## seq2pathway.data Vignette

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### November 4, 2025

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### 1 Abstract

Seq2pathway.data is supporting data for the seq2pathway package. The package includes pre-defined gene sets which are constructed from R package org.Hs.eg.db[1] for GeneOntology and the Molecular Signatures Database (MsigDB)[2] for other functional gene sets. The gene locus definitions in the package is built from the GENCODE project[3], including GENCODE 36 (hg38), GENCODE 19 (hg19), GENCODE mmvM15 (mm10) and GENCODE mmvM1 (mm9) currently.

## 2 Data

We grouped all data to nine categories. The data 2.1 to 2.5 is aimed to internal funtion use only. The data 2.6 to 2.9 is demo data for the seq2pathway package.

## 2.1 GO\_BP\_list; GO\_MF\_list; GO\_CC\_list

These data contains all gene symbol lists extracted from an R object org.Hs.egG02EG. Note that org.Hs.egG02EG "provides mappings between entrez gene identifiers and the GO identifiers that they are directly associated with. This mapping and its reverse mapping do NOT associate the child terms from the GO ontology with the gene. Only the directly evidenced terms are represented here."[1]

The list <code>GO\_BP\_list</code> includes 9407 GO biological process terms, and the names of list stand for the names of GO terms. The corresponding element of each list is the gene symbol of each term. Similarly, the list <code>GO\_MF\_list</code> includes 3529 GO molecular function terms, and the list <code>GO\_CC\_list</code> includes 1198 GO cell component terms.

Data in this category will mainly be invoked internally by the functions in the seq2pathway package.

## 2.2 Des BP list; Des MF list; Des CC list

These three lists include the description information of each GO terms. The name of each list stands for the name of GO terms. The corresponding element of each list is the description of each term. The description is extracted from R object GO.db[4]. The length of the list Des\_BP\_list is equal to the length of the list GO\_BP\_list. The similar pattern applies to the other two lists.

Data in this category will mainly be invoked internally by the functions in the seq2pathway package.

# 2.3 GO\_GENCODE\_df\_hg\_v19; GO\_GENCODE\_df\_hg\_v36; GO\_GENCODE\_df\_mm\_vM1; GO\_GENCODE\_df\_mm\_vM25

The gene set varies from different gene databese sources. The gene symbols of GO terms are from org.Hs.egGO2EG[1], the gene set in our annoation package is from the GENCODE[3] data sets, which attribute gene set by differenct organisms and assembly versions. These data frames record the common genes from different databases. For example, the GO\_GENCODE\_df\_hg\_v19 data frame records the common genes from org.Hs.egGO2EG and GENCODE human species 19 version. In summary, there are 17998 genes in GO\_GENCODE\_df\_hg\_v19, 20455 genes in GO\_GENCODE\_df\_hg\_v36, 14328 genes in GO\_GENCODE\_df\_mm\_vM1, and 22972 genes in GO\_GENCODE\_df\_mm\_vM25 respectively.

Data in this category will mainly be invoked internally by the functions in the seq2pathway package.

## 

These four data frames record the common genes from Molecular Signatures Database (MsigDB)[2] and the GENCODE[3] data sets. MsigDB is a collection of annotated gene sets. There are 22751 genes in Msig\_GENCODE\_df\_hg\_v19, 22721 genes in Msig\_GENCODE\_df\_hg\_v36, 15420 genes in Msig\_GENCODE\_df\_mm\_vM1, and 16762 genes in Msig\_GENCODE\_df\_mm\_vM25.

Data in this category will mainly be invoked internally by the functions in the seq2pathway package.

### 2.5 gencode coding

gencode\_coding is an R vector which collects all protein coding gene symbols from GENCODE[3] human version20. There are 19810 unique coding genes symbols in gencode coding object.

Data gencode\_coding will mainly be invoked internally by the functions in the seq2pathway package.

### 2.6 MsigDB C5

MsigDB\_C5 is a gene-sets collection from MsigDB[2] in GSA.genesets format, which could be open with R package GSA[5]. MsigDB\_C5 consists of genes annotated by the GO terms. As an R object, MsigDB\_C5 is a list of 3 elements. The first element includes multiple sub lists. Each sub list is a gene list for one gene set. The second element records the names of genesets, and the last element is the descriptions of genesets. Seq2pathway.data is a supporting data package for the seq2pathway package. MsigDB\_C5 is used as demo data for functions in the seq2pathway package. More details could be found in the vignette of seq2pathway package.

```
> data(MsigDB_C5,package="seq2pathway.data")
```

<sup>&</sup>gt; class(MsigDB\_C5)

## 2.7 gene\_description

gene\_description is a data frame with two columns. The data give the gene descrption based on the biomaRt[6] package. gene\_description is a demo data for the seq2pathway package. More details could be found at the Vignette (5.1.4 Add description for genes) of the seq2pathway package.

```
> data(gene_description,package="seq2pathway.data")
> head(gene_description)
```

| hgnc_symbol | description   |  |
|-------------|---|--|
| ABCD4       | ATP-binding cassette, sub-family D (ALD), member 4 [Source:HGNC Symbol;Acc:68 ] |  |
| ABHD12B     | abhydrolase domain containing 12B [Source:HGNC Symbol;Acc:19837]                |  |
| ABHD4       | abhydrolase domain containing 4 [Source:HGNC Symbol;Acc:20154]                  |  |
| ACIN1       | apoptotic chromatin condensation inducer 1 [Source:HGNC Symbol;Acc:17066]       |  |
| ACOT1       | acyl-CoA thioesterase 1 [Source:HGNC Symbol;Acc:33128]                          |  |
| ACOT2       | acyl-CoA thioesterase 2 [Source:HGNC Symbol;Acc:18431]                          |  |

## 2.8 dat\_gene2path\_chip; dat\_gene2path\_RNA

dat\_gene2path\_chip and dat\_gene2path\_RNA are demo data for functions in the seq2pathway package. Each of them is an R list object with 2 elements. The first element gene2pathway\_result.2 is a list of gene2pathway test result, and the second element gene2pathway\_result.FET is a list of Fisher's exact test result. More details and usage could be found at Examples section in the vignette of seq2pathway package.

