# Package 'hgnc'

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Type Package

**Title** Download and Import the HUGO Gene Nomenclature Committee ('HGNC') Data Set into R

Version 0.1.2

**Description** A set of routines to quickly download and import the 'HGNC' data set on mapping of gene symbols to gene entries in other popular databases or resources.

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URL https://github.com/maialab/hgnc, https://maialab.org/hgnc/

#### BugReports https://github.com/maialab/hgnc/issues

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download\_archive Download HGNC dataset

#### Description

Download the latest HGNC approved data set.

#### Usage

```
download_archive(
    url = latest_archive_url(),
    path = getwd(),
    filename = basename(url),
    ...
)
```

#### Arguments

url	A character string naming the URL of the HGNC dataset. It defaults to the latest available archive.
path	A character string with the directory path where the downloaded file is to be saved. By default, this is the current working directory.
filename	A character string with the name of the saved file. By default, this is inferred from the last part of the URL.
	Additional arguments passed on to download.file().

filter\_by\_keyword Filter genes by keyword

#### Description

Filter the HGNC data set by a keyword to be looked up in the columns containing gene names or symbols. By default, it will look up in symbol, name, alias\_symbol, alias\_name, prev\_symbol and prev\_name. Note that this function in dive into list-columns and match return a hit result if at least one of the strings matches the keyword.

#### Usage

```
filter_by_keyword(
   tbl,
   keyword,
   cols = c("symbol", "name", "alias_symbol", "alias_name", "prev_symbol", "prev_name")
)
```

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#### Arguments

tbl	A tibble containing the HGNC data set, typically obtained with import_hgnc_dataset().
keyword	A keyword or a regular expression to be used as search criterion.
cols	Columns to be looked up.

#### Value

A tibble of the HGNC data set filtered by observations matching the keyword.

#### Examples

```
## Not run:
# Start by retrieving the HGNC data set
hgnc_tbl <- import_hgnc_dataset()</pre>
# Search for entries containing "TP53" in the HGNC data set
hgnc_tbl %>%
  filter_by_keyword('TP53') %>%
  dplyr::select(1:4)
# The same as above but restrict the search to the `symbol` column
hgnc_tbl %>%
  filter_by_keyword('TP53', cols = 'symbol') %>%
  dplyr::select(1:4)
# Match "TP53" exactly in the `symbol` column
hgnc_tbl %>%
  filter_by_keyword('^TP53$', cols = 'symbol') %>%
  dplyr::select(1:4)
# `filter_by_keyword()` is vectorised over `keyword`
hgnc_tbl %>%
  filter_by_keyword(c('^TP53$', '^PIK3CA$'), cols = 'symbol') %>%
  dplyr::select(1:4)
## End(Not run)
```

import\_hgnc\_dataset Import HGNC data

#### Description

This function imports into memory, as a tibble, the complete HGNC data set from a TSV file.

#### Usage

```
import_hgnc_dataset(file = latest_archive_url(), ...)
```

#### Arguments

file	A file or URL of the complete HGNC data set (in TSV format).
	Additional arguments to be passed on to readr::read_tsv().

#### Value

A tibble of the HGNC data set consisting of 55 columns:

- hgnc\_id A unique ID provided by the HGNC for each gene with an approved symbol. IDs are of the format HGNC:n, where n is a unique number. HGNC IDs remain stable even if a name or symbol changes.
- hgnc\_id2 A stripped down version of hgnc\_id where the prefix "HGNC:" has been removed (this column is added by the package {hgnc}).
- symbol The official gene symbol approved by the HGNC, which is typically a short form of the gene name. Symbols are approved in accordance with the Guidelines for Human Gene Nomenclature.
- name The full gene name approved by the HGNC; corresponds to the approved symbol above.
- locus\_group A group name for a set of related locus types as defined by the HGNC. One of: "protein-coding gene", "non-coding RNA", "pseudogene" or "other".

locus\_type Specifies the genetic class of each gene entry:

