

Package ‘LDlinkR’

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Type Package

Title Calculating Linkage Disequilibrium (LD) in Human Population
Groups of Interest

Version 1.4.0

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Description Provides access to the 'LDlink' API (<<https://ldlink.nih.gov/?tab=apiaccess>>) using the R console. This programmatic access facilitates researchers who are interested in performing batch queries in 1000 Genomes Project (2015) <[doi:10.1038/nature15393](https://doi.org/10.1038/nature15393)> data using 'LDlink'. 'LDlink' is an interactive and powerful suite of web-based tools for querying germline variants in human population groups of interest. For more details, please see Machiela et al. (2015) <[doi:10.1093/bioinformatics/btv402](https://doi.org/10.1093/bioinformatics/btv402)>.

License GPL (>= 2)

URL <https://ldlink.nih.gov>

BugReports <https://github.com/CBIIT/LDlinkR/issues>

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Contents

LDexpress	2
LDhap	4
LDmatrix	5
LDpair	6
LDpop	7
LDproxy	8
LDproxy_batch	9
LDtrait	10
list_chips	11
list_gtex_tissues	12
list_pop	13
SNPchip	13
SNPclip	14
Index	16

LDexpress	<i>Determine if genomic variants are associated with gene expression.</i>
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Description

Search if a list of genomic variants (or variants in LD with those variants) is associated with gene expression in tissues of interest. Quantitative trait loci data is downloaded from the GTEx Portal (<https://gtexportal.org/home/>).

Usage

```
LDexpress(
  snps,
  pop = "CEU",
  tissue = "ALL",
  r2d = "r2",
  r2d_threshold = 0.1,
  p_threshold = 0.1,
  win_size = 5e+05,
  genome_build = "grch37",
  token = NULL,
  file = FALSE,
  api_root = "https://ldlink.nih.gov/LDlinkRest"
)
```

Arguments

snps	between 1 - 10 variants, using an rsID or chromosome coordinate (e.g. "chr7:24966446")
------	--

pop	a 1000 Genomes Project population, (e.g. YRI or CEU), multiple allowed, default = "CEU". Use the 'list_pop' function to see a list of available human reference populations.
tissue	select from 1 - 54 non-diseased tissue sites collected for the GTEx project, multiple allowed. Acceptable user input is taken either from "tissue_name_ldexpress" or "tissue_abbrev_ldexpress" (tissue abbreviation) code listed in available GTEx tissue sites using the list_getex_tissues() function (e.g. "ADI_SUB" for Adipose Subcutaneous). Input is case sensitive. Default = "ALL" for all available tissue types.
r2d	either "r2" for LD R2 or "d" for LD D', default = "r2".
r2d_threshold	R2 or D' (depends on 'r2d' user input parameter) threshold for LD filtering. Any variants within +/- of the specified genomic window and R ² or D' less than the threshold will be removed. Value needs to be in the range 0 to 1. Default value is 0.1.
p_threshold	define the eQTL significance threshold used for returning query results. Default value is 0.1 which returns all GTEx eQTL associations with P-value less than 0.1.
win_size	set genomic window size for LD calculation. Specify a value greater than or equal to zero and less than or equal to 1,000,000 basepairs (bp). Default value is +/- 500,000bp.
genome_build	Choose between one of the three options... 'grch37' for genome build GRCh37 (hg19), 'grch38' for GRCh38 (hg38), or 'grch38_high_coverage' for GRCh38 High Coverage (hg38) 1000 Genome Project data sets. Default is GRCh37 (hg19).
token	LDlink provided user token, default = NULL, register for token at https://ldlink.nih.gov/?tab=apiaccess
file	Optional character string naming a path and file for saving results. If file = FALSE, no file will be generated, default = FALSE.
api_root	Optional alternative root url for API.

Value

A data frame of all query variant RS numbers, respective QTL which are in LD with query variant, and associated gene expression.

Examples

```
## Not run: LDexpress(snps = c("rs345", "rs456"),
  pop = c("YRI", "CEU"),
  tissue = c("ADI_SUB", "ADI_VIS_OME"),
  r2d = "r2",
  r2d_threshold = "0.1",
  p_threshold = "0.1",
  win_size = "500000",
  genome_build = "grch37",
  token = Sys.getenv("LDLINK_TOKEN")
)
```

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

LDhap	<i>Calculates population specific haplotype frequencies of all haplotypes observed for a list of query variants.</i>
-------	--

Description

Calculates population specific haplotype frequencies of all haplotypes observed for a list of query variants.

