".

#### See Also

```
Other Expression Data Endpoints: get_clustered_median_exon_expression(), get_clustered_median_gene_expression get_clustered_median_junction_expression(), get_expression_pca(), get_gene_expression(), get_median_exon_expression(), get_median_gene_expression(), get_median_junction_expression(), get_median_transcript_expression(), get_single_nucleus_gex(), get_single_nucleus_gex_summary(), get_top_expressed_genes()
```

# **Description**

This service returns the collapsed exons in the gene model of the given gene. Gene-level and exon-level expression quantification were based on the GENCODE annotation, collapsed to a single transcript model for each gene using an algorithm developed by the GTEx analysis team.

By default, this service queries the models used by the latest GTEx release.

GTEx Portal API documentation

# Usage

```
get_collapsed_gene_model_exon(
  gencodeId,
  datasetId = "gtex_v8",
  page = 0,
  itemsPerPage = 250
)
```

## Arguments

```
gencodeId String. A Versioned GENCODE ID of a gene, e.g. "ENSG00000065613.9".

datasetId String. Unique identifier of a dataset. Usually includes a data source and data release. Options: "gtex_v8", "gtex_snrnaseq_pilot".

page Integer (default = 0).

itemsPerPage Integer (default = 250).
```

## Value

A tibble

#### See Also

```
Other Datasets Endpoints: get_annotation(), get_downloads_page_data(), get_file_list(), get_full_get_collapsed_gene_model_exon(), get_functional_annotation(), get_linkage_disequilibrium_by_get_linkage_disequilibrium_data(), get_sample_datasets_endpoints(), get_subject(), get_tissue_site_detail(), get_variant(), get_variant_by_location()
```

```
## Not run:
    get_collapsed_gene_model_exon(gencodeId = "ENSG000000132693.12")
## End(Not run)
```

get\_dataset\_info

get\_dataset\_info

Get Dataset Info

# **Description**

GTEx Portal API documentation

# Usage

```
get_dataset_info(datasetId = "gtex_v8", organizationName = "GTEx Consortium")
```

# **Arguments**

datasetId

String. Unique identifier of a dataset. Usually includes a data source and data release. Options: "gtex\_v8", "gtex\_snrnaseq\_pilot".

organizationName

String. Options: "GTEx Consortium" "Kid's First".

# **Details**

Note: this current only returns empty values.

# Value

A tibble.

# **Examples**

```
## Not run:
get_dataset_info()
## End(Not run)
```

```
get_downloads_page_data
```

Get Downloads Page Data

# **Description**

Retrieves all the files belonging to the given project\_id for display on the Downloads Page GTEx Portal API documentation

```
get_downloads_page_data(project_id = "adult-gtex")
```

# Arguments

```
project_id String. Options: "gtex", "adult-gtex", "egtex".
```

#### **Details**

Note: The GTEx Portal API documentation states "GTEx currently has one project available: gtex". However, project\_id values "adult-gtex" and "egtex" both return results, whereas "gtex" does not (see examples).

#### Value

A Tibble

#### See Also

```
Other Datasets Endpoints: get_annotation(), get_collapsed_gene_model_exon(), get_file_list(), get_full_get_collapsed_gene_model_exon(), get_functional_annotation(), get_linkage_disequilibrium_by_get_linkage_disequilibrium_data(), get_sample_datasets_endpoints(), get_subject(), get_tissue_site_detail(), get_variant(), get_variant_by_location()
```

```
## Not run:
# "adult-gtex" (default `project_id` value) and "egtex" both return results
get_downloads_page_data()
egtex <- get_downloads_page_data("egtex")</pre>
 egtex
 # ... "gtex" does not
 get_downloads_page_data("gtex")
 # get details for whole blood methylation data, including download URL
 purrr::pluck(
  egtex$children,
   1,
   "folders",
   "Methylation - EPIC Array",
   "children",
   "folders",
   "mQTLs",
  "children",
   "files",
   "WholeBlood.mQTLs.regular.txt.gz"
)
## End(Not run)
```

get\_eqtl\_genes 21

# Description

Retrieve eGenes (eQTL Genes).

- This service returns eGenes (eQTL Genes) from the specified dataset.
- eGenes are genes that have at least one significant cis-eQTL acting upon them.
- Results may be filtered by tissue. By default, the service queries the latest GTEx release.

For each eGene, the results include the allelic fold change (log2AllelicFoldChange), p-value (pValue), p-value threshold (pValueThreshold), empirical p-value (empiricalPValue), and q-value (qValue).

- The log2AllelicFoldChange is the allelic fold change (in log2 scale) of the most significant eQTL.
- The pValue is the nominal p-value of the most significant eQTL.
- The pValueThreshold is the p-value threshold used to determine whether a cis-eQTL for this gene is significant. For more details see https://gtexportal.org/home/documentationPage#staticTextAnalysisMethods.
- The empirical PValue is the beta distribution-adjusted empirical p-value from FastQTL.
- The qValues were calculated based on the empirical p-values. A false discovery rate (FDR) threshold of <= 0.05 was applied to identify genes with a significant eQTL.

GTEx Portal API documentation.

## Usage

```
get_eqtl_genes(
  tissueSiteDetailIds,
  datasetId = "gtex_v8",
  page = 0,
  itemsPerPage = 250
)
```

## Arguments

tissueSiteDetailIds

Character vector of IDs for tissues of interest. Can be GTEx specific IDs (e.g. "Whole\_Blood"; use get\_tissue\_site\_detail() to see valid values) or Ontology IDs.

datasetId String. Unique identifier of a dataset. Usually includes a data source and data

release. Options: "gtex\_v8", "gtex\_snrnaseq\_pilot".

page Integer (default = 0). itemsPerPage Integer (default = 250). 22 get\_exons

## Value

A tibble.

# See Also

```
Other Static Association Endpoints: get_fine_mapping(), get_independent_eqtl(), get_multi_tissue_eqtls(), get_significant_single_tissue_eqtls(), get_significant_single_tissue_eqtls_by_location(), get_significant_single_tissue_isqtls(), get_signif
```

# **Examples**

```
## Not run:
get_eqtl_genes(c("Whole_Blood", "Artery_Aorta"))
## End(Not run)
```

get\_exons

Get Exons

## **Description**

This service returns exons from all known transcripts of the given gene.

- A versioned GENCODE ID is required to ensure that all exons are from a single gene.
- A dataset ID or both GENCODE version and genome build must be provided.
- Although annotated exons are not dataset dependent, specifying a dataset here is equivalent to specifying the GENCODE version and genome build used by that dataset.

GTEx Portal API documentation

```
get_exons(
  gencodeId,
  gencodeVersion = NULL,
  genomeBuild = NULL,
  datasetId = "gtex_v8",
  page = 0,
  itemsPerPage = 250
)
```

get\_expression\_pca 23

# Arguments

```
gencodeId String. A Versioned GENCODE ID of a gene, e.g. "ENSG00000065613.9".

gencodeVersion String (default = "v26"). GENCODE annotation release. Either "v26" or "v19".

genomeBuild String. Options: "GRCh38/hg38", "GRCh37/hg19". Default = "GRCh38/hg38".

datasetId String. Unique identifier of a dataset. Usually includes a data source and data release. Options: "gtex_v8", "gtex_snrnaseq_pilot".

page Integer (default = 0).

itemsPerPage Integer (default = 250).
```

#### Value

A tibble.

#### See Also

```
Other Reference Genome Endpoints: get_gene_search(), get_genes(), get_genomic_features(), get_gwas_catalog_by_location(), get_neighbor_gene(), get_transcripts()
```

# **Examples**

# Description

Find gene expression PCA data.

- Returns gene expression PCA (principal component analysis) in tissues.
- Results may be filtered by tissue, sample, or dataset.

By default, the service queries the latest GTEx release.

GTEx Portal API documentation

```
get_expression_pca(
  tissueSiteDetailIds,
  datasetId = "gtex_v8",
  sampleId = NULL,
  page = 0,
  itemsPerPage = 250
)
```

24 get\_file\_list

## **Arguments**

tissueSiteDetailIds

Character vector of IDs for tissues of interest. Can be GTEx specific IDs (e.g. "Whole\_Blood"; use get\_tissue\_site\_detail() to see valid values) or On-

tology IDs.

datasetId String. Unique identifier of a dataset. Usually includes a data source and data

release. Options: "gtex\_v8", "gtex\_snrnaseq\_pilot".

sampleId String. ^GTEX-[A-Z0-9]{5}-[0-9]{4}-SM-[A-Z0-9]{5}\$

page Integer (default = 0). itemsPerPage Integer (default = 250).

#### Value

A tibble.

#### See Also

```
Other Expression Data Endpoints: get_clustered_median_exon_expression(), get_clustered_median_gene_expression(), get_clustered_median_junction_expression(), get_clustered_median_transcript_expression(), get_gene_expression(), get_median_exon_expression(), get_median_gene_expression(), get_median_junction_expression(), get_median_transcript_expression(), get_single_nucleus_gex(), get_single_nucleus_gex_summary(), get_top_expressed_genes()
```

## **Examples**

get\_file\_list

Get File List

## **Description**

Get all the files in GTEx dataset for Download page

GTEx Portal API documentation

```
get_file_list()
```

get\_fine\_mapping 25

## **Details**

The returned tibble includes a nested list column, "filesets". This details files, sub-categorised by fileset (see examples section).