- "gene with protein product" Protein-coding genes (the protein may be predicted and of unknown function), SO:0001217.
- "RNA, cluster" Region containing a cluster of small non-coding RNA genes.
- "RNA, long non-coding" Non-protein coding genes that encode long non-coding RNAs (lncR-NAs) SO:0001877; these are at least 200 nt in length. Subtypes include intergenic SO:0001463, intronic SO:0001903 and antisense SO:0001904.
- "RNA, micro" Non-protein coding genes that encode microRNAs (miRNAs), SO:0001265.
- "RNA, ribosomal" Non-protein coding genes that encode ribosomal RNAs (rRNAs), SO:0001637.
- "RNA, small nuclear" Non-protein coding genes that encode small nuclear RNAs (snR-NAs), SO:0001268.
- "RNA, small nucleolar" Non-protein coding genes that encode small nucleolar RNAs (snoR-NAs) containing C/D or H/ACA box domains, SO:0001267.
- "RNA, small cytoplasmic" Non-protein coding genes that encode small cytoplasmic RNAs (scRNAs), SO:0001266.
- "RNA, transfer" Non-protein coding genes that encode transfer RNAs (tRNAs), SO:0001272.
- "RNA, small misc" Non-protein coding genes that encode miscellaneous types of small ncR-NAs, such as vault (SO:0000404) and Y (SO:0000405) RNA genes.
- "phenotype only" Mapped phenotypes where the causative gene has not been identified, SO:0001500.
- "pseudogene" Genomic DNA sequences that are similar to protein-coding genes but do not encode a functional protein, SO:0000336.
- "complex locus constituent" Transcriptional unit that is part of a named complex locus.
- "endogenous retrovirus" Integrated retroviral elements that are transmitted through the germline, SO:0000100.

"fragile site" A heritable locus on a chromosome that is prone to DNA breakage.

- "immunoglobulin gene" Gene segments that undergo somatic recombination to form heavy or light chain immunoglobulin genes (SO:0000460). Also includes immunoglobulin gene segments with open reading frames that either cannot undergo somatic recombination, or encode a peptide that is not predicted to fold correctly; these are identified by inclusion of the term "non-functional" in the gene name.
- "immunoglobulin pseudogene" Immunoglobulin gene segments that are inactivated due to frameshift mutations and/or stop codons in the open reading frame.
- "protocadherin" Gene segments that constitute the three clustered protocadherins (alpha, beta and gamma)
- "readthrough" A naturally occurring transcript containing coding sequence from two or more genes that can also be transcribed individually.
- "region" Extents of genomic sequence that contain one or more genes, also applied to nongene areas that do not fall into other types.
- "T cell receptor gene" Gene segments that undergo somatic recombination to form either alpha, beta, gamma or delta chain T cell receptor genes (SO:0000460). Also includes T cell receptor gene segments with open reading frames that either cannot undergo somatic recombination, or encode a peptide that is not predicted to fold correctly; these are identified by inclusion of the term "non-functional" in the gene name.
- "T cell receptor pseudogene" T cell receptor gene segments that are inactivated due to frameshift mutations and/or stop codons in the open reading frame.
- "transposable element" A segment of repetitive DNA that can move, or retrotranspose, to new sites within the genome (SO:0000101).
- "unknown" Entries where the locus type is currently unknown.
- "virus integration site" Target sequence for the integration of viral DNA into the genome.
- status Status of the symbol report, which can be either "Approved" or "Entry Withdrawn".
- location Chromosomal location. Indicates the cytogenetic location of the gene or region on the chromsome, e.g. "19q13.43". In the absence of that information one of the following may be listed:
  - "not on reference assembly" Named gene is not annotated on the current version of the Genome Reference Consortium human reference assembly; may have been annotated on previous assembly versions or on a non-reference human assembly.
  - "unplaced" Named gene is annotated on an unplaced/unlocalized scaffold of the human reference assembly.
  - "reserved" Named gene has never been annotated on any human assembly.
- location\_sortable A sortable version of the location column (see above).
- alias\_symbol Alternative symbols that have been used to refer to the gene. Aliases may be from literature, from other databases or may be added to represent membership of a gene group.
- alias\_name Alternative names for the gene. Aliases may be from literature, from other databases or may be added to represent membership of a gene group.
- prev\_symbol This field displays any symbols that were previously HGNC-approved nomenclature.
- prev\_name This field displays any names that were previously HGNC-approved nomenclature.
- gene\_group A gene group. Each gene has been assigned to one or more groups, according to either sequence similarity or information from publications, specialist advisors for that group or other databases. Groups may be either structural or functional.