Usage

```
LDhap(
  snps,
  pop = "CEU",
  token = NULL,
  file = FALSE,
  table_type = "haplotype",
  genome_build = "grch37",
  api_root = "https://ldlink.nih.gov/LDlinkRest"
)
```

Arguments

snps	list of between 1 - 30 variants, using an rsID or chromosome coordinate (e.g. "chr7:24966446")
pop	a 1000 Genomes Project population, (e.g. YRI or CEU), multiple allowed, default = "CEU"
token	LDlink provided user token, default = NULL, register for token at https://ldlink.nih.gov/?tab=apiaccess
file	Optional character string naming a path and file for saving results. If file = FALSE, no file will be generated, default = FALSE.
table_type	Choose from one of four options available to determine output format type... 'haplotype', 'variant', 'both' and 'merged'. Default = "haplotype".
genome_build	Choose between one of the three options... 'grch37' for genome build GRCh37 (hg19), 'grch38' for GRCh38 (hg38), or 'grch38_high_coverage' for GRCh38 High Coverage (hg38) 1000 Genome Project data sets. Default is GRCh37 (hg19).
api_root	Optional alternative root url for API.

Value

a data frame or list

Examples

```
## Not run: LDhap(c("rs3", "rs4", "rs148890987"), "CEU", token = Sys.getenv("LDLINK_TOKEN"))
## Not run: LDhap("rs148890987", c("YRI", "CEU"), token = Sys.getenv("LDLINK_TOKEN"))
```

LDmatrix	<i>Generates a data frame of pairwise linkage disequilibrium statistics.</i>
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Description

Generates a data frame of pairwise linkage disequilibrium statistics.

Usage

```
LDmatrix(
  snps,
  pop = "CEU",
  r2d = "r2",
  token = NULL,
  file = FALSE,
  genome_build = "grch37",
  api_root = "https://ldlink.nih.gov/LDlinkRest"
)
```

Arguments

snps	list of between 2 - 2500 variants, using an rsID or chromosome coordinate (e.g. "chr7:24966446")
pop	a 1000 Genomes Project population, (e.g. YRI or CEU), multiple allowed, default = "CEU"
r2d	r2d, either "r2" for LD R2 or "d" for LD D', default = "r2"
token	LDlink provided user token, default = NULL, register for token at https://ldlink.nih.gov/?tab=apiaccess
file	Optional character string naming a path and file for saving results. If file = FALSE, no file will be generated, default = FALSE.
genome_build	Choose between one of the three options... 'grch37' for genome build GRCh37 (hg19), 'grch38' for GRCh38 (hg38), or 'grch38_high_coverage' for GRCh38 High Coverage (hg38) 1000 Genome Project data sets. Default is GRCh37 (hg19).
api_root	Optional alternative root url for API.

Value

a data frame

Examples

```
## Not run: LDmatrix(c("rs3", "rs4", "rs148890987"),
                    "YRI", "r2",
                    token = Sys.getenv("LDLINK_TOKEN"))

## End(Not run)
```

LDpair

Investigates potentially correlated alleles for a pair of variants.

Description

Investigates potentially correlated alleles for a pair of variants.

Usage

```
LDpair(
  var1,
  var2,
  pop = "CEU",
  token = NULL,
  output = "table",
  file = FALSE,
  genome_build = "grch37",
  api_root = "https://ldlink.nih.gov/LDlinkRest"
)
```

Arguments

var1	the first RS number or genomic coordinate (e.g. "chr7:24966446")
var2	the second RS number or genomic coordinate (e.g. "ch7:24966446")
pop	a 1000 Genomes Project population(s), (e.g. YRI or CEU), multiple allowed, default = "CEU"
token	LDlink provided user token, default = NULL, register for token at https://ldlink.nih.gov/?tab=apiaccess
output	two output options available, "text", which displays a two-by-two matrix displaying haplotype counts and allele frequencies along with other statistics, or "table", which displays the same data in rows and columns, default = "table"
file	Optional character string naming a path and file for saving results. If file = FALSE, no file will be generated, default = FALSE.
genome_build	Choose between one of the three options... 'grch37' for genome build GRCh37 (hg19), 'grch38' for GRCh38 (hg38), or 'grch38_high_coverage' for GRCh38 High Coverage (hg38) 1000 Genome Project data sets. Default is GRCh37 (hg19).
api_root	Optional alternative root url for API.