## Value

A tibble

#### See Also

```
Other Datasets Endpoints: get_annotation(), get_collapsed_gene_model_exon(), get_downloads_page_data(), get_full_get_collapsed_gene_model_exon(), get_functional_annotation(), get_linkage_disequilibrium_by_get_linkage_disequilibrium_data(), get_sample_datasets_endpoints(), get_subject(), get_tissue_site_detail(), get_variant(), get_variant_by_location()
```

# **Examples**

get\_fine\_mapping

Get Fine Mapping

# **Description**

Retrieve Fine Mapping Data

- Finds and returns Fine Mapping data for the provided list of genes
- By default, this endpoint fetches data from the latest GTEx version

The retrieved data is split into pages with items\_per\_page entries per page

GTEx Portal API documentation

26 get\_fine\_mapping

## Usage

```
get_fine_mapping(
  gencodeIds,
  datasetId = "gtex_v8",
  variantId = NULL,
  tissueSiteDetailIds = NULL,
  page = 0,
  itemsPerPage = 250
)
```

## Arguments

gencodeIds A character vector of Versioned GENCODE IDs, e.g. c("ENSG00000132693.12",

"ENSG00000203782.5").

datasetId String. Unique identifier of a dataset. Usually includes a data source and data

release. Options: "gtex\_v8", "gtex\_snrnaseq\_pilot".

variantId String. A gtex variant ID.

tissueSiteDetailIds

Character vector of IDs for tissues of interest. Can be GTEx specific IDs (e.g. "Whole\_Blood"; use get\_tissue\_site\_detail() to see valid values) or On-

tology IDs.

page Integer (default = 0). itemsPerPage Integer (default = 250).

## Value

A tibble.

## See Also

```
Other Static Association Endpoints: get_eqtl_genes(), get_independent_eqtl(), get_multi_tissue_eqtls(), get_significant_single_tissue_eqtls(), get_significant_single_tissue_eqtls_by_location(), get_significant_single_tissue_isqtls(), get_signific
```

```
get_full_get_collapsed_gene_model_exon
```

```
27
```

```
## End(Not run)
```

# **Description**

This service allows the user to query the full Collapsed Gene Model Exon of a specific gene by gencode ID

GTEx Portal API documentation

# Usage

```
get_full_get_collapsed_gene_model_exon(gencodeId, page = 0, itemsPerPage = 250)
```

## Arguments

gencodeId String. A Versioned GENCODE ID of a gene, e.g. "ENSG00000065613.9".

page Integer (default = 0).

itemsPerPage Integer (default = 250).

#### Value

A tibble

## See Also

```
Other Datasets Endpoints: get_annotation(), get_collapsed_gene_model_exon(), get_downloads_page_data(), get_file_list(), get_functional_annotation(), get_linkage_disequilibrium_by_variant_data(), get_linkage_disequilibrium_data(), get_sample_datasets_endpoints(), get_subject(), get_tissue_site_detail(), get_variant(), get_variant_by_location()
```

```
## Not run:
    get_full_get_collapsed_gene_model_exon(gencodeId = "ENSG00000203782.5")
## End(Not run)
```

## **Description**

This endpoint retrieves the functional annotation of a certain chromosome location. Default to most recent dataset release.

GTEx Portal API documentation

# Usage

```
get_functional_annotation(
  datasetId = "gtex_v8",
  chromosome,
  start,
  end,
  page = 0,
  itemsPerPage = 250
)
```

# **Arguments**

 $\label{eq:datasetId} String. \ Unique identifier of a dataset. \ Usually includes a data source and data release. Options: "gtex_v8", "gtex_snrnaseq_pilot". \\ chromosome \ String. \ One of "chr1", "chr2", "chr3", "chr4", "chr5", "chr6", "chr7", "chr8", "chr9", "chr10", "chr11", "chr12", "chr13", "chr14", "chr15", "chr16", "chr17", "chr18", "chr19", "chr20", "chr21", "chr22", "chrM", "chrX", "chrY". \\ start \ Integer. \\ end \ Integer. \\ page \ Integer (default = 0). \\$ 

Integer (default = 250).

# Value

A tibble

itemsPerPage

# See Also

```
Other Datasets Endpoints: get_annotation(), get_collapsed_gene_model_exon(), get_downloads_page_data(), get_file_list(), get_full_get_collapsed_gene_model_exon(), get_linkage_disequilibrium_by_variant_data get_linkage_disequilibrium_data(), get_sample_datasets_endpoints(), get_subject(), get_tissue_site_detail(), get_variant(), get_variant_by_location()
```

get\_genes 29

# **Examples**

```
## Not run:
   get_functional_annotation(chromosome = "chr1", start = 192168000, end = 192169000)
## End(Not run)
```

get\_genes

Get Genes

# **Description**

This service returns information about reference genes. A genome build and GENCODE version must be provided.

- Genes are searchable by gene symbol, GENCODE ID and versioned GENCODE ID.
- Versioned GENCODE ID is recommended to ensure unique ID matching.
- By default, this service queries the genome build and GENCODE version used by the latest GTEx release.

# GTEx API Portal documentation

# Usage

```
get_genes(
  geneIds,
  gencodeVersion = "v26",
  genomeBuild = "GRCh38/hg38",
  page = 0,
  itemsPerPage = 250
)
```

Integer (default = 250).

# **Arguments**

geneIds A character vector of gene symbols, versioned gencodeIds, or unversioned gencodeIds.

gencodeVersion String (default = "v26"). GENCODE annotation release. Either "v26" or "v19".

genomeBuild String. Options: "GRCh38/hg38", "GRCh37/hg19". Default = "GRCh38/hg38".

page Integer (default = 0).

#### Value

A tibble

itemsPerPage

30 get\_gene\_expression

## See Also

```
Other Reference Genome Endpoints: get_exons(), get_gene_search(), get_genomic_features(), get_gwas_catalog_by_location(), get_neighbor_gene(), get_transcripts()
```

## **Examples**

## **Description**

Find normalized gene expression data.

- Returns normalized gene expression in tissues at the sample level.
- Results may be filtered by dataset, gene or tissue, but at least one gene must be provided.

By default, this service queries the latest GTEx release.

GTEx Portal API documentation

## Usage

```
get_gene_expression(
  gencodeIds,
  datasetId = "gtex_v8",
  tissueSiteDetailIds = NULL,
  attributeSubset = NULL,
  page = 0,
  itemsPerPage = 250
)
```

## **Arguments**

get\_gene\_search 31

```
page Integer (default = 0).
itemsPerPage Integer (default = 250).
```

#### Value

A tibble.

## See Also

```
Other Expression Data Endpoints: get_clustered_median_exon_expression(), get_clustered_median_gene_expression(), get_clustered_median_junction_expression(), get_clustered_median_transcript_expression(), get_expression_pca(), get_median_exon_expression(), get_median_gene_expression(), get_median_junction_expression(), get_median_transcript_expression(), get_single_nucleus_gex(), get_single_nucleus_gex_summary(), get_top_expressed_genes()
```

# **Examples**

```
## Not run:
 # multiple genes, selected tissues
 get_gene_expression(gencodeIds = c("ENSG00000132693.12",
                                     "ENSG00000203782.5"),
                      tissueSiteDetailIds = c("Thyroid", "Whole_Blood"))
 # single gene, selected (single) tissue
 get_gene_expression(gencodeIds = "ENSG00000132693.12",
                      tissueSiteDetailIds = "Whole_Blood")
 # subset by sex
 get_gene_expression(gencodeIds = "ENSG00000132693.12";
                      tissueSiteDetailIds = "Whole_Blood",
                      attributeSubset = "sex")
 # subset by age bracket
 get_gene_expression(gencodeIds = "ENSG00000132693.12",
                      tissueSiteDetailIds = "Whole_Blood",
                      attributeSubset = "ageBracket")
## End(Not run)
```

get\_gene\_search

Get Gene Search

## **Description**

Find genes that are partial or complete match of a gene\_id

- gene\_id could be a gene symbol, a gencode ID, or an Ensemble ID
- Gencode Version and Genome Build must be specified

GTEx Portal API documentation

32 get\_genomic\_features

## Usage

```
get_gene_search(
  geneId,
  gencodeVersion = "v26",
  genomeBuild = "GRCh38/hg38",
  page = 0,
  itemsPerPage = 250
)
```

## **Arguments**

#### Value

A tibble.

## See Also

```
Other Reference Genome Endpoints: get_exons(), get_genes(), get_genomic_features(), get_gwas_catalog_by_location(), get_neighbor_gene(), get_transcripts()
```

# **Examples**

```
## Not run:
get_gene_search("CRP")
## End(Not run)
```

```
get_genomic_features Get Genomic Features
```

# Description

GTEx API Portal documentation

```
get_genomic_features(.featureId, datasetId = "gtex_v8")
```

# **Arguments**

. featureId String. A genomic feature e.g. GENCODE ID, RSID or GTEx Variant ID.

datasetId String. Unique identifier of a dataset. Usually includes a data source and data

release. Options: "gtex\_v8", "gtex\_snrnaseq\_pilot".

