

The biomaRt user's guide

Steffen Durinck* Wolfgang Huber†

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1 Introduction

In recent years a wealth of biological data has become available in public data repositories. Easy access to these valuable data resources and firm integration with data analysis is needed for comprehensive bioinformatics data analysis. The *biomaRt* package, provides an interface to a growing collection of databases implementing the BioMart software suite (<http://www.biomart.org>). The package enables retrieval of large amounts of data in a uniform way without the need to know the underlying database schemas or write complex SQL queries. Examples of BioMart databases are Ensembl, Uniprot and HapMap. These major databases give biomaRt users direct access to a diverse set of data and enable a wide range of powerful online queries from R.

2 Selecting a BioMart database and dataset

Every analysis with *biomaRt* starts with selecting a BioMart database to use. A first step is to check which BioMart web services are available. The function `listMarts` will display all available BioMart web services

```
> library("biomaRt")
> listMarts()
```

*durincks@gene.com

†huber@ebi.ac.uk

```

      biomart           version
1 ENSEMBL_MART_ENSEMBL    Ensembl Genes 84
2     ENSEMBL_MART_SNP  Ensembl Variation 84
3 ENSEMBL_MART_FUNCGEN Ensembl Regulation 84
4     ENSEMBL_MART_VEGA          Vega 64

```

Note: if the function `useMart` runs into proxy problems you should set your proxy first before calling any biomaRt functions. You can do this using the `Sys.getenv` command:

```
Sys.getenv("http_proxy" = "http://my.proxy.org:9999")
```

Some users have reported that the workaround above does not work, in this case an alternative proxy solution below can be tried:

```
options(RCurlOptions = list(proxy="uscache.kcc.com:80",proxyuserpwd="-----:-----"))
```

The `useMart` function can now be used to connect to a specified BioMart database, this must be a valid name given by `listMarts`. In the next example we choose to query the Ensembl BioMart database.

```
> ensembl=useMart("ensembl")
```

BioMart databases can contain several datasets, for Ensembl every species is a different dataset. In a next step we look at which datasets are available in the selected BioMart by using the function `listDatasets`.

```
> listDatasets(ensembl)
```

	dataset	description	version
1	oanatinus_gene_ensembl	Ornithorhynchus anatinus genes (OANA5)	OANA5
2	cporcellus_gene_ensembl	Cavia porcellus genes (cavPor3)	cavPor3
3	gaculeatus_gene_ensembl	Gasterosteus aculeatus genes (BROADS1)	BROADS1
4	itridescemlineatus_gene_ensembl	Ictidomys tridecemlineatus genes (spetri2)	spetri2
5	lafricana_gene_ensembl	Loxodonta africana genes (loxAfr3)	loxAfr3
6	choffmanni_gene_ensembl	Choloepus hoffmanni genes (choHof1)	choHof1
7	csavignyi_gene_ensembl	Ciona savignyi genes (CSAV2.0)	CSAV2.0
8	fcatus_gene_ensembl	Felis catus genes (Felis_catus_6.2)	Felis_catus_6.2
9	rnorvegicus_gene_ensembl	Rattus norvegicus genes (Rnor_6.0)	Rnor_6.0
10	psinensis_gene_ensembl	Pelodiscus sinensis genes (PelSin_1.0)	PelSin_1.0
11	cjacchus_gene_ensembl	Callithrix jacchus genes (C_jacchus3.2.1)	C_jacchus3.2.1
12	ttruncatus_gene_ensembl	Tursiops truncatus genes (turTru1)	turTru1
13	scerevisiae_gene_ensembl	Saccharomyces cerevisiae genes (R64-1-1)	R64-1-1
14	celegans_gene_ensembl	Caenorhabditis elegans genes (WBcel235)	WBcel235
15	csabaeus_gene_ensembl	Chlorocebus sabaeus genes (ChlSab1.1)	ChlSab1.1
16	oniloticus_gene_ensembl	Oreochromis niloticus genes (Orenil1.0)	Orenil1.0
17	amexicanus_gene_ensembl	Astyanax mexicanus genes (AstMex102)	AstMex102
18	trubripes_gene_ensembl	Takifugu rubripes genes (FUGU4.0)	FUGU4.0
19	pmarinus_gene_ensembl	Petromyzon marinus genes (Pmarinus_7.0)	Pmarinus_7.0