Value

text or data frame, depending on the output option

Examples

```
## Not run: LDpair(var1 = "rs3", var2 = "rs4", pop = "YRI", token = Sys.getenv("LDLINK_TOKEN"))
## Not run: LDpair("rs3", "rs4", "YRI", token = Sys.getenv("LDLINK_TOKEN"), "text")
```

LDpop	<i>Investigates allele frequencies and linkage disequilibrium patterns across 1000 Genomes Project populations.</i>
-------	---

Description

Investigates allele frequencies and linkage disequilibrium patterns across 1000 Genomes Project populations.

Usage

```
LDpop(
  var1,
  var2,
  pop = "CEU",
  r2d = "r2",
  token = NULL,
  file = FALSE,
  genome_build = "grch37",
  api_root = "https://ldlink.nih.gov/LDlinkRest"
)
```

Arguments

var1	the first RS number or genomic coordinate (e.g. "chr7:24966446")
var2	the second RS number or genomic coordinate (e.g. "ch7:24966446")
pop	a 1000 Genomes Project population(s), (e.g. YRI or CEU), multiple allowed, default = "CEU"
r2d	either "r2" for LD R2 or "d" for LD D', default = "r2"
token	LDlink provided user token, default = NULL, register for token at https://ldlink.nih.gov/?tab=apiaccess
file	Optional character string naming a path and file for saving results. If file = FALSE, no file will be generated, default = FALSE.
genome_build	Choose between one of the three options... 'grch37' for genome build GRCh37 (hg19), 'grch38' for GRCh38 (hg38), or 'grch38_high_coverage' for GRCh38 High Coverage (hg38) 1000 Genome Project data sets. Default is GRCh37 (hg19).
api_root	Optional alternative root url for API.

Value

a data frame

Examples

```
## Not run: LDpop(var1 = "rs3", var2 = "rs4",
                 pop = "YRI", r2d = "r2",
                 token = Sys.getenv("LDLINK_TOKEN"))

## End(Not run)
```

LDproxy

Explore proxy and putative functional variants for a single query variant.

Description

Explore proxy and putative functional variants for a single query variant.

Usage

```
LDproxy(
  snp,
  pop = "CEU",
  r2d = "r2",
  token = NULL,
  file = FALSE,
  genome_build = "grch37",
  win_size = "500000",
  api_root = "https://ldlink.nih.gov/LDlinkRest"
)
```

Arguments

snp	an rsID or chromosome coordinate (e.g. "chr7:24966446"), one per query
pop	a 1000 Genomes Project population, (e.g. YRI or CEU), multiple allowed, default = "CEU"
r2d	either "r2" for LD R2 or "d" for LD D', default = "r2"
token	LDlink provided user token, default = NULL, register for token at https://ldlink.nih.gov/?tab=apiaccess
file	Optional character string naming a path and file for saving results. If file = FALSE, no file will be generated, default = FALSE.
genome_build	Choose between one of the three options... 'grch37' for genome build GRCh37 (hg19), 'grch38' for GRCh38 (hg38), or 'grch38_high_coverage' for GRCh38 High Coverage (hg38) 1000 Genome Project data sets. Default is GRCh37 (hg19).

win_size	set base pair (bp) window size. Specify a value greater than or equal to zero and less than or equal to 1,000,000bp. Default value is 500,000bp.
api_root	Optional alternative root url for API.

Value

a data frame

Examples

```
## Not run: LDproxy("rs456", "YRI", "r2", token = Sys.getenv("LDLINK_TOKEN"))
```

LDproxy_batch	<i>Query LDproxy using a list of query variants, one per line.</i>
---------------	--

Description

Query LDproxy using a list of query variants, one per line.

Usage

```
LDproxy_batch(
  snp,
  pop = "CEU",
  r2d = "r2",
  token = NULL,
  append = FALSE,
  genome_build = "grch37",
  win_size = "500000",
  api_root = "https://ldlink.nih.gov/LDlinkRest"
)
```

Arguments

snp	a character string or data frame listing rsID's or chromosome coordinates (e.g. "chr7:24966446"), one per line
pop	a 1000 Genomes Project population, (e.g. YRI or CEU), multiple allowed, default = "CEU"
r2d	either "r2" for LD R2 or "d" for LD D', default = "r2"
token	LDlink provided user token, default = NULL, register for token at https://ldlink.nih.gov/?tab=apiaccess
append	A logical. If TRUE, output for each query variant is appended to a text file. If FALSE, output of each query variant is saved in its own text file. Default is FALSE.

genome_build	Choose between one of the three options...‘grch37’ for genome build GRCh37 (hg19), ‘grch38’ for GRCh38 (hg38), or ‘grch38_high_coverage’ for GRCh38 High Coverage (hg38) 1000 Genome Project data sets. Default is GRCh37 (hg19).
win_size	set base pair (bp) window size. Specify a value greater than or equal to zero and less than or equal to 1,000,000bp. Default value is 500,000bp.
api_root	Optional alternative root url for API.

Value

text file(s) are saved to the current working directory.

Examples

```
## Not run: snps_to_upload <- c("rs3", "rs4")
## Not run: LDproxy_batch(snp = snps_to_upload, token = Sys.getenv("LDLINK_TOKEN"), append = FALSE)
```

LDtrait	<i>Determine if genomic variants are associated with a trait or disease.</i>
---------	--

Description

Search if a list of variants (or variants in LD with those variants) have been previously associated with a trait or disease. Trait and disease data is updated nightly from the GWAS Catalog (<https://www.ebi.ac.uk/gwas/docs/file-downloads>).

Usage

```
LDtrait(
  snps,
  pop = "CEU",
  r2d = "r2",
  r2d_threshold = 0.1,
  win_size = 5e+05,
  token = NULL,
  file = FALSE,
  genome_build = "grch37",
  api_root = "https://ldlink.nih.gov/LDlinkRest"
)
```

Arguments

snps	between 1 - 50 variants, using an rsID or chromosome coordinate (e.g. "chr7:24966446"). All input variants must match a bi-allelic variant.
pop	a 1000 Genomes Project population, (e.g. YRI or CEU), multiple allowed, default = "CEU". Use the ‘list_pop’ function to see a list of available human reference populations.

r2d	use "r2" to filter desired output from a threshold based on estimated LD R2 (R squared) or "d" for LD D' (D-prime), default = "r2".
r2d_threshold	R2 or D' (depends on 'r2d' user input parameter) threshold for LD filtering. Any variants within +/- of the specified genomic window and R^2 or D' less than the threshold will be removed. Value needs to be in the range 0 to 1. Default value is 0.1.
win_size	set genomic window size for LD calculation. Specify a value greater than or equal to zero and less than or equal to 1,000,000bp. Default value is +/- 500,000 bp.
token	LDlink provided user token, default = NULL, register for token at https://ldlink.nih.gov/?tab=apiaccess
file	Optional character string naming a path and file for saving results. If file = FALSE, no file will be generated, default = FALSE.
genome_build	Choose between one of the three options... 'grch37' for genome build GRCh37 (hg19), 'grch38' for GRCh38 (hg38), or 'grch38_high_coverage' for GRCh38 High Coverage (hg38) 1000 Genome Project data sets. Default is GRCh37 (hg19).
api_root	Optional alternative root url for API.

Value

A data frame of all query variant RS numbers with a list of queried variants in LD with a variant reported in the GWAS Catalog (<https://www.ebi.ac.uk/gwas/docs/file-downloads>).

Examples

```
## Not run: LDtrait(snps = "rs456",
  pop = c("YRI", "CEU"),
  r2d = "r2",
  r2d_threshold = "0.1",
  win_size = "500000",
  token = Sys.getenv("LDLINK_TOKEN")
)

## End(Not run)
```

list_chips	<i>Provides a data frame listing the names and abbreviation codes for available commercial SNP Chip Arrays from Illumina and Affymetrix.</i>
------------	--

Description

Provides a data frame listing the names and abbreviation codes for available commercial SNP Chip Arrays from Illumina and Affymetrix.

Usage

```
list_chips()
```

Value

a data frame listing the names and abbreviation codes for available SNP Chip Arrays from Illumina and Affymetrix

Examples

```
list_chips()
```

list_gtex_tissues	<i>Provides a data frame listing the GTEx full names, 'LDexpress' full names (without spaces) and acceptable abbreviation codes of the 54 non-diseased tissue sites collected for the GTEx Portal and used as input for the 'LDexpress' function.</i>
-------------------	---

Description

Provides a data frame listing the GTEx full names, 'LDexpress' full names (without spaces) and acceptable abbreviation codes of the 54 non-diseased tissue sites collected for the GTEx Portal and used as input for the 'LDexpress' function.

Usage

```
list_gtex_tissues()
```

Value

a data frame listing the GTEx tissues, their names and abbreviation codes used as input for LDexpress.

Examples

```
list_gtex_tissues()
```

list_pop	<i>Provides a data frame listing the available reference populations from the 1000 Genomes Project.</i>
----------	---

Description

Provides a data frame listing the available reference populations from the 1000 Genomes Project.

Usage

```
list_pop()
```

Value

a data frame listing the available reference populations, continental (ex: European, African, and Admixed American) and sub-populations (ex: Finnish, Gambian, and Peruvian)

Examples

```
list_pop()
```

SNPchip	<i>Find commercial genotyping chip arrays for variants of interest.</i>
---------	---

Description

Find commercial genotyping chip arrays for variants of interest.

Usage

```
SNPchip(  
  snps,  
  chip = "ALL",  
  token = NULL,  
  file = FALSE,  
  genome_build = "grch37",  
  api_root = "https://ldlink.nih.gov/LDlinkRest"  
)
```

Arguments

snp	between 1 - 5,000 variants, using an rsID or chromosome coordinate (e.g. "chr7:24966446")
chip	chip or arrays, platform code(s) for a SNP chip array, ALL_Illumina, ALL_Affy or ALL, default=ALL
token	LDlink provided user token, default = NULL, register for token at https://ldlink.nih.gov/?tab=apiaccess
file	Optional character string naming a path and file for saving results. If file = FALSE, no file will be generated, default = FALSE.
genome_build	Choose between one of the three options... 'grch37' for genome build GRCh37 (hg19), 'grch38' for GRCh38 (hg38), or 'grch38_high_coverage' for GRCh38 High Coverage (hg38) 1000 Genome Project data sets. Default is GRCh37 (hg19).
api_root	Optional alternative root url for API.

Value

a data frame

Examples

```
## Not run: SNPclip(c("rs3", "rs4", "rs148890987"), "ALL",
  token = Sys.getenv("LDLINK_TOKEN"))

## End(Not run)
## Not run: SNPclip(c("rs3", "rs4", "rs148890987"),
  c("A_CHB2", "A_SNP5.0"),
  token = Sys.getenv("LDLINK_TOKEN"))

## End(Not run)
## Not run: SNPclip("rs148890987", "ALL_Affy", token = Sys.getenv("LDLINK_TOKEN"))
```

SNPclip

Prune a list of variants by linkage disequilibrium.

Description

Prune a list of variants by linkage disequilibrium.

Usage

```
SNPclip(
  snps,
  pop = "CEU",
  r2_threshold = "0.1",
  maf_threshold = "0.01",
```

```

    token = NULL,
    file = FALSE,
    genome_build = "grch37",
    api_root = "https://ldlink.nih.gov/LDlinkRest"
  )

```

Arguments

snps	a list of between 1 - 5,000 variants, using an rsID or chromosome coordinate (e.g. "chr7:24966446")
pop	a 1000 Genomes Project population, (e.g. YRI or CEU), multiple allowed, default = "CEU"
r2_threshold	LD R2 threshold between 0-1, default = 0.1
maf_threshold	minor allele frequency threshold between 0-1, default = 0.01
token	LDlink provided user token, default = NULL, register for token at https://ldlink.nih.gov/?tab=apiaccess
file	Optional character string naming a path and file for saving results. If file = FALSE, no file will be generated, default = FALSE.
genome_build	Choose between one of the three options... 'grch37' for genome build GRCh37 (hg19), 'grch38' for GRCh38 (hg38), or 'grch38_high_coverage' for GRCh38 High Coverage (hg38) 1000 Genome Project data sets. Default is GRCh37 (hg19).
api_root	Optional alternative root url for API.

Value

a data frame

Examples

```

## Not run: SNPclip(c("rs3", "rs4", "rs148890987"), "YRI", "0.1", "0.01",
                    token = Sys.getenv("LDLINK_TOKEN"))

## End(Not run)

```

Index

LDexpress, [2](#)
LDhap, [4](#)
LDmatrix, [5](#)
LDpair, [6](#)
LDpop, [7](#)
LDproxy, [8](#)
LDproxy_batch, [9](#)
LDtrait, [10](#)
list_chips, [11](#)
list_gtex_tissues, [12](#)
list_pop, [13](#)

SNPchip, [13](#)
SNPclip, [14](#)