## **Details**

This endpoint takes a path parameter "featureId".

## Value

A tibble.

#### See Also

```
Other Reference Genome Endpoints: get_exons(), get_gene_search(), get_genes(), get_gwas_catalog_by_location get_neighbor_gene(), get_transcripts()
```

## **Examples**

```
## Not run:
# gene symbol
get_genomic_features("brca1")

# GENCODE ID
get_genomic_features("ENSG00000132693.12")

# RSID
get_genomic_features("rs1815739")

# GTEx variant ID
get_genomic_features("chr11_66561023_G_GTTA_b38")

## End(Not run)
```

# Description

Find the GWAS Catalog on a certain chromosome between start and end locations.

GTEx API Portal documentation

34 get\_image

## Usage

```
get_gwas_catalog_by_location(
   start,
   end,
   chromosome,
   page = 0,
   itemsPerPage = 250
)
```

## Arguments

start Integer. end Integer.

chromosome String. One of "chr1", "chr2", "chr3", "chr4", "chr5", "chr6", "chr6", "chr7", "chr8",

"chr9", "chr10", "chr11", "chr12", "chr13", "chr14", "chr15", "chr16", "chr17",

"chr18", "chr19", "chr20", "chr21", "chr22", "chrM", "chrX", "chrY".

page Integer (default = 0). itemsPerPage Integer (default = 250).

## Value

A tibble.

## See Also

```
Other Reference Genome Endpoints: get_exons(), get_gene_search(), get_genes(), get_genomic_features(), get_neighbor_gene(), get_transcripts()
```

# **Examples**

```
## Not run:
get_gwas_catalog_by_location(start = 1, end = 10000000, chromosome = "chr1")
## End(Not run)
```

get\_image

Get Image

# Description

GTEx Portal API documentation

```
get_image(tissueSampleIds = NULL, page = 0, itemsPerPage = 250)
```

get\_independent\_eqtl 35

## **Arguments**

```
tissueSampleIds  Array \ of \ strings. \ A \ list \ of \ Tissue \ Sample \ ID(s).  page  Integer \ (default = 0).   itemsPerPage \qquad Integer \ (default = 250).
```

#### Value

A tibble.

# **Examples**

```
## Not run:
get_image()

# filter by `tissueSampleId`
result <- get_image(tissueSampleIds = "GTEX-1117F-0526")
print(result)

# note that `pathologyNotesCategories` (if present) is a list column
print(result$pathologyNotesCategories)

## End(Not run)</pre>
```

```
get_independent_eqtl Get Independent Eqtl
```

## **Description**

Retrieve Independent eQTL Data

- Finds and returns Independent eQTL Data data for the provided list of genes
- By default, this endpoint fetches data from the latest GTEx version

The retrieved data is split into pages with items\_per\_page entries per page

GTEx portal API documentation

```
get_independent_eqtl(
  gencodeIds,
  tissueSiteDetailIds = NULL,
  datasetId = "gtex_v8",
  page = 0,
  itemsPerPage = 250
)
```

## **Arguments**

gencodeIds A character vector of Versioned GENCODE IDs, e.g. c("ENSG00000132693.12",

"ENSG00000203782.5").

tissueSiteDetailIds

Character vector of IDs for tissues of interest. Can be GTEx specific IDs (e.g. "Whole\_Blood"; use get\_tissue\_site\_detail() to see valid values) or On-

tology IDs.

datasetId String. Unique identifier of a dataset. Usually includes a data source and data

release. Options: "gtex\_v8", "gtex\_snrnaseq\_pilot".

page Integer (default = 0). itemsPerPage Integer (default = 250).

#### Value

A tibble.

## See Also

```
Other Static Association Endpoints: get_eqtl_genes(), get_fine_mapping(), get_multi_tissue_eqtls(), get_significant_single_tissue_eqtls(), get_significant_single_tissue_eqtls_by_location(), get_significant_single_tissue_isqtls(), get_significant_single_tisqtls(), get_significant_single_tissue_isqtls(), get_significant_single
```

# **Examples**

# **Description**

Find linkage disequilibrium (LD) data for a given variant

GTEx Portal API documentation

### **Usage**

```
get_linkage_disequilibrium_by_variant_data(
  variantId,
  page = 0,
  itemsPerPage = 250
)
```

### **Arguments**

```
variantId String. A gtex variant ID.

page Integer (default = 0).

itemsPerPage Integer (default = 250).
```

### Value

A tibble.

### See Also

```
Other Datasets Endpoints: get_annotation(), get_collapsed_gene_model_exon(), get_downloads_page_data(), get_file_list(), get_full_get_collapsed_gene_model_exon(), get_functional_annotation(), get_linkage_disequilibrium_data(), get_sample_datasets_endpoints(), get_subject(), get_tissue_site_detail(), get_variant(), get_variant_by_location()
```

### **Examples**

```
get_linkage_disequilibrium_by_variant_data("chr1_159245536_C_T_b38")
```

```
get_linkage_disequilibrium_data
```

Get Linkage Disequilibrium Data

# **Description**

Find linkage disequilibrium (LD) data for a given gene.

This endpoint returns linkage disequilibrium data for the cis-eQTLs found associated with the provided gene in a specified dataset. Results are queried by gencode ID. By default, the service queries the latest GTEx release. Specify a dataset ID to fetch results from a different dataset.

GTEx Portal API documentation

```
get_linkage_disequilibrium_data(
  gencodeId,
  datasetId = "gtex_v8",
  page = 0,
  itemsPerPage = 250
)
```

### **Arguments**

gencodeId String. A Versioned GENCODE ID of a gene, e.g. "ENSG00000065613.9".

datasetId String. Unique identifier of a dataset. Usually includes a data source and data

release. Options: "gtex\_v8", "gtex\_snrnaseq\_pilot".

page Integer (default = 0). itemsPerPage Integer (default = 250).

#### Value

A tibble.

# See Also

```
Other Datasets Endpoints: get_annotation(), get_collapsed_gene_model_exon(), get_downloads_page_data(), get_file_list(), get_full_get_collapsed_gene_model_exon(), get_functional_annotation(), get_linkage_disequilibrium_by_variant_data(), get_sample_datasets_endpoints(), get_subject(), get_tissue_site_detail(), get_variant(), get_variant_by_location()
```

### **Examples**

```
get_linkage_disequilibrium_data(gencodeId = "ENSG00000132693.12")
```

get\_maintenance\_message

Get Maintenance Message

### **Description**

Getting all the maintenance messages from the database that are enabled.

GTEx Portal API documentation.

### Usage

```
get_maintenance_message(page = 0, itemsPerPage = 250)
```

#### **Arguments**

```
page Integer (default = 0).
itemsPerPage Integer (default = 250).
```

#### **Details**

Note this typically returns an empty tibble.

### Value

A tibble.

### See Also

```
Other Admin Endpoints: get_news_item()
```

### **Examples**

```
## Not run:
get_maintenance_message()
## End(Not run)
```

```
get_median_exon_expression
```

Get Median Exon Expression

# Description

Find median exon expression data.

- Returns median exon read counts, in tissues, of a collapsed gene model.
- Results may be filtered by dataset, gene or tissue, but at least one gene must be provided

By default, this service queries the latest GTEx release.

GTEx Portal API documentation

#### Usage

```
get_median_exon_expression(
  gencodeIds,
  datasetId = "gtex_v8",
  tissueSiteDetailIds = NULL,
  page = 0,
  itemsPerPage = 250
)
```

### **Arguments**

gencodeIds A character vector of Versioned GENCODE IDs, e.g. c("ENSG00000132693.12",

"ENSG00000203782.5").

datasetId String. Unique identifier of a dataset. Usually includes a data source and data

release. Options: "gtex\_v8", "gtex\_snrnaseq\_pilot".

tissueSiteDetailIds

Character vector of IDs for tissues of interest. Can be GTEx specific IDs (e.g. "Whole\_Blood"; use  $get_tissue_site_detail()$  to see valid values) or On-

tology IDs.

page Integer (default = 0). itemsPerPage Integer (default = 250).

### Value

A tibble.

#### See Also

```
Other Expression Data Endpoints: get_clustered_median_exon_expression(), get_clustered_median_gene_expression(), get_clustered_median_junction_expression(), get_clustered_median_transcript_expression(), get_expression_pca(), get_gene_expression(), get_median_gene_expression(), get_median_junction_expression(), get_median_transcript_expression(), get_single_nucleus_gex(), get_single_nucleus_gex_summary(), get_top_expressed_genes()
```

# **Examples**

### **Description**

Find median gene expression data along with hierarchical clusters.

- Returns median gene expression in tissues.
- By default, this endpoint queries the latest GTEx release.