20	eeuropaeus_gene_ensembl	Erinaceus europaeus genes (eriEur1)	eriEur1
21	falbicoloris_gene_ensembl	Ficedula albicollis genes (FicAlb_1.4)	FicAlb_1.4
22	etelfairi_gene_ensembl	Echinops telfairi genes (TENREC)	TENREC
23	cintestinalis_gene_ensembl	Ciona intestinalis genes (KH)	KH
24	ptroglodytes_gene_ensembl	Pan troglodytes genes (CHIMP2.1.4)	CHIMP2.1.4
25	nleucogenys_gene_ensembl	Nomascus leucogenys genes (Nleu1.0)	Nleu1.0
26	sscrofa_gene_ensembl	Sus scrofa genes (SScrofa10.2)	SScrofa10.2
27	ocuniculus_gene_ensembl	Oryctolagus cuniculus genes (OryCun2.0)	OryCun2.0
28	dnovemcinctus_gene_ensembl	Dasyurus novemcinctus genes (Dasnov3.0)	Dasnov3.0
29	pcapensis_gene_ensembl	Procavia capensis genes (proCap1)	proCap1
30	tguttata_gene_ensembl	Taeniopygia guttata genes (taeGut3.2.4)	taeGut3.2.4
31	mlucifugus_gene_ensembl	Myotis lucifugus genes (myoLuc2)	myoLuc2
32	hsapiens_gene_ensembl	Homo sapiens genes (GRCh38.p5)	GRCh38.p5
33	pformosa_gene_ensembl	Poecilia formosa genes (PoeFor_5.1.2)	PoeFor_5.1.2
34	tbelangeri_gene_ensembl	Tupaia belangeri genes (tupBel1)	tupBel1
35	mfuro_gene_ensembl	Mustela putorius furo genes (MusPutFur1.0)	MusPutFur1.0
36	ggallus_gene_ensembl	Gallus gallus genes (Galgal4)	Galgal4
37	xtropicalis_gene_ensembl	Xenopus tropicalis genes (JGI4.2)	JGI4.2
38	ecaballus_gene_ensembl	Equus caballus genes (EquCab2)	EquCab2
39	pabelii_gene_ensembl	Pongo abelii genes (PPYG2)	PPYG2
40	drerio_gene_ensembl	Danio rerio genes (GRCz10)	GRCz10
41	xmaculatus_gene_ensembl	Xiphophorus maculatus genes (Xipmac4.4.2)	Xipmac4.4.2
42	tnigroviridis_gene_ensembl	Tetraodon nigroviridis genes (TETRAODON8.0)	TETRAODON8.0
43	lchalumnae_gene_ensembl	Latimeria chalumnae genes (LatChal1)	LatChal1
44	amelanoleuca_gene_ensembl	Ailuropoda melanoleuca genes (ailMeli1)	ailMeli1
45	mmulatta_gene_ensembl	Macaca mulatta genes (MMUL_1)	MMUL_1
46	pvampyrus_gene_ensembl	Pteropus vampyrus genes (pteVam1)	pteVam1
47	panubis_gene_ensembl	Papio anubis genes (PapAnu2.0)	PapAnu2.0
48	mdomestica_gene_ensembl	Monodelphis domestica genes (monDom5)	monDom5
49	acarolinensis_gene_ensembl	Anolis carolinensis genes (AnoCar2.0)	AnoCar2.0
50	vpacos_gene_ensembl	Vicugna pacos genes (vicPac1)	vicPac1
51	tsyrichta_gene_ensembl	Tarsius syrichta genes (tarSyr1)	tarSyr1
52	ogarnettii_gene_ensembl	Otolemur garnettii genes (OtoGar3)	OtoGar3
53	dmelanogaster_gene_ensembl	Drosophila melanogaster genes (BDGP6)	BDGP6
54	mmurinus_gene_ensembl	Microcebus murinus genes (micMur1)	micMur1
55	loculatus_gene_ensembl	Lepisosteus oculatus genes (LepOcu1)	LepOcu1
56	olatipes_gene_ensembl	Oryzias latipes genes (HdrR)	HdrR
57	oprinceps_gene_ensembl	Ochotona princeps genes (OchPri2.0)	OchPri2.0
58	ggorilla_gene_ensembl	Gorilla gorilla genes (gorGor3.1)	gorGor3.1
59	dordii_gene_ensembl	Dipodomys ordii genes (dipOrd1)	dipOrd1
60	oaries_gene_ensembl	Ovis aries genes (Oar_v3.1)	Oar_v3.1
61	mmusculus_gene_ensembl	Mus musculus genes (GRCh38.p4)	GRCh38.p4
62	mgallopavo_gene_ensembl	Meleagris gallopavo genes (UMD2)	UMD2
63	gmorhua_gene_ensembl	Gadus morhua genes (gadMor1)	gadMor1
64	saraneus_gene_ensembl	Sorex araneus genes (sorAra1)	sorAra1
65	aplatyrhynchos_gene_ensembl	Anas platyrhynchos genes (BGI_duck_1.0)	BGI_duck_1.0
66	sharrisii_gene_ensembl	Sarcophilus harrisii genes (DEVIL7.0)	DEVIL7.0
67	meugenii_gene_ensembl	Macropus eugenii genes (Meug_1.0)	Meug_1.0
68	btaurus_gene_ensembl	Bos taurus genes (UMD3.1)	UMD3.1
69	cfamiliaris_gene_ensembl	Canis familiaris genes (CanFam3.1)	CanFam3.1