Small code for checking the data dat\_gene2path\_chip is provided below:

|            | Des   | peakscore                | peakscore            | Intersect | Intersect   |
|------------|---|--------------------------|----------------------|-----------|---|
|            |   | pathscore<br>_Normalized | pathscore<br>_Pvalue | _Count    | _gene   |
|            |   |                          |                      |           |   |
| GO:0000082 | The mitotic cell cycle transition by which a cell in G1 commits to S phase. The process begins with the build up of G1 cyclin-dependent kinase (G1 CDK), resulting in the activation of transcription of G1 cyclins. The process ends with the positive feedback of the G1 cyclins on the G1 CDK which commits the cell to S phase, in which DNA replication is initiated.  | 0.32017745               | 0.12                 | 11        | CDKN3 GPR132 MNAT1 POLE2 PSMA3 PSMA6 PSME<br>PSMC1 PSMC6 PSME1 PSME2  |
| GO:0000086 | The mitotic cell cycle transition by which a cell in G2 commits to M phase. The process begins when the kinase activity of M cyclin/CDK complex reaches a threshold high enough for the cell cycle to proceed. This is accomplished by activating a positive feedback loop that results in the accumulation of unphosphorylated and active M cyclin/CDK complex.  | -0.33586010              | 0.49                 | 5         | AJUBA DYNC1H1 HSP90AA1 LIN52 MNAT1  |
| GO:0000122 | Any process that stops, prevents, or reduces the frequency, rate or extent of transcription from an RNA polymerase II promoter.   | -0.11535853              | 0.16                 | 20        | AJUBA BMP4 DACT1 DICER1 ESR2 FOXA1 GSC JDP2 NKX<br>1 PPM1A PRMT5 PSEN1 RCOR1 SALL2 SIX1 SNW1 STRN<br>YY1 ZBTB1 ZBTB42 |
| GO:0000209 | Addition of multiple ubiquitin groups to a protein, forming a ubiquitin chain.  | 0.17070465               | 0.11                 | 11        | ASB2 G2E3 PSMA3 PSMA6 PSMB11 PSMB5 PSMC1 PSMC<br>PSME1 PSME2 RNF31  |
| GO:0000278 | Progression through the phases of the mitotic cell cycle, the most common eu-<br>karyotic cell cycle, which canonically comprises four successive phases called<br>G1, S, G2, and M and includes replication of the genome and the subsequent<br>segregation of chromosomes into daughter cells. In some variant cell cycles<br>nuclear replication or nuclear division may not be followed by cell division, or<br>G1 and G2 phases may be absent. | 0.06368249               | 0.04                 | 16        | AJUBA DYNC1H1 HSP90A41 LIN52 MNAT1 NEK9 POLE<br>PSMA3 PSMA6 PSMB11 PSMB5 PSMC1 PSMC6 PSME<br>PSME2 VRK1               |
| GO:0000398 | The joining together of exons from one or more primary transcripts of mes-<br>senger RNA (mRNA) and the excision of intron sequences, via a spliceosomal<br>mechanism, so that mRNA consisting only of the joined exons is produced.  | -0.55767621              | 0.59                 | 8         | CPSF2 HNRNPC NOVA1 PABPN1 PAPOLA PNN SNW1 SRSI  |

## 

dat\_seq2pathway\_GOterms and dat\_seq2pathway\_Msig are demo data for functions in the seq2pathway package. Each of them is an R list object with 3 elements. The first element seq2gene\_result is a list with annotation tables. The seconde element gene2pathway\_result.FAIME is a list of gene2pathway FAIME[7] test result. And the third element gene2pathway\_result.FET is a list of Fisher's exact test results. More details and usage could be found in the Examples section in the vignette of the seq2pathway package.

## References

- [1] Carlson M., org. Hs. eg. db: Genome wide annotation for Human., R package version 3.0.0.
- [2] Subramanian, Tamayo, et al., Gene set enrichment analysis: A knowledge-based approach for interpreting genome-wide expression profiles, PNAS 102 (2005), 1545–1550.
- [3] Harrow J, et al., *GENCODE: The reference human genome annotation for The ENCODE Project*, Genome Res. **9** (2012), 1760–1774.
- [4] Carlson M., GO.db: A set of annotation maps describing the entire Gene Ontology., R package version 3.0.0.
- [5] Efron, B. and Tibshirani, R. On testing the significance of sets of genes., Stanford tech report rep 2006.
- [6] Durinck S, Spellman P, Birney E and Huber W, *Mapping identifiers for the integration of genomic datasets with the R/Bioconductor package biomaRt*, Nature Protocols **4** (2009), 1184–1192.

[7] X. Yang, K. Regan, Y. Huang, Q. Zhang, J. Li, T. Y. Seiwert, et al., Single sample expression-anchored mechanisms predict survival in head and neck cancer, PLoS Comput Biol 8 (2012), e1002350.