GTEx Portal API documentation

```
get_median_gene_expression(
  gencodeIds,
  datasetId = "gtex_v8",
  tissueSiteDetailIds = NULL,
  page = 0,
  itemsPerPage = 250
)
```

#### **Arguments**

gencodeIds A character vector of Versioned GENCODE IDs, e.g. c("ENSG00000132693.12",

"ENSG00000203782.5").

datasetId String. Unique identifier of a dataset. Usually includes a data source and data

release. Options: "gtex\_v8", "gtex\_snrnaseq\_pilot".

tissueSiteDetailIds

Character vector of IDs for tissues of interest. Can be GTEx specific IDs (e.g. "Whole\_Blood"; use get\_tissue\_site\_detail() to see valid values) or On-

tology IDs.

page Integer (default = 0). itemsPerPage Integer (default = 250).

#### Value

A tibble.

#### See Also

```
Other Expression Data Endpoints: get_clustered_median_exon_expression(), get_clustered_median_gene_expression(), get_clustered_median_junction_expression(), get_clustered_median_transcript_expression(), get_expression_pca(), get_gene_expression(), get_median_exon_expression(), get_median_junction_expression(), get_median_transcript_expression(), get_single_nucleus_gex(), get_single_nucleus_gex_summary(), get_top_expressed_genes()
```

### **Examples**

```
## Not run:
get_median_gene_expression(gencodeIds = "ENSG00000132693.12")
## End(Not run)
```

```
get_median_junction_expression
```

Get Median Junction Expression

# **Description**

Find junction gene expression data.

- Returns median junction read counts in tissues of a given gene from all known transcripts.
- Results may be filtered by dataset or tissue.

By default, this service queries the latest GTEx release.

GTEx Portal API documentation

### Usage

```
get_median_junction_expression(
  gencodeIds,
  datasetId = "gtex_v8",
  tissueSiteDetailIds = NULL,
  page = 0,
  itemsPerPage = 250
)
```

#### **Arguments**

gencodeIds A character vector of Versioned GENCODE IDs, e.g. c("ENSG00000132693.12",

"ENSG00000203782.5").

datasetId String. Unique identifier of a dataset. Usually includes a data source and data

release. Options: "gtex\_v8", "gtex\_snrnaseq\_pilot".

tissueSiteDetailIds

Character vector of IDs for tissues of interest. Can be GTEx specific IDs (e.g. "Whole\_Blood"; use get\_tissue\_site\_detail() to see valid values) or On-

tology IDs.

page Integer (default = 0).

itemsPerPage Integer (default = 250).

#### Value

A tibble.

### See Also

```
Other Expression Data Endpoints: get_clustered_median_exon_expression(), get_clustered_median_gene_expression(), get_clustered_median_junction_expression(), get_clustered_median_transcript_expression(), get_expression_pca(), get_gene_expression(), get_median_exon_expression(), get_median_gene_expression() get_median_transcript_expression(), get_single_nucleus_gex(), get_single_nucleus_gex_summary(), get_top_expressed_genes()
```

# Examples

```
## Not run:
get_median_junction_expression(gencodeIds = "ENSG000000132693.12")
## End(Not run)
```

### Description

Find median transcript expression data of all known transcripts of a gene.

- Returns median normalized expression in tissues of all known transcripts of a given gene.
- Results may be filtered by dataset or tissue.

By default, this service queries the latest GTEx release.

GTEx Portal API documentation

### Usage

```
get_median_transcript_expression(
  gencodeIds,
  datasetId = "gtex_v8",
  tissueSiteDetailIds = NULL,
  page = 0,
  itemsPerPage = 250
)
```

### **Arguments**

# Value

A tibble.

#### See Also

```
Other Expression Data Endpoints: get_clustered_median_exon_expression(), get_clustered_median_gene_expression(), get_clustered_median_junction_expression(), get_clustered_median_transcript_expression(), get_expression_pca(), get_gene_expression(), get_median_exon_expression(), get_median_gene_expression() get_median_junction_expression(), get_single_nucleus_gex(), get_single_nucleus_gex_summary(), get_top_expressed_genes()
```

### **Examples**

```
## Not run:
    get_median_transcript_expression(gencodeIds = "ENSG00000132693.12")

## End(Not run)

get_multi_tissue_eqtls
```

Get Multi Tissue Eqtls

# **Description**

Find multi-tissue eQTL Metasoft results.

- This service returns multi-tissue eQTL Metasoft results for a given gene and variant in a specified dataset.
- A Versioned GENCODE ID must be provided.
- For each tissue, the results include: m-value (mValue), normalized effect size (nes), p-value (pValue), and standard error (se).
- The m-value is the posterior probability that an eQTL effect exists in each tissue tested in the cross-tissue meta-analysis (Han and Eskin, PLoS Genetics 8(3): e1002555, 2012).
- The normalized effect size is the slope of the linear regression of normalized expression data versus the three genotype categories using single-tissue eQTL analysis, representing eQTL effect size.
- The p-value is from a t-test that compares observed NES from single-tissue eQTL analysis to a null NES of 0.

By default, the service queries the latest GTEx release. The retrieved data is split into pages with items\_per\_page entries per page

GTEx Portal API documentation

```
get_multi_tissue_eqtls(
  gencodeIds,
  variantId = NULL,
  datasetId = "gtex_v8",
  page = 0,
  itemsPerPage = 250
)
```

get\_neighbor\_gene 45

### **Arguments**

gencodeIds A character vector of Versioned GENCODE IDs, e.g. c("ENSG00000132693.12",

"ENSG00000203782.5").

variantId String. A gtex variant ID.

datasetId String. Unique identifier of a dataset. Usually includes a data source and data

release. Options: "gtex\_v8", "gtex\_snrnaseq\_pilot".

page Integer (default = 0).

itemsPerPage Integer (default = 250).

#### Value

A tibble.

#### See Also

```
Other Static Association Endpoints: get_eqtl_genes(), get_fine_mapping(), get_independent_eqtl(), get_significant_single_tissue_eqtls(), get_significant_single_tissue_eqtls_by_location(), get_significant_single_tissue_isqtls(), get_significant_si
```

# **Examples**

get\_neighbor\_gene

Get Neighbor Gene

### **Description**

Find all neighboring genes on a certain chromosome around a position with a certain window size.

GTEx API Portal documentation

```
get_neighbor_gene(pos, chromosome, bp_window, page = 0, itemsPerPage = 250)
```

get\_news\_item

### **Arguments**

pos Integer, vector.

chromosome String. One of "chr1", "chr2", "chr3", "chr4", "chr5", "chr6", "chr7", "chr8",

"chr9", "chr10", "chr11", "chr12", "chr13", "chr14", "chr15", "chr16", "chr17",

"chr18", "chr19", "chr20", "chr21", "chr22", "chrM", "chrX", "chrY".

bp\_window Integer.

page Integer (default = 0). itemsPerPage Integer (default = 250).

#### Value

A tibble.

### See Also

```
Other Reference Genome Endpoints: get_exons(), get_gene_search(), get_genes(), get_genomic_features(), get_gwas_catalog_by_location(), get_transcripts()
```

# **Examples**

```
## Not run:
get_neighbor_gene(pos = 1000000, chromosome = "chr1", bp_window = 10000)
## End(Not run)
```

get\_news\_item

Get News Item

# **Description**

Getting all the news items from the database that are current.

GTEx Portal API documentation.

# Usage

```
get_news_item(page = 0, itemsPerPage = 250)
```

# **Arguments**

```
page Integer (default = 0).
itemsPerPage Integer (default = 250).
```

### Value

A tibble.

### See Also

```
Other Admin Endpoints: get_maintenance_message()
```

# **Examples**

### **Description**

GTEx Portal API documentation

#### Usage

```
get_sample_biobank_data(
  draw = NULL,
  materialTypes = NULL,
  tissueSiteDetailIds = NULL,
  pathCategory = NULL,
  tissueSampleIds = NULL,
  sex = NULL,
  sortBy = "sampleId",
  sortDirection = "asc",
  searchTerm = NULL,
  sampleIds = NULL,
  subjectIds = NULL,
  ageBrackets = NULL,
  hardyScales = NULL,
  hasExpressionData = NULL,
  hasGenotype = NULL,
  page = 0,
  itemsPerPage = 250
)
```

### **Arguments**

```
draw Integer.

materialTypes String, vector. Options: "Cells:Cell Line Viable", "DNA:DNA Genomic", "DNA:DNA Somatic", "RNA:Total RNA", "Tissue:PAXgene Preserved", "Tissue:PAXgene Preserved Paraffin-embedded", "Tissue:Fresh Frozen Tissue".
```

#### tissueSiteDetailIds

Character vector of IDs for tissues of interest. Can be GTEx specific IDs (e.g. "Whole\_Blood"; use get\_tissue\_site\_detail() to see valid values) or Ontology IDs.

pathCategory

Character vector. Options: "adenoma", "amylacea", "atelectasis", "atherosclerosis", "atherosis", "atrophy", "calcification", "cirrhosis", "clean\_specimens", "congestion", "corpora\_albicantia", "cyst", "desquamation", "diabetic", "dysplasia", "edema", "emphysema", "esophagitis", "fibrosis", "gastritis", "glomerulosclerosis", "goiter", "gynecomastoid", "hashimoto", "heart\_failure\_cells", "hemorrhage", "hepatitis", "hyalinization", "hypereosinophilia", "hyperplasia", "hypertrophy", "hypoxic", "infarction", "inflammation", "ischemic\_changes", "macrophages", "mastopathy", "metaplasia", "monckeberg", "necrosis", "nephritis", "nephrosclerosis", "no\_abnormalities", "nodularity", "pancreatitis", "pigment", "pneumonia", "post\_menopausal", "prostatitis", "saponification", "scarring", "sclerotic", "solar\_elastosis", "spermatogenesis", "steatosis", "sweat\_glands", "tma".

### tissueSampleIds

Array of strings. A list of Tissue Sample ID(s).

sex String. Options: "male", "female".

sortBy String. Options: "sampleId", "ischemicTime", "aliquotId", "tissueSampleId",

"hardyScale", "pathologyNotes", "ageBracket", "tissueSiteDetailId", "sex".

sortDirection String. Options: "asc", "desc". Default = "asc".

searchTerm String.

sampleIds Character vector. GTEx sample ID.
subjectIds Character vector. GTEx subject ID.

ageBrackets The age bracket(s) of the donors of interest. Options: "20-29", "30-39", "40-49",

"50-59", "60-69", "70-79".

hardyScales Character vector. A list of Hardy Scale(s) of interest. Options: "Ventilator case",

"Fast death - violent", "Fast death - natural causes", "Intermediate death", "Slow

death".

hasExpressionData

Logical.

hasGenotype Logical.

page Integer (default = 0).

itemsPerPage Integer (default = 250).

#### Value

A tibble

#### See Also

Other Biobank Data Endpoints: download()

### **Examples**

# Description

This service returns information of samples used in analyses from all datasets. Results may be filtered by dataset ID, sample ID, subject ID, sample metadata, or other provided parameters. By default, this service queries the latest GTEx release.

GTEx Portal API documentation

#### Usage

```
get_sample_datasets_endpoints(
  datasetId = "gtex_v8",
  sampleIds = NULL,
  tissueSampleIds = NULL,
  subjectIds = NULL,
  ageBrackets = NULL,
  sex = NULL,
  pathCategory = NULL,
  tissueSiteDetailId = NULL,
  aliquotIds = NULL,
  autolysisScores = NULL,
  hardyScales = NULL,
  ischemicTime = NULL,
  ischemicTimeGroups = NULL,
  rin = NULL,
  uberonIds = NULL,
  dataTypes = NULL,
  sortBy = NULL,
  sortDirection = NULL,
  page = NULL,
  itemsPerPage = NULL
)
```

#### **Arguments**

datasetId

String. Unique identifier of a dataset. Usually includes a data source and data release. Options: "gtex\_v8", "gtex\_snrnaseq\_pilot".

sampleIds Character vector. GTEx sample ID.

tissueSampleIds

Array of strings. A list of Tissue Sample ID(s).

subjectIds Character vector. GTEx subject ID.

ageBrackets The age bracket(s) of the donors of interest. Options: "20-29", "30-39", "40-49",

"50-59", "60-69", "70-79".

sex String. Options: "male", "female".

pathCategory Character vector. Options: "adenoma", "amylacea", "atelectasis", "atheroscle-

rosis", "atherosis", "atrophy", "calcification", "cirrhosis", "clean\_specimens", "congestion", "corpora\_albicantia", "cyst", "desquamation", "diabetic", "dysplasia", "edema", "emphysema", "esophagitis", "fibrosis", "gastritis", "glomerulosclerosis", "goiter", "gynecomastoid", "hashimoto", "heart\_failure\_cells", "hemorrhage", "hepatitis", "hyalinization", "hypereosinophilia", "hyperplasia", "hy-

pertrophy", "hypoxic", "infarction", "inflammation", "ischemic\_changes", "macrophages",

"mastopathy", "metaplasia", "monckeberg", "necrosis", "nephritis", "nephrosclerosis", "no\_abnormalities", "nodularity", "pancreatitis", "pigment", "pneumonia", "post\_menopausal", "prostatitis", "saponification", "scarring", "sclerotic", "solar\_elastosis", "spermatogenesis", "steatosis", "sweat\_glands", "tma".

tissueSiteDetailId

String. The ID of the tissue of interest. Can be a GTEx specific ID (e.g. "Whole\_Blood"; use get\_tissue\_site\_detail() to see valid values) or an

Ontology ID.

aliquotIds Character vector.

autolysisScores

Character vector. Options: "None", "Mild", "Moderate", "Severe".

hardyScales Character vector. A list of Hardy Scale(s) of interest. Options: "Ventilator case",

"Fast death - violent", "Fast death - natural causes", "Intermediate death", "Slow

death".

ischemicTime Integer.
ischemicTimeGroups

Character vector. Options: "<= 0", "1 - 300", "301 - 600", "601 - 900", "901 -

1200", "1201 - 1500", "> 1500".

rin Integer vector.

uberonIds Character vector of Uberon IDs (e.g. "UBERON:EFO\_0000572"; use get\_tissue\_site\_detail()

to see valid values).

dataTypes Character vector. Options: "RNASEQ", "WGS", "WES", "OMNI", "EXCLUDE".

sortBy String. Options: "sampleId", "ischemicTime", "aliquotId", "tissueSampleId",

"hardyScale", "pathologyNotes", "ageBracket", "tissueSiteDetailId", "sex".

sortDirection String. Options: "asc", "desc". Default = "asc".

page Integer (default = 0). itemsPerPage Integer (default = 250).

### Value

A tibble

get\_service\_info 51

# See Also

```
Other Datasets Endpoints: get_annotation(), get_collapsed_gene_model_exon(), get_downloads_page_data(), get_file_list(), get_full_get_collapsed_gene_model_exon(), get_functional_annotation(), get_linkage_disequilibrium_by_variant_data(), get_linkage_disequilibrium_data(), get_subject(), get_tissue_site_detail(), get_variant(), get_variant_by_location()
```

# **Examples**

```
## Not run:
   get_sample_datasets_endpoints()
## End(Not run)
```

get\_service\_info

Get Service Info

# **Description**

General information about the GTEx service.

GTEx Portal API documentation.

# Usage

```
get_service_info()
```

### Value

A tibble.

# **Examples**

```
## Not run:
get_service_info()
## End(Not run)
```

### **Description**

Find significant single tissue eQTLs.

- This service returns precomputed significant single tissue eQTLs.
- Results may be filtered by tissue, gene, variant or dataset.
- To search by gene, use the versioned GENCODE ID.
- To search by variant, use the dbSNP rs ID (snpId).

By default, the service queries the latest GTEx release and the retrieved data is split into pages with items\_per\_page entries per page

GTEx Portal API documentation.

#### Usage

```
get_significant_single_tissue_eqtls(
  gencodeIds = NULL,
  variantIds = NULL,
  tissueSiteDetailIds = NULL,
  datasetId = "gtex_v8",
  page = 0,
  itemsPerPage = 250
)
```

### **Arguments**

gencodeIds A character vector of Versioned GENCODE IDs, e.g. c("ENSG00000132693.12", "ENSG00000203782.5"). variantIds Character vector. Gtex variant IDs. tissueSiteDetailIds Character vector of IDs for tissues of interest. Can be GTEx specific IDs (e.g. "Whole\_Blood"; use get\_tissue\_site\_detail() to see valid values) or Ontology IDs. String. Unique identifier of a dataset. Usually includes a data source and data datasetId release. Options: "gtex\_v8", "gtex\_snrnaseq\_pilot". Integer (default = 0). page itemsPerPage Integer (default = 250).

#### **Details**

**Note:** although the GTEx Portal API documentation says to use the dbSNP rsID when searching by variant, this returns no results. Instead use gtex variant IDs e.g. use "chr1\_153209640\_C\_A\_b38" instead of "rs1410858".

#### Value

A tibble.

#### See Also

```
Other Static Association Endpoints: get_eqtl_genes(), get_fine_mapping(), get_independent_eqtl(), get_multi_tissue_eqtls(), get_significant_single_tissue_eqtls_by_location(), get_significant_single_t get_significant_single_tissue_isqtls(), get_significant_single_tissue_sqtls(), get_sqtl_genes()
```

### **Examples**

#### **Description**

Find significant single tissue eQTLs using Chromosomal Locations.

- This service returns precomputed significant single tissue eQTLs.
- Results may be filtered by tissue, and/or dataset.

By default, the service queries the latest GTEx release. Since this endpoint is used to support a third party program on the portal, the return structure is different from other endpoints and is not paginated.

GTEx Portal API documentation

### Usage

```
get_significant_single_tissue_eqtls_by_location(
  tissueSiteDetailId,
  start,
  end,
  chromosome,
  datasetId = "gtex_v8"
)
```

# **Arguments**

tissueSiteDetailId

String. The ID of the tissue of interest. Can be a GTEx specific ID (e.g. "Whole\_Blood"; use get\_tissue\_site\_detail() to see valid values) or an

Ontology ID.

start Integer. end Integer.

chromosome String. One of "chr1", "chr2", "chr3", "chr4", "chr5", "chr6", "chr7", "chr8",

"chr9", "chr10", "chr11", "chr12", "chr13", "chr14", "chr15", "chr16", "chr17", "chr17", "chr17", "chr18", "c

"chr18", "chr19", "chr20", "chr21", "chr22", "chrM", "chrX", "chrY".

datasetId String. Unique identifier of a dataset. Usually includes a data source and data

release. Options: "gtex\_v8", "gtex\_snrnaseq\_pilot".

### Value

A tibble.

### See Also

```
Other Static Association Endpoints: get_eqtl_genes(), get_fine_mapping(), get_independent_eqtl(), get_multi_tissue_eqtls(), get_significant_single_tissue_eqtls(), get_significant_single_tissue_ieqtls get_significant_single_tissue_isqtls(), get_significant_single_tissue_sqtls(), get_sqtl_genes()
```

# **Examples**

```
## Not run:
get_significant_single_tissue_eqtls_by_location(
   tissueSiteDetailId = "Artery_Aorta",
   start = 10000,
   end = 250000,
   chromosome = "chr11")
## End(Not run)
```

# **Description**

Retrieve Interaction eQTL Data.

- This service returns cell type interaction eQTLs (ieQTLs), from a specified dataset.
- Results may be filtered by tissue
- By default, the service queries the latest GTEx release.

The retrieved data is split into pages with items\_per\_page entries per page

GTEx Portal API documentation

### Usage

```
get_significant_single_tissue_ieqtls(
  gencodeIds,
  variantIds = NULL,
  tissueSiteDetailIds = NULL,
  datasetId = "gtex_v8",
  page = 0,
  itemsPerPage = 250
)
```

# **Arguments**

gencodeIds A character vector of Versioned GENCODE IDs, e.g. c("ENSG00000132693.12", "ENSG00000203782.5"). variantIds Character vector. Gtex variant IDs. tissueSiteDetailIds Character vector of IDs for tissues of interest. Can be GTEx specific IDs (e.g. "Whole\_Blood"; use get\_tissue\_site\_detail() to see valid values) or Ontology IDs. datasetId String. Unique identifier of a dataset. Usually includes a data source and data release. Options: "gtex\_v8", "gtex\_snrnaseq\_pilot". Integer (default = 0). page itemsPerPage Integer (default = 250).

#### Value

A tibble

### See Also

```
Other Static Association Endpoints: get_eqtl_genes(), get_fine_mapping(), get_independent_eqtl(), get_multi_tissue_eqtls(), get_significant_single_tissue_eqtls(), get_significant_single_tissue_eqtls_get_significant_single_tissue_isqtls(), get_significant_single_tissue_sqtls(), get_sqtl_genes()
```

# **Examples**

### **Description**

Retrieve Interaction sQTL Data.

- This service retrieves cell type interaction sQTLs (isQTLs), from a specified dataset.
- Results may be filtered by tissue
- By default, the service queries the latest GTEx release.

The retrieved data is split into pages with items\_per\_page entries per page

GTEx Portal API documentation.

#### Usage

```
get_significant_single_tissue_isqtls(
  gencodeIds,
  variantIds = NULL,
  tissueSiteDetailIds = NULL,
  datasetId = "gtex_v8",
  page = 0,
  itemsPerPage = 250
)
```

### **Arguments**

```
gencodeIds A character vector of Versioned GENCODE IDs, e.g. c("ENSG00000132693.12", "ENSG00000203782.5").

variantIds Character vector. Gtex variant IDs.
```

tissueSiteDetailIds

Character vector of IDs for tissues of interest. Can be GTEx specific IDs (e.g. "Whole\_Blood"; use get\_tissue\_site\_detail() to see valid values) or Ontology IDs

tology IDs.

datasetId String. Unique identifier of a dataset. Usually includes a data source and data

release. Options: "gtex\_v8", "gtex\_snrnaseq\_pilot".

page Integer (default = 0). itemsPerPage Integer (default = 250).

#### Value

A tibble

#### See Also

```
Other Static Association Endpoints: get_eqtl_genes(), get_fine_mapping(), get_independent_eqtl(), get_multi_tissue_eqtls(), get_significant_single_tissue_eqtls(), get_significant_single_tissue_eqtls_get_significant_single_tissue_ieqtls(), get_significant_single_tissue_sqtls(), get_sqtl_genes()
```

# **Examples**

# Description

Retrieve Single Tissue sQTL Data.

- This service returns single tissue sQTL data for the given genes, from a specified dataset.
- Results may be filtered by tissue
- By default, the service queries the latest GTEx release.

The retrieved data is split into pages with items\_per\_page entries per page

GTEx Portal API documentation.

### Usage

```
get_significant_single_tissue_sqtls(
  gencodeIds,
  variantIds = NULL,
  tissueSiteDetailIds = NULL,
  datasetId = "gtex_v8"
)
```

### **Arguments**

gencodeIds A character vector of Versioned GENCODE IDs, e.g. c("ENSG00000132693.12",

"ENSG00000203782.5").

variantIds Character vector. Gtex variant IDs.

tissueSiteDetailIds

Character vector of IDs for tissues of interest. Can be GTEx specific IDs (e.g. "Whole\_Blood"; use get\_tissue\_site\_detail() to see valid values) or On-

tology IDs.

datasetId String. Unique identifier of a dataset. Usually includes a data source and data

release. Options: "gtex\_v8", "gtex\_snrnaseq\_pilot".

### Value

A tibble

#### See Also

```
Other Static Association Endpoints: get_eqtl_genes(), get_fine_mapping(), get_independent_eqtl(), get_multi_tissue_eqtls(), get_significant_single_tissue_eqtls(), get_significant_single_tissue_eqtls_get_significant_single_tissue_isqtls(), get_sqtl_genes()
```

# **Examples**

# **Description**

Retrieve Single Nucleus Gene Expression Data for a given Gene.

GTEx Portal API documentation

### Usage

```
get_single_nucleus_gex(
  gencodeIds,
  datasetId = "gtex_snrnaseq_pilot",
  tissueSiteDetailIds = NULL,
  excludeDataArray = TRUE,
  page = 0,
  itemsPerPage = 250
)
```

# **Arguments**

gencodeIds A character vector of Versioned GENCODE IDs, e.g. c("ENSG00000132693.12",

"ENSG00000203782.5").

datasetId String. Unique identifier of a dataset. Usually includes a data source and data

release. Options: "gtex\_v8", "gtex\_snrnaseq\_pilot".

tissueSiteDetailIds

Character vector of IDs for tissues of interest. Can be GTEx specific IDs (e.g. "Whole\_Blood"; use get\_tissue\_site\_detail() to see valid values) or Ontology IDs.

excludeDataArray

String. Options are TRUE or FALSE

page Integer (default = 0). itemsPerPage Integer (default = 250).

#### Value

A tibble

#### See Also

```
Other Expression Data Endpoints: get_clustered_median_exon_expression(), get_clustered_median_gene_expression(), get_clustered_median_transcript_expression(), get_expression(), get_expression(), get_expression(), get_median_exon_expression(), get_median_gene_expression(), get_median_junction_expression(), get_median_transcript_expression(), get_single_nucleus_gex_summary(get_top_expressed_genes()
```

# **Examples**

### Description

Retrieve Summarized Single Nucleus Gene Expression Data.

GTEx Portal API documentation

# Usage

```
get_single_nucleus_gex_summary(
  datasetId = "gtex_snrnaseq_pilot",
  tissueSiteDetailIds = NULL,
  page = 0,
  itemsPerPage = 250
)
```

# Arguments

datasetId String. Unique identifier of a dataset. Usually includes a data source and data release. Options: "gtex\_v8", "gtex\_snrnaseq\_pilot".

tissueSiteDetailIds Character vector of IDs for tissues of interest. Can be GTEx specific IDs (e.g. "Whole\_Blood"; use get\_tissue\_site\_detail() to see valid values) or Ontology IDs.

page Integer (default = 0).

itemsPerPage Integer (default = 250).

### Value

A tibble.

get\_sqtl\_genes 61

### See Also

```
Other Expression Data Endpoints: get_clustered_median_exon_expression(), get_clustered_median_gene_expression(), get_clustered_median_junction_expression(), get_clustered_median_transcript_expression(), get_expression_pca(), get_gene_expression(), get_median_exon_expression(), get_median_gene_expression() get_median_junction_expression(), get_median_transcript_expression(), get_single_nucleus_gex(), get_top_expressed_genes()
```

### **Examples**

get\_sqtl\_genes

Get Sqtl Genes

### **Description**

Retrieve sGenes (sQTL Genes).

- This service returns sGenes (sQTL Genes) from the specified dataset.
- Results may be filtered by tissue.
- By default, the service queries the latest GTEx release.

The retrieved data is split into pages with items\_per\_page entries per page

GTEx Portal API documentation.

```
get_sqtl_genes(
  tissueSiteDetailId,
  datasetId = "gtex_v8",
  page = 0,
  itemsPerPage = 250
)
```

62 get\_subject

# **Arguments**

tissueSiteDetailId

String. The ID of the tissue of interest. Can be a GTEx specific ID (e.g. "Whole\_Blood"; use get\_tissue\_site\_detail() to see valid values) or an

Ontology ID.

datasetId String. Unique identifier of a dataset. Usually includes a data source and data

release. Options: "gtex\_v8", "gtex\_snrnaseq\_pilot".

page Integer (default = 0). itemsPerPage Integer (default = 250).

#### Value

A tibble.

#### See Also

```
Other Static Association Endpoints: get_eqtl_genes(), get_fine_mapping(), get_independent_eqtl(), get_multi_tissue_eqtls(), get_significant_single_tissue_eqtls(), get_significant_single_tissue_eqtls_get_significant_single_tissue_isqtls(), get_significant_single_tissue_isqtls(), get_single_tissue_isqtls(), get_single_tiss
```

### **Examples**

```
## Not run:
get_sqtl_genes("Whole_Blood")
## End(Not run)
```

get\_subject

Get Subject

### **Description**

This service returns information of subjects used in analyses from all datasets. Results may be filtered by dataset ID, subject ID, sex, age bracket or Hardy Scale. By default, this service queries the latest GTEx release.

GTEx Portal API documentation

```
get_subject(
  datasetId = "gtex_v8",
  sex = NULL,
  ageBrackets = NULL,
  hardyScales = NULL,
  subjectIds = NULL,
  page = 0,
  itemsPerPage = 250
)
```

get\_tissue\_site\_detail 63

### **Arguments**

datasetId String. Unique identifier of a dataset. Usually includes a data source and data

release. Options: "gtex\_v8", "gtex\_snrnaseq\_pilot".

sex String. Options: "male", "female".

ageBrackets The age bracket(s) of the donors of interest. Options: "20-29", "30-39", "40-49",

"50-59", "60-69", "70-79".

hardyScales Character vector. A list of Hardy Scale(s) of interest. Options: "Ventilator case",

"Fast death - violent", "Fast death - natural causes", "Intermediate death", "Slow

death".

subjectIds Character vector. GTEx subject ID.

page Integer (default = 0). itemsPerPage Integer (default = 250).

#### Value

A Tibble

#### See Also

```
Other Datasets Endpoints: get_annotation(), get_collapsed_gene_model_exon(), get_downloads_page_data(), get_file_list(), get_full_get_collapsed_gene_model_exon(), get_functional_annotation(), get_linkage_disequilibrium_by_variant_data(), get_linkage_disequilibrium_data(), get_sample_datasets_get_tissue_site_detail(), get_variant(), get_variant_by_location()
```

### **Examples**

```
## Not run:
   get_subject()
## End(Not run)
```

```
get_tissue_site_detail
```

Get Tissue Site Detail

# Description

Retrieve all tissue site detail information in the database

GTEx Portal API documentation

```
get_tissue_site_detail(page = 0, itemsPerPage = 250)
```

#### **Arguments**

```
page Integer (default = 0).
itemsPerPage Integer (default = 250).
```

#### Value

A tibble

#### See Also

```
Other Datasets Endpoints: get_annotation(), get_collapsed_gene_model_exon(), get_downloads_page_data(), get_file_list(), get_full_get_collapsed_gene_model_exon(), get_functional_annotation(), get_linkage_disequilibrium_by_variant_data(), get_linkage_disequilibrium_data(), get_sample_datasets_get_subject(), get_variant(), get_variant_by_location()
```

### **Examples**

```
## Not run:
    # returns a tibble with one row per tissue
    get_tissue_site_detail()

# `eqtlSampleSummary` and `rnaSeqSampleSummary` are list columns
bladder_site_details <- get_tissue_site_detail() |>
        dplyr::filter(tissueSiteDetailId == "Bladder")

purrr::pluck(bladder_site_details, "eqtlSampleSummary", 1)

purrr::pluck(bladder_site_details, "rnaSeqSampleSummary", 1)

## End(Not run)

get_top_expressed_genes
```

Get Top Expressed Genes

### **Description**

Find top expressed genes for a specified tissue.

- Returns top expressed genes for a specified tissue in a dataset, sorted by median expression.
- When the optional parameter filterMtGene is set to true, mitochondrial genes will be excluded from the results. By default, this service queries the latest GTEx release.

GTEx Portal API documentation

### **Usage**

```
get_top_expressed_genes(
   tissueSiteDetailId,
   datasetId = "gtex_v8",
   filterMtGene = TRUE,
   page = 0,
   itemsPerPage = 250
)
```

### **Arguments**

tissueSiteDetailId

String. The ID of the tissue of interest. Can be a GTEx specific ID (e.g. "Whole\_Blood"; use get\_tissue\_site\_detail() to see valid values) or an

Ontology ID.

datasetId String. Unique identifier of a dataset. Usually includes a data source and data

release. Options: "gtex\_v8", "gtex\_snrnaseq\_pilot".

filterMtGene Logical. Exclude mitochondrial genes.

page Integer (default = 0). itemsPerPage Integer (default = 250).

### Value

A tibble.

### See Also

```
Other Expression Data Endpoints: get_clustered_median_exon_expression(), get_clustered_median_gene_expression(), get_clustered_median_transcript_expression(), get_expression(), get_expression(), get_expression(), get_median_exon_expression(), get_median_gene_expression(), get_median_junction_expression(), get_median_transcript_expression(), get_single_nucleus_gex(), get_single_nucleus_gex_summary()
```

# **Examples**

```
## Not run:
get_top_expressed_genes(tissueSiteDetailId = "Artery_Aorta")
## End(Not run)
```

get\_transcripts

### **Description**

Find all transcripts of a reference gene.

- This service returns information about transcripts of the given versioned GENCODE ID.
- A genome build and GENCODE version must be provided.
- By default, this service queries the genome build and GENCODE version used by the latest GTEx release.

GTEx API Portal documentation

# Usage

```
get_transcripts(
  gencodeId,
  gencodeVersion = "v26",
  genomeBuild = "GRCh38/hg38",
  page = 0,
  itemsPerPage = 250
)
```

# **Arguments**

#### Value

A tibble.

# See Also

```
Other Reference Genome Endpoints: get_exons(), get_gene_search(), get_genes(), get_genomic_features(), get_gwas_catalog_by_location(), get_neighbor_gene()
```

### **Examples**

```
## Not run:
get_transcripts(gencodeId = "ENSG00000203782.5")
## End(Not run)
```

get\_variant 67

get\_variant Get Variant

### **Description**

This service returns information about a variant, including position, dbSNP RS ID, the reference allele, the alternative allele, and whether the minor allele frequency is >= 1%. For GTEx v6p, there is also information about whether the whole exome sequence and chip sequencing data are available. Results may be queried by GTEx variant ID (variantId), dbSNP RS ID (snpId) or genomic location (chromosome and pos). Variants are identified based on the genotype data of each dataset cohort, namely, are dataset-dependent. Each variant is assigned a unique GTEx variant ID (i.e. the primary key). Not all variants have a mappable dbSNP RS ID. By default, this service queries the latest GTEx release.

GTEx Portal API documentation

#### **Usage**

```
get_variant(
   snpId = NULL,
   variantId = NULL,
   chromosome = NULL,
   pos = NULL,
   datasetId = "gtex_v8",
   page = 0,
   itemsPerPage = 250
)
```

#### **Arguments**

snpId String

variantId String. A gtex variant ID.

chromosome String. One of "chr1", "chr2", "chr3", "chr4", "chr5", "chr6", "chr7", "chr8", "chr9", "chr10", "chr11", "chr12", "chr13", "chr14", "chr15", "chr16", "chr17", "chr18", "chr19", "chr20", "chr21", "chr22", "chrM", "chrX", "chrY".

pos Integer, vector.

datasetId String. Unique identifier of a dataset. Usually includes a data source and data release. Options: "gtex\_v8", "gtex\_snrnaseq\_pilot".

page Integer (default = 0). itemsPerPage Integer (default = 250).

#### Value

A tibble.

### See Also

```
Other Datasets Endpoints: get_annotation(), get_collapsed_gene_model_exon(), get_downloads_page_data(), get_file_list(), get_full_get_collapsed_gene_model_exon(), get_functional_annotation(), get_linkage_disequilibrium_by_variant_data(), get_linkage_disequilibrium_data(), get_sample_datasets_get_subject(), get_tissue_site_detail(), get_variant_by_location()
```

# **Examples**

```
get_variant_by_location
```

Get Variant By Location

# **Description**

This service allows the user to query information about variants on a certain chromosome at a certain location.

GTEx Portal API documentation

# Usage

```
get_variant_by_location(
   start,
   end,
   chromosome,
   sortBy = "pos",
   sortDirection = "asc",
   page = 0,
   itemsPerPage = 250
)
```

#### **Arguments**

```
\label{eq:start} \begin{array}{ll} & \text{Integer.} \\ & \text{end} & \text{Integer.} \\ & \text{chromosome} & \text{String. One of "chr1", "chr2", "chr3", "chr4", "chr5", "chr6", "chr7", "chr8", "chr9", "chr10", "chr11", "chr12", "chr13", "chr14", "chr15", "chr16", "chr17", "chr18", "chr19", "chr20", "chr21", "chr22", "chrM", "chrX", "chrY".} \\ \end{array}
```

```
sortBy String. Options: "sampleId", "ischemicTime", "aliquotId", "tissueSampleId",
```

"hardyScale", "pathologyNotes", "ageBracket", "tissueSiteDetailId", "sex".

sortDirection String. Options: "asc", "desc". Default = "asc".

page Integer (default = 0). itemsPerPage Integer (default = 250).

### Value

A tibble.

# See Also

```
Other Datasets Endpoints: get_annotation(), get_collapsed_gene_model_exon(), get_downloads_page_data(), get_file_list(), get_full_get_collapsed_gene_model_exon(), get_functional_annotation(), get_linkage_disequilibrium_by_variant_data(), get_linkage_disequilibrium_data(), get_sample_datasets_get_subject(), get_tissue_site_detail(), get_variant()
```

# **Examples**

# **Index**

```
* Admin Endpoints
                                                    get_expression_pca, 23
    get_maintenance_message, 38
                                                    get_gene_expression, 30
    get_news_item, 46
                                                    get_median_exon_expression, 39
* Biobank Data Endpoints
                                                    get_median_gene_expression, 40
    download, 9
                                                    get_median_junction_expression, 41
    get_sample_biobank_data, 47
                                                    get_median_transcript_expression,
* Datasets Endpoints
    get_annotation, 11
                                                    get_single_nucleus_gex, 58
    get_collapsed_gene_model_exon, 17
                                                    get_single_nucleus_gex_summary, 60
    get_downloads_page_data, 19
                                                    get_top_expressed_genes, 64
                                                * GTEx Portal API Info
    get_file_list, 24
                                                    get_service_info, 51
    get_full_get_collapsed_gene_model_exon,
                                                * Histology Endpoints
        2.7
    get_functional_annotation, 28
                                                    get_image, 34
    get_linkage_disequilibrium_by_variant_data, Metadata Endpoints
                                                    get_dataset_info, 19
                                                * Reference Genome Endpoints
    get_linkage_disequilibrium_data,
                                                    get_exons, 22
                                                    {\tt get\_gene\_search}, {\tt 31}
    get_sample_datasets_endpoints, 49
    get_subject, 62
                                                    get_genes, 29
    get_tissue_site_detail, 63
                                                    get_genomic_features, 32
    get_variant, 67
                                                    get_gwas_catalog_by_location, 33
    get_variant_by_location, 68
                                                    get_neighbor_gene, 45
* Dynamic Association Endpoints
                                                    get_transcripts, 66
    calculate_expression_quantitative_trait_loStatic Association Endpoints
                                                    get_eqtl_genes, 21
                                                    {\tt get\_fine\_mapping}, {\tt 25}
    calculate_ieqtls, 5
    calculate_isqtls, 6
                                                    get_independent_eqt1, 35
    calculate_splicing_quantitative_trait_loci,
                                                    get_multi_tissue_eqtls, 44
                                                    get_significant_single_tissue_eqtls,
* Expression Data Endpoints
    get_clustered_median_exon_expression,
                                                    get_significant_single_tissue_eqtls_by_location,
    get_clustered_median_gene_expression,
                                                    get_significant_single_tissue_ieqtls,
        13
    get_clustered_median_junction_expression,
                                                    get_significant_single_tissue_isqtls,
    get_clustered_median_transcript_expression,
                                                    get_significant_single_tissue_sqtls,
        16
                                                        57
```

INDEX 71

<pre>get_sqtl_genes, 61</pre>	get_genomic_features, 23, 30, 32, 32, 34,
	46, 66
calculate_expression_quantitative_trait_loci	get_gwas_catalog_by_location, 23, 30, 32,
3, 6–8	33, 33, 46, 66
calculate_ieqtls, 4, 5, 7, 8	<pre>get_image, 34</pre>
calculate_isqtls, $4$ , $6$ , $6$ , $8$	get_independent_eqt1, 22, 26, 35, 45, 53,
<pre>calculate_splicing_quantitative_trait_loci,</pre>	54, 56–58, 62
4, 6, 7, 8	<pre>get_linkage_disequilibrium_by_variant_data,</pre>
1 1 10 40	11, 18, 20, 25, 27, 28, 36, 38, 51, 63,
download, 9, 48	64, 68, 69
11 10 20 25 27 20 27	<pre>get_linkage_disequilibrium_data, 11, 18,</pre>
get_annotation, 11, 18, 20, 25, 27, 28, 37,	20, 25, 27, 28, 37, 37, 51, 63, 64, 68,
38, 51, 63, 64, 68, 69	69
get_clustered_median_exon_expression,	<pre>get_maintenance_message, 38, 47</pre>
12, 14, 15, 17, 24, 31, 40–43, 59, 61,	<pre>get_median_exon_expression, 12, 14, 15,</pre>
65	17, 24, 31, 39, 41–43, 59, 61, 65
get_clustered_median_gene_expression,	<pre>get_median_gene_expression, 12, 14, 15,</pre>
12, 13, 15, 17, 24, 31, 40–43, 59, 61,	17, 24, 31, 40, 40, 42, 43, 59, 61, 65
65	get_median_junction_expression, 12, 14,
<pre>get_clustered_median_junction_expression,</pre>	15, 17, 24, 31, 40, 41, 41, 43, 59, 61,
12, 14, 14, 17, 24, 31, 40–43, 59, 61,	65
65	get_median_transcript_expression, <i>12</i> ,
<pre>get_clustered_median_transcript_expression,</pre>	14, 15, 17, 24, 31, 40–42, 43, 59, 61,
12, 14, 15, 16, 24, 31, 40–43, 59, 61,	65
65	get_multi_tissue_eqtls, 22, 26, 36, 44, 53,
<pre>get_collapsed_gene_model_exon, 11, 17,</pre>	
20, 25, 27, 28, 37, 38, 51, 63, 64, 68,	54, 56–58, 62
69	get_neighbor_gene, 23, 30, 32–34, 45, 66
get_dataset_info, 19	get_news_item, 39, 46
get_downloads_page_data, 11, 18, 19, 25,	get_sample_biobank_data, 10, 47
27, 28, 37, 38, 51, 63, 64, 68, 69	<pre>get_sample_datasets_endpoints, 11, 18,</pre>
get_eqtl_genes, 21, 26, 36, 45, 53, 54,	20, 25, 27, 28, 37, 38, 49, 63, 64, 68,
56–58, 62	69
get_exons, 22, 30, 32–34, 46, 66	<pre>get_service_info, 51</pre>
get_expression_pca, <i>12</i> , <i>14</i> , <i>15</i> , <i>17</i> , 23, <i>31</i> ,	<pre>get_significant_single_tissue_eqtls,</pre>
40–43, 59, 61, 65	22, 26, 36, 45, 52, 54, 56–58, 62
get_file_list, 11, 18, 20, 24, 27, 28, 37, 38,	<pre>get_significant_single_tissue_eqtls_by_location,</pre>
51, 63, 64, 68, 69	22, 26, 36, 45, 53, 53, 56–58, 62
get_fine_mapping, 22, 25, 36, 45, 53, 54,	<pre>get_significant_single_tissue_ieqtls,</pre>
56–58, 62	22, 26, 36, 45, 53, 54, 55, 57, 58, 62
<pre>get_full_get_collapsed_gene_model_exon,</pre>	<pre>get_significant_single_tissue_isqtls,</pre>
11, 18, 20, 25, 27, 28, 37, 38, 51, 63,	22, 26, 36, 45, 53, 54, 56, 56, 58, 62
64, 68, 69	<pre>get_significant_single_tissue_sqtls,</pre>
get_functional_annotation, 11, 18, 20, 25,	22, 26, 36, 45, 53, 54, 56, 57, 57, 62
27, 28, 37, 38, 51, 63, 64, 68, 69	get_single_nucleus_gex, 12, 14, 15, 17, 24,
get_gene_expression, <i>12</i> , <i>14</i> , <i>15</i> , <i>17</i> , <i>24</i> , 30,	31, 40–43, 58, 61, 65
40–43, 59, 61, 65	<pre>get_single_nucleus_gex_summary, 12, 14,</pre>
get_gene_search, 23, 30, 31, 33, 34, 46, 66	15, 17, 24, 31, 40–43, 59, 60, 65
get_genes, 23, 29, 32–34, 46, 66	get_sqtl_genes, 22, 26, 36, 45, 53, 54,

72 INDEX

```
\begin{array}{c} 56-58, 61 \\ \text{get\_subject}, 11, 18, 20, 25, 27, 28, 37, 38, \\ 51, 62, 64, 68, 69 \\ \text{get\_tissue\_site\_detail}, 11, 18, 20, 25, 27, \\ 28, 37, 38, 51, 63, 63, 68, 69 \\ \text{get\_tissue\_site\_detail}(), 3, 6-9, 12, 14, \\ 15, 17, 21, 24, 26, 30, 36, 39, 41-43, \\ 48, 50, 52, 54, 55, 57-60, 62, 65 \\ \text{get\_top\_expressed\_genes}, 12, 14, 15, 17, \\ 24, 31, 40-43, 59, 61, 64 \\ \text{get\_transcripts}, 23, 30, 32-34, 46, 66 \\ \text{get\_variant}, 11, 18, 20, 25, 27, 28, 37, 38, \\ 51, 63, 64, 67, 69 \\ \text{get\_variant\_by\_location}, 11, 18, 20, 25, \\ 27, 28, 37, 38, 51, 63, 64, 68, 68 \\ \end{array}
```