To select a dataset we can update the `Mart` object using the function `useDataset`. In the example below we choose to use the `hsapiens` dataset.

```
ensembl = useDataset("hsapiens_gene_ensembl",mart=ensembl)
```

Or alternatively if the dataset one wants to use is known in advance, we can select a BioMart database and dataset in one step by:

```
> ensembl = useMart("ensembl",dataset="hsapiens_gene_ensembl")
```

3 How to build a biomaRt query

The `getBM` function has three arguments that need to be introduced: filters, attributes and values. *Filters* define a restriction on the query. For example you want to restrict the output to all genes located on the human X chromosome then the filter `chromosome_name` can be used with value 'X'. The `listFilters` function shows you all available filters in the selected dataset.

```
> filters = listFilters(ensembl)
> filters[1:5,]

      name      description
1 chromosome_name Chromosome name
2          start Gene Start (bp)
3          end   Gene End (bp)
4    band_start     Band Start
5    band_end       Band End
```

Attributes define the values we are interested in to retrieve. For example we want to retrieve the gene symbols or chromosomal coordinates. The `listAttributes` function displays all available attributes in the selected dataset.

```
> attributes = listAttributes(ensembl)
> attributes[1:5,]

      name      description      page
1  ensembl_gene_id Ensembl Gene ID feature_page
2 ensembl_transcript_id Ensembl Transcript ID feature_page
3  ensembl_peptide_id Ensembl Protein ID feature_page
4  ensembl_exon_id   Ensembl Exon ID feature_page
5      description           Description feature_page
```

The `getBM` function is the main query function in biomaRt. It has four main arguments:

- attributes: is a vector of attributes that one wants to retrieve (= the output of the query).
- filters: is a vector of filters that one wil use as input to the query.
- values: a vector of values for the filters. In case multple filters are in use, the values argument requires a list of values where each position in the list corresponds to the position of the filters in the filters argument (see examples below).
- mart: is and object of class **Mart**, which is created by the **useMart** function.

Note: for some frequently used queries to Ensembl, wrapper functions are available: **getGene** and **getSequence**. These functions call the **getBM** function with hard coded filter and attribute names.

Now that we selected a BioMart database and dataset, and know about attributes, filters, and the values for filters; we can build a biomaRt query. Let's make an easy query for the following problem: We have a list of Affymetrix identifiers from the u133plus2 platform and we want to retrieve the corresponding EntrezGene identifiers using the Ensembl mappings.

The u133plus2 platform will be the filter for this query and as values for this filter we use our list of Affymetrix identifiers. As output (attributes) for the query we want to retrieve the EntrezGene and u133plus2 identifiers so we get a mapping of these two identifiers as a result. The exact names that we will have to use to specify the attributes and filters can be retrieved with the **listAttributes** and **listFilters** function respectively. Let's now run the query:

```
> affyids=c("202763_at", "209310_s_at", "207500_at")
> getBM(attributes=c('affy_hg_u133_plus_2', 'entrezgene'), filters = 'affy_hg_u133_plus_2', values = affyids, mart =
```

	affy_hg_u133_plus_2	entrezgene
1	209310_s_at	837
2	207500_at	838
3	202763_at	836

4 Examples of biomaRt queries

In the sections below a variety of example queries are described. Every example is written as a task, and we have to come up with a biomaRt solution to the problem.

4.1 Task 1: Annotate a set of Affymetrix identifiers with HUGO symbol and chromosomal locations of corresponding genes

We have a list of Affymetrix hgu133plus2 identifiers and we would like to retrieve the HUGO gene symbols, chromosome names, start and end positions and the bands of the corresponding genes. The `listAttributes` and the `listFilters` functions give us an overview of the available attributes and filters and we look in those lists to find the corresponding attribute and filter names we need. For this query we'll need the following attributes: `hgnc_symbol`, `chromosome_name`, `start_position`, `end_position`, `band` and `affy_hg_u133_plus_2` (as we want these in the output to provide a mapping with our original Affymetrix input identifiers. There is one filter in this query which is the `affy_hg_u133_plus_2` filter as we use a list of Affymetrix identifiers as input. Putting this all together in the `getBM` and performing the query gives:

```
> affyids=c("202763_at", "209310_s_at", "207500_at")
> getBM(attributes=c('affy_hg_u133_plus_2', 'hgnc_symbol', 'chromosome_name', 'start_position', 'end_position', 'band',
+   filters = 'affy_hg_u133_plus_2', values = affyids, mart = ensembl)

affy_hg_u133_plus_2 hgnc_symbol chromosome_name start_position end_position band
1      209310_s_at      CASP4             11     104813593    104840163 q22.3
2      207500_at       CASP5             11     104864962    104893895 q22.3
3      202763_at       CASP3              4     185548850    185570663 q35.1
```

4.2 Task 2: Annotate a set of EntrezGene identifiers with GO annotation

In this task we start out with a list of EntrezGene identifiers and we want to retrieve GO identifiers related to biological processes that are associated with these entrezgene identifiers. Again we look at the output of `listAttributes` and `listFilters` to find the filter and attributes we need. Then we construct the following query:

```
> entrez=c("673", "837")
> goids = getBM(attributes=c('entrezgene', 'go_id'), filters='entrezgene', values=entrez, mart=ensembl)
> head(goids)

entrezgene      go_id
1      673 GO:0000186
2      673 GO:0006468
3      673 GO:0006916
4      673 GO:0007264
5      673 GO:0007268
```

4.3 Task 3: Retrieve all HUGO gene symbols of genes that are located on chromosomes 17,20 or Y , and are associated with one the following GO terms: "GO:0051330","GO:0000080","GO:0000114","GO:0000082" (here we'll use more than one filter)

The `getBM` function enables you to use more than one filter. In this case the filter argument should be a vector with the filter names. The values should be a list, where the first element of the list corresponds to the first filter and the second list element to the second filter and so on. The elements of this list are vectors containing the possible values for the corresponding filters.

```
go=c("GO:0051330","GO:0000080","GO:0000114")
chrom=c(17,20,"Y")
getBM(attributes= "hgnc_symbol",
      filters=c("go_id","chromosome_name"),
      values=list(go,chrom), mart=ensembl)

hgnc_symbol
1           E2F1
```

4.4 Task 4: Annotate set of identifiers with INTERPRO protein domain identifiers

In this example we want to annotate the following two RefSeq identifiers: NM_005359 and NM_000546 with INTERPRO protein domain identifiers and a description of the protein domains.

```
> refseqids = c("NM_005359", "NM_000546")
> ipro = getBM(attributes=c("refseq_mrna","interpro","interpro_description"), filters
ipro
  refseq_mrna    interpro          interpro_description
  1 NM_000546 IPR002117          p53 tumor antigen
  2 NM_000546 IPR010991          p53, tetramerisation
  3 NM_000546 IPR011615          p53, DNA-binding
  4 NM_000546 IPR013872 p53 transactivation domain (TAD)
  5 NM_000546 IPR000694          Proline-rich region
  6 NM_005359 IPR001132          MAD homology 2, Dwarfin-type
  7 NM_005359 IPR003619          MAD homology 1, Dwarfin-type
  8 NM_005359 IPR013019          MAD homology, MH1
```

4.5 Task 5: Select all Affymetrix identifiers on the hgu133plus2 chip and Ensembl gene identifiers for genes located on chromosome 16 between basepair 1100000 and 1250000.

In this example we will again use multiple filters: `chromosome_name`, `start`, and `end` as we filter on these three conditions. Note that when a chromo-

some name, a start position and an end position are jointly used as filters, the BioMart webservice interprets this as return everything from the given chromosome between the given start and end positions.

```
> getBM(c('affy_hg_u133_plus_2', 'ensembl_gene_id'), filters = c('chromosome_name', 'start', 'end'),
+ values=list(16, 1100000, 1250000), mart=ensembl)

  affy_hg_u133_plus_2 ensembl_gene_id
1                 ENSG00000260702
2        215502_at ENSG00000260532
3                 ENSG00000273551
4        205845_at ENSG00000196557
5                 ENSG00000196557
6                 ENSG00000260403
7                 ENSG00000259910
8                 ENSG00000261294
9        220339_s_at ENSG00000116176
10                ENSG00000277010
11       217023_x_at ENSG00000197253
12       210084_x_at ENSG00000197253
13       215382_x_at ENSG00000197253
14       216474_x_at ENSG00000197253
15       207134_x_at ENSG00000197253
16       205683_x_at ENSG00000197253
17       217023_x_at ENSG00000172236
18       210084_x_at ENSG00000172236
19       215382_x_at ENSG00000172236
20       207741_x_at ENSG00000172236
21       216474_x_at ENSG00000172236
22       207134_x_at ENSG00000172236
23       205683_x_at ENSG00000172236
```

4.6 Task 6: Retrieve all entrezgene identifiers and HUGO gene symbols of genes which have a "MAP kinase activity" GO term associated with it.

The GO identifier for MAP kinase activity is GO:0004707. In our query we will use go as filter and entrezgene and hgnc_symbol as attributes. Here's the query:

```
> getBM(c('entrezgene', 'hgnc_symbol'), filters='go', values='GO:0004707', mart=ensembl)

  entrezgene hgnc_symbol
1         5601      MAPK9
2        225689      MAPK15
3         5599      MAPK8
4         5594      MAPK1
5         6300      MAPK12
```

4.7 Task 7: Given a set of EntrezGene identifiers, retrieve 100bp upstream promoter sequences

All sequence related queries to Ensembl are available through the `getSequence` wrapper function. `getBM` can also be used directly to retrieve sequences

but this can get complicated so using `getSequence` is recommended. Sequences can be retrieved using the `getSequence` function either starting from chromosomal coordinates or identifiers. The chromosome name can be specified using the `chromosome` argument. The `start` and `end` arguments are used to specify `start` and `end` positions on the chromosome. The type of sequence returned can be specified by the `seqType` argument which takes the following values: 'cdna';'peptide' for protein sequences; '3utr' for 3' UTR sequences,'5utr' for 5' UTR sequences; 'gene_exon' for exon sequences only; 'transcript_exon' for transcript specific exonic sequences only;'transcript_exon_intron' gives the full unspