# Description of the biomaRt package

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# 1 Introduction

The BioConductor *biomaRt* package enables to directly query databases based on biomaRt such as Ensembl, a software system which produces and maintains automatic annotation on metazoan genomes. This way you can annotate the features on your array with the latest annotations starting from identifiers such as affy id's, locuslink, RefSeq and more. Annotation includes gene names, GO and OMIM annotation (depending on species).

# 2 objects

## 2.1 Mart-class

An object of the Mart class stores connections to BioMart databases and aditional information about the BioMarts. It has the following slots:

- connections: stores the RMySQLConnections
- arrayToSpecies: Stores mapping from affy arrays to species

#### 2.2 martTable-class

An object of the martTable class is the output of most biomaRt funtions and has the following slots:

- id: stores the id used for querying
- table: is a list of vectors storing the retrieved data

# 3 Functions

#### 3.1 martConnect

A first step in using the biomaRt package is to connect to a BioMart database. The function martConnect establishes a connection with one or more of the following BioMart databases: snp, ensembl, uniprot, sequence and vega. Default this function will connect to public BioMart databases. If no biomart is specified, only a connection to ensembl will be established. If you want to use local BioMart install you have to set the local argument to TRUE and specify host, password and user details in the corresponding arguments.

Examples:

#### > library(biomaRt)

```
Loading required package: Biobase Welcome to Bioconductor
```

Vignettes contain introductory material. To view,

simply type: openVignette()

For details on reading vignettes, see

the openVignette help page.

Loading required package: RMySQL Loading required package: DBI

> mart <- martConnect()</pre>

connected to: ensembl\_mart\_31

#### 3.2 martDisconnect

You can only hold a limited number of connections with different BioMarts. The function martDisconnect can be used to close a mart connection.

Examples:

> martDisconnect(mart)

# 3.3 getGene

The function getGene uses a query id to look up identification and chromosomal information of the corresponding gene. Depending on the selected output, this function returns a mart-Table. Currently the getGene function takes identifiers from entrezgene, ensembl, refseq, affy, hugo and embl. Besides the id argument, this function also has a species, array and type argument.

The id argument is either a vector of identifiers or a single identifier to be annotated.

The species argument should have the species from which the identifier comes as value. For the value of species, we use the full name of the species where separate words are separated by an underscore, e.g. 'gallusgallus'. A list of possible species to choose from can be obtained by executing the function getSpecies.

The array argument takes affy array identifiers as values. A list of possible identifiers supported by the package can be obtained by executing the function getAffyArrays.

The mart argument is a mart connection, which was obtained using the method martConnect

The type takes the values of 'entrezgene', 'refseq' and 'embl' to clarify which type of identifier is specified in the id argument.

The output can be changed using the output argument. One can choose between a mart-Table (default) and an output of Gene/Multi-Gene objects. Depending on the identifier, different additional arguments will have to be given, summarized below:

• Affy id's: id, array, mart

- Entrez-Gene: id, type, species, mart
- RefSeq: id, type, species, mart
- embl: id, type, species, mart

Note: make sure you have a Mart object with connections to ensembl and vega (only if you want to retrieve data from VEGA)

Examples:

\$end

\$martID

[1] 7531642

[1] "ENSG00000141510"

```
> mart <- martConnect(biomarts = c("ensembl", "snp", "sequence",
      "uniprot", "vega"))
connected to: ensembl_mart_31
connected to: snp_mart_31
connected to: sequence_mart_31
connected to: uniprot_mart_16
connected to: vega_mart_31
> getGene(id = "1939_at", array = "hg_u95av2", mart = mart)
An object of class "martTable"
Slot "id":
[1] "1939_at"
Slot "table":
$symbol
[1] "TP53"
$description
[1] "Cellular tumor antigen p53 (Tumor suppressor p53) (Phosphoprotein p53) (Antigen NY-
$band
[1] "p13.1"
$chromosome
[1] "17"
$start
[1] 7512464
```

```
> getGene(id = 672, type = "entrezgene", species = "hsapiens",
      mart = mart)
An object of class "martTable"
Slot "id":
[1] "672"
Slot "table":
$symbol
[1] "BRCA1"
$description
[1] "Breast cancer type 1 susceptibility protein (RING finger protein 53). [Source:Unipr
$band
[1] "q21.31"
$chromosome
[1] "17"
$start
[1] 38449844
$end
[1] 38530934
$martID
```

# 3.4 getGO

[1] "ENSG00000012048"

The function getGO uses a query id to look up GO annotation of the corresponding gene. Currently the getGO function takes identifiers from entrezgene, hugo, ensembl, refseq, affy and embl. Besides the id argument, this function also has a species, array and type argument.

The id argument is either a vector of identifiers or a single identifier to be annotated.

The species argument should have the species from which the identifier comes as value. A list of possible species to choose from can be obtained by executing the function getSpecies. The array argument takes affy array identifiers as values. A list of possible identifiers supported by the package can be obtained by executing the function getAffyArrays.

The mart argument is a mart connection, which was obtained using the method martConnect

A last argument of this function is the type argument which, takes the values of 'entrez-gene', 'refseq' and 'embl' to clarify which type of identifier is specified in the id argument.

Depending on the identifier, different additional arguments will have to be given, summarized below:

- Affy id's: id, array, mart
- Entrez-Gene: id, type, species, mart
- RefSeq: id, type, species, mart
- embl: id, type, species, mart

Note: make sure you have a Mart object with a connection to ensembl Examples:

```
> getGO(id = "1939_at", array = "hg_u95av2", mart = mart)
An object of class "martTable"
Slot "id":
 [1] "1939_at" "1939_at" "1939_at" "1939_at" "1939_at" "1939_at" "1939_at"
 [8] "1939_at" "1939_at" "1939_at" "1939_at" "1939_at" "1939_at" "1939_at"
[15] "1939_at" "1939_at" "1939_at" "1939_at" "1939_at" "1939_at" "1939_at"
[22] "1939_at" "1939_at" "1939_at" "1939_at" "1939_at" "1939_at"
Slot "table":
$GOID
 [1] "GO:0005739" "GO:0005730" "GO:0051262" "GO:0051097" "GO:0046902"
 [6] "GD:0045786" "GD:0030308" "GD:0030154" "GD:0008635" "GD:0008630"
[11] "GD:0008628" "GD:0008283" "GD:0007569" "GD:0007050" "GD:0006915"
[16] "GD:0006355" "GD:0006310" "GD:0006289" "GD:0006284" "GD:0000075"
[21] "G0:0008270" "G0:0005524" "G0:0005515" "G0:0005507" "G0:0004518"
[26] "GD:0003700" "GD:0000739"
$description
 [1] "mitochondrion"
 [2] "nucleolus"
 [3] "protein tetramerization"
 [4] "negative regulation of helicase activity"
 [5] "regulation of mitochondrial membrane permeability"
 [6] "negative regulation of cell cycle"
 [7] "negative regulation of cell growth"
 [8] "cell differentiation"
 [9] "caspase activation via cytochrome c"
[10] "DNA damage response, signal transduction resulting in induction of apoptosis"
```

[11] "induction of apoptosis by hormones"

[12] "cell proliferation"

- [13] "cell aging"
- [14] "cell cycle arrest"
- [15] "apoptosis"
- [16] "regulation of transcription, DNA-dependent"
- [17] "DNA recombination"
- [18] "nucleotide-excision repair"
- [19] "base-excision repair"
- [20] "cell cycle checkpoint"
- [21] "zinc ion binding"
- [22] "ATP binding"
- [23] "protein binding"
- [24] "copper ion binding"
- [25] "nuclease activity"
- [26] "transcription factor activity"
- [27] "DNA strand annealing activity"

#### \$evidence

- [1] "IDA" "IDA" "TAS" "TAS" "TAS" "IEA" "IMP" "TAS" "IDA" "TAS" "TAS" "TAS"
- [13] "IMP" "TAS" "IDA" "IDA" "TAS" "IMP" "TAS" "TAS" "TAS" "IDA" "IPI" "IDA"
- [25] "TAS" "IDA" "IDA"

#### \$martID

- [1] "ENSG00000141510" "ENSG00000141510" "ENSG00000141510" "ENSG00000141510"
- [5] "ENSG00000141510" "ENSG00000141510" "ENSG00000141510" "ENSG00000141510"
- [9] "ENSG00000141510" "ENSG00000141510" "ENSG00000141510" "ENSG00000141510"
- [13] "ENSG00000141510" "ENSG00000141510" "ENSG00000141510" "ENSG00000141510"
- [17] "ENSG00000141510" "ENSG00000141510" "ENSG00000141510" "ENSG00000141510"
- [21] "ENSG00000141510" "ENSG00000141510" "ENSG00000141510" "ENSG00000141510"
- [25] "ENSG00000141510" "ENSG00000141510" "ENSG00000141510"
- > getGO(id = 672, type = "entrezgene", species = "hsapiens", mart = mart)

An object of class "martTable"

Slot "id":

- [1] "672" "672" "672" "672" "672" "672" "672" "672" "672" "672" "672" "672" "672"
- [13] "672" "672" "672" "672" "672" "672" "672" "672" "672" "672" "672" "672" "672"
- [25] "672" "672"

#### Slot "table":

#### \$GOID

- [1] "GD:0008372" "GD:0000075" "GD:0005554" "GD:0005622" "GD:0008274"
- [6] "GD:0005667" "GD:0005634" "GD:0005615" "GD:0000151" "GD:0046600"
- [11] "G0:0045786" "G0:0045739" "G0:0042981" "G0:0042127" "G0:0016567"
- [16] "GD:0006978" "GD:0006359" "GD:0006357" "GD:0016563" "GD:0015631"
- [21] "G0:0008270" "G0:0005515" "G0:0004842" "G0:0003713" "G0:0003684"

#### [26] "GD:0008270"

## \$description

- [1] "cellular\_component unknown"
- [2] "cell cycle checkpoint"
- [3] "molecular\_function unknown"
- [4] "intracellular"
- [5] "gamma-tubulin ring complex"
- [6] "transcription factor complex"
- [7] "nucleus"
- [8] "extracellular space"
- [9] "ubiquitin ligase complex"
- [10] "negative regulation of centriole replication"
- [11] "negative regulation of cell cycle"
- [12] "positive regulation of DNA repair"
- [13] "regulation of apoptosis"
- [14] "regulation of cell proliferation"
- [15] "protein ubiquitination"
- [16] "DNA damage response, signal transduction by p53 class mediator resulting in transc
- [17] "regulation of transcription from Pol III promoter"
- [18] "regulation of transcription from Pol II promoter"
- [19] "transcriptional activator activity"
- [20] "tubulin binding"
- [21] "zinc ion binding"
- [22] "protein binding"
- [23] "ubiquitin-protein ligase activity"
- [24] "transcription coactivator activity"
- [25] "damaged DNA binding"
- [26] "zinc ion binding"

#### \$evidence

- [1] "ND" "NAS" "ND" "IEA" "NAS" "TAS" "TAS" "TAS" "IEA" "NAS" "IEA" "NAS"
- [13] "TAS" "TAS" "IEA" "TAS" "TAS" "TAS" "TAS" "NAS" "TAS" "IPI" "IEA" "TAS"
- [25] "NR" "IEA"

#### \$martID

- [1] "ENSG00000012048" "ENSG00000012048" "ENSG00000012048" "ENSG00000012048"
- [5] "ENSG00000012048" "ENSG00000012048" "ENSG00000012048" "ENSG00000012048"
- [9] "ENSG00000012048" "ENSG00000012048" "ENSG00000012048" "ENSG00000012048"
- [13] "ENSG00000012048" "ENSG00000012048" "ENSG00000012048" "ENSG00000012048"
- [17] "ENSG00000012048" "ENSG00000012048" "ENSG00000012048" "ENSG00000012048"
- [21] "ENSG0000012048" "ENSG00000012048" "ENSG00000012048" "ENSG00000012048"
- [25] "ENSG00000012048" "ENSG00000012048"

# 3.5 getOMIM

The function getOMIM uses a query id to look up OMIM annotation of the corresponding gene. Currently the getOMIM function takes identifiers from entrezgene, affy, refseq, ensembl, hugo and embl. Besides the id argument, this function also has an array ,type and mart argument.

The id argument is either a vector of identifiers or a single identifier to be annotated.

The array argument takes affy array identifiers as values. A list of possible identifiers supported by the package can be obtained by executing the function getAffyArrays.

The type argument takes the values of 'entrezgene', 'refseq' and 'embl' to clarify which type of identifier is specified in the id argument. If the argument array is used then biomaRt knows the identifiers given corresponf to affy id's. The mart argument is a mart connection, which was obtained using the method martConnect

Depending on the identifier, different additional arguments will have to be given, summarized below:

• Affy id's: id, array, mart

• Entrez-Gene: id, type, mart

• RefSeq: id, type, mart

• embl: id, type, mart

Note: make sure you have a Mart object with a connection to ensembl Examples:

```
> getOMIM(id = "1939_at", array = "hg_u95av2", mart = mart)
An object of class "martTable"
Slot "id":
[1] "1939_at" "1939_at"

Slot "table":
$OMIMID
[1] 191170 191170

$disease
[1] "Colorectal cancer, 114500 (3)" "Li-Fraumeni syndrome (3)"
$martID
[1] "ENSG00000141510" "ENSG00000141510"

> getOMIM(id = 672, type = "entrezgene", mart = mart)
```

```
An object of class "martTable"
Slot "id":
[1] "672" "672"

Slot "table":
$0MIMID
[1] 113705 113705

$disease
[1] "Breast cancer-1 (3)" "Ovarian cancer (3)"
$martID
[1] "ENSG00000012048" "ENSG00000012048"
```

# 3.6 getINTERPRO

The function getINTERPRO uses a query id to look up INTERPRO protein domains of the corresponding protein. Currently the getINTERPRO function takes identifiers from entrezgene, affy, refseq and embl, hugo, ensembl. Besides the id argument, this function also has a species, array and type argument.

The id argument is either a vector of identifiers or a single identifier to be annotated.

The species argument should have the species from which the identifier comes as value. A list of possible species to choose from can be obtained by executing the function getSpecies. The array argument takes affy array identifiers as values. A list of possible identifiers supported by the package can be obtained by executing the function getAffyArrays.

The mart argument is a mart connection, which was obtained using the method martConnect

A last argument of this function is the type argument which, takes the values of 'entrezgene', 'refseq' and 'embl' to clarify which type of identifier is specified in the id argument. Depending on the identifier, different additional arguments will have to be given, summarized below:

- Affy id's: id, array, mart
- Entrez-Gene: id, type, species, mart
- RefSeq: id, type, species, mart
- embl: id, type, species, mart

Note: make sure you have a Mart object with a connections to ensembl and uniprot Examples:

```
> getINTERPRO(id = "1939_at", array = "hg_u95av2", mart = mart)
```

```
An object of class "martTable"
Slot "id":
[1] "1939_at" "1939_at" "1939_at" "1939_at" "1939_at"
Slot "table":
$interproid
[1] "IPR002117" "IPR008967" "IPR010991" "IPR011615" "IPR012346"
$name
[1] "p53 tumor antigen"
[2] "p53-like transcription factor, DNA-binding"
[3] "p53, tetramerisation"
[4] "p53, DNA-binding"
[5] "p53 and RUNT-type transcription factor, DNA-binding"
$shortname
[1] "P53"
                      "P53_like_DNA_bnd" "p53_tetrameristn" "p53_DNA_bind"
[5] "P53_RUNT_DNA_bnd"
$type
[1] "F" "D" "D" "D" "D"
> getINTERPRO(id = 100, type = "entrezgene", species = "hsapiens",
+ mart = mart)
An object of class "martTable"
Slot "id":
[1] "100" "100" "100"
Slot "table":
$interproid
[1] "IPR001365" "IPR006330" "IPR006650"
$name
[1] "Adenosine/AMP deaminase"
                                        "Adenosine deaminase"
[3] "Adenosine/AMP deaminase active site"
$shortname
[1] "A/AMP_deaminase" "A_deaminase" "A/AMP_deam_AS"
$type
[1] "F" "F" "A"
```

# 3.7 getFeature

The function getFeature looks up affy identifiers on a given affy array which correspond to a given symbol. As output this function returns a martTable. Currently the getFeature function takes identifiers from affy only. Besides the symbol argument, this function also has array and mart argument.

The mart argument is a mart connection, which was obtained using the method martConnect

A last argument of this function is the type argument which, takes the values of 'affy', 'locuslink', 'refseq' and 'embl' to clarify which type of identifier is specified in the id argument.

Note: make sure you have a Mart object with a connection to ensembl Examples:

```
> getFeature(symbol = "P53", array = "hg_u95av2", mart = mart)
An object of class "martTable"
Slot "id":
[1] "36079_at" "1974_s_at" "1939_at" "31618_at" "1711_at" "33749_at"
[7] "34822_at" "1860_at"
Slot "table":
$symbol
[1] "TP53I3" "TP53" "TP53" "TP53" "TP53BP1" "TP53BP1" "TP53BP2"
[8] "TP53BP2"
```

#### \$description

- [1] "tumor protein p53 inducible protein 3 [Source:RefSeq\_peptide;Acc:NP\_671713]"
- [2] "Cellular tumor antigen p53 (Tumor suppressor p53) (Phosphoprotein p53) (Antigen NY-
- [3] "Cellular tumor antigen p53 (Tumor suppressor p53) (Phosphoprotein p53) (Antigen NY-
- [4] "Cellular tumor antigen p53 (Tumor suppressor p53) (Phosphoprotein p53) (Antigen NY-
- [5] "Tumor suppressor p53-binding protein 1 (p53-binding protein 1) (53BP1). [Source:Uni
- [6] "TP53 activated protein 1 [Source:RefSeq\_peptide;Acc:NP\_009164]"
- [7] "Apoptosis stimulating of p53 protein 2 (Tumor suppressor p53-binding protein 2) (p5
- [8] "Apoptosis stimulating of p53 protein 2 (Tumor suppressor p53-binding protein 2) (p5

#### \$martID

- [1] "ENSG00000115129" "ENSG00000141510" "ENSG00000141510" "ENSG00000141510"
- [5] "ENSG00000067369" "ENSG00000182165" "ENSG00000143514" "ENSG00000143514"

## 3.8 getSequence

The function getSequence retrieves the sequence given it's chromosome, start and end position. As output this function returns a martTable. The mart argument is a mart connection,

which was obtained using the method martConnect and should in this case be the sequence mart.

Note: make sure you have a Mart object with connections to ensembl and sequence Examples:

```
> getSequence(species = "ggallus", chromosome = 1, start = 400,
+ end = 500, mart = mart)

An object of class "martTable"
Slot "id":
[1] "1_400_500"

Slot "table":
$chromosome
[1] 1

$start
[1] 400

$end
[1] 500
```

#### \$sequence

[1] "GTGACATTTCCAGCATTCAGTGTGTCAAAGCCTAGCTTCATTTTTGAATGTATTGAGGGGCAGATGTCCATCTCATGAATCAT

## 3.9 getSNP

The function getSNP retrieves all SNP's between a given a start and end position on a gives chromosome. As output this function returns a martTable. The mart argument is a mart connection, which was obtained using the method martConnect and should in this case be the snp mart.

Note: make sure you have a Mart object with connections to ensembl and snp Examples:

```
> getSNP(chromosome = 8, start = 148350, end = 148612, species = "hsapiens",
+ mart = mart)

An object of class "martTable"
Slot "id":
  [1] "TSC1421398" "TSC1421399" "TSC1421400" NA "TSC1421401"
  [6] NA "TSC1421402" "TSC1737607" NA NA
```

Slot "table":

#### \$snpStart

[1] 148394 148411 148462 148471 148499 148525 148533 148535 148539 148601

#### \$allele

[1] "C/A" "A/G" "C/T" "T/G" "G/A" "G/A" "G/A" "C/T" "C/T" "G/A"

#### \$coding

[1] NA NA NA NA NA NA NA NA NA

#### \$intronic

[1] NA NA NA NA NA NA NA NA NA

#### \$syn

[1] NA NA NA NA NA NA NA NA NA

#### \$utr5

[1] NA NA NA NA NA NA NA NA NA

#### \$utr3

[1] 1 1 1 1 1 1 1 1 1 1

# 3.10 getSpecies

The function getSpecies looks up which species are present in the BioMart. This function currently works only for ensembl.

Note: make sure you have a Mart object with a connection to ensembl Examples:

# > getSpecies(mart = mart)

```
[1] "agambiae" "amellifera" "celegans" "cfamiliaris" [5] "cintestinalis" "dmelanogaster" "drerio" "frubripes" [9] "ggallus" "hsapiens" "mmusculus" "ptroglodytes" [13] "rnorvegicus" "scerevisiae" "tnigroviridis" "xtropicalis"
```

# 3.11 getAffyArrays

The function getAffyArrays retrieves the Affymetrix array identifiers which are present in ensembl and which can be queried using the biomaRt package.

Note: make sure you have a Mart object with a connection to ensembl Examples:

## > getAffyArrays(mart = mart)

```
V1
                            V2
           canine cfamiliaris
1
2
        zebrafish
                        drerio
3
         hg_focus
                      hsapiens
4
  hg_u133_plus_2
                      hsapiens
5
       hg_u133a_2
                      hsapiens
6
         hg_u133a
                      hsapiens
7
         hg_u133b
                      hsapiens
8
        hg_u95av2
                      hsapiens
9
          hg_u95b
                      hsapiens
10
          hg_u95c
                      hsapiens
11
          hg_u95d
                      hsapiens
12
          hg_u95e
                     hsapiens
13
         u133_x3p
                      hsapiens
                    mmusculus
14
        mg_u74av2
15
        mg_u74bv2
                    mmusculus
16
        mg_u74cv2
                    mmusculus
17
       mouse430_2
                    mmusculus
18
      mouse430a_2
                    mmusculus
19
        mu11ksuba
                    mmusculus
20
        mu11ksubb
                    mmusculus
21
         rat230_2 rnorvegicus
22
          rg_u34a rnorvegicus
23
          rg_u34b rnorvegicus
24
          rg_u34c rnorvegicus
```

## 3.12 getHomolog

This function retrieves homologs of genes of one species in another species Note: make sure you have a Mart object with a connection to ensembl Example:

```
> getHomolog(id = 1:20, from.species = "hsapiens", to.species = "mmusculus",
+    from.type = "entrezgene", to.type = "refseq", mart = mart)

An object of class "martTable"
Slot "id":
  [1] "1" "2" "2" "2" "2" "3" "4" "5" "6" "7" "8" "9" "9" "9" "10"
[16] "11" "12" "12" "12" "12" "13" "14" "15" "16" "17" "18" "19" "20"
Slot "table":
$MappedID
```

```
[1] NA "NM_175628" "NM_008646" "NM_008645" "NM_007376" NA
[7] NA NA NA NA NA "NM_008673"
[13] "NM_010874" "NM_008674" "NM_010874" NA "NM_173024" "NM_008458"
[19] "NM_011458" "NM_009253" "NM_009252" "NM_023383" "NM_146110" "NM_009591"
[25] "NM_146217" NA "NM_172961" "NM_013454" "NM_007379"
```

# 3.13 getPossibleXrefs

This function retrieves the possible cross-references present in Ensembl. This is a very general function to see what can be extracted from En sembl. The results of this function can be used in the getXref function to extract the data of interest.

Note: make sure you have a Mart object with a connection to ensembl Example:

# 3.14 getXref

This powerful function retrieves any cross reference in Ensembl Note: make sure you have a Mart object with a connection to ensembl Example:

```
> getXref(id = "1939_at", from.species = "hsapiens", to.species = "mmusculus",
+ from.xref = "affy_hg_u95av2", to.xref = "affy_mouse430_2",
+ mart = mart)

An object of class "martTable"
Slot "id":
[1] "1939_at" "1939_at"
```

```
Slot "table":
$from.id
[1] "1939_at" "1939_at"

$to.id
[1] "1427739_a_at" "1426538_a_at"

$martID
[1] 75949 75949
```