- gene\_group\_id Gene group identifier, an integer number. This column contains the gene group identifiers, see gene\_group for the gene group name.
- date\_approved\_reserved The date the entry was first approved.
- date\_symbol\_changed The date the gene symbol was last changed.
- date\_name\_changed The date the gene name was last changed.
- date\_modified Date the entry was last modified.
- entrez\_id Entrez gene identifier.
- ensembl\_gene\_id Ensembl gene identifier.
- vega\_id VEGA gene identifier.
- ucsc\_id UCSC gene identifier.
- ena International Nucleotide Sequence Database Collaboration (GenBank, ENA and DDBJ) accession number(s).
- refseq\_accession The Reference Sequence (RefSeq) identifier for that entry, provided by the NCBI.
- ccds\_id Consensus CDS identifier.
- uniprot\_ids UniProt protein accession.
- pubmed\_id Pubmed and Europe Pubmed Central PMIDs.
- mgd\_id Mouse genome informatics database identifier.
- rgd\_id Rat genome database gene identifier.
- 1sdb The name of the Locus Specific Mutation Database and URL for the gene.
- cosmic Symbol used within the Catalogue of somatic mutations in cancer for the gene.
- omim\_id Online Mendelian Inheritance in Man (OMIM) identifier.
- mirbase miRBase identifier.
- homeodb Homeobox Database identifier.
- snornabase snoRNABase identifier.
- bioparadigms\_slc Symbol used to link to the SLC tables database at bioparadigms.org for the gene.
- orphanet Orphanet identifier.
- pseudogene.org Pseudogene.org identifier.
- horde\_id Symbol used within HORDE for the gene.
- merops Identifier used to link to the MEROPS peptidase database.
- imgt Symbol used within international ImMunoGeneTics information system.
- iuphar The objectId used to link to the IUPHAR/BPS Guide to PHARMACOLOGY database.
- kznf\_gene\_catalog Lawrence Livermore National Laboratory Human KZNF Gene Catalog (LLNL) identifier.
- mamit-trnadb Identifier to link to the Mamit-tRNA database.
- cd Symbol used within the Human Cell Differentiation Molecule database for the gene.
- Incrnadb IncRNA Database identifier.

#### last\_update

enzyme\_id ENZYME EC accession number.

intermediate\_filament\_db Identifier used to link to the Human Intermediate Filament Database.

- rna\_central\_ids Identifier in the RNAcentral, The non-coding RNA sequence database.
- Incipedia The LNCipedia identifier to which the gene belongs. This will only appear if the gene is a long non-coding RNA.
- gtrnadb The GtRNAdb identifier to which the gene belongs. This will only appear if the gene is a tRNA.
- agr The Alliance of Genomic Resources HGNC ID for the Human gene page within the resource.
- mane\_select MANE Select nucleotide accession with version (i.e. NCBI RefSeq or Ensembl transcript ID and version).

gencc Gene Curation Coalition (GenCC) Database identifier.

#### Examples

## Not run: import\_hgnc\_dataset()

last\_update Last update of HGNC data set

#### Description

This function returns the date of the most recent update of the HGNC data set.

#### Usage

last\_update()

#### Value

A POSIXct date-time object.

#### Examples

try(last\_update())

latest\_archive\_url Latest HGNC archive URL

#### Description

Latest HGNC archive URL

#### Usage

```
latest_archive_url(type = c("tsv", "json"))
```

#### Arguments

type The format of the archive: "tsv" or "json".

#### Value

A string with the latest HGNC archive URL.

#### Examples

latest\_archive\_url()

list\_archives List monthly and quarterly archives

## Description

This function lists the monthly and quarterly archives currently available.

#### Usage

list\_archives()

#### Value

A tibble of available archives for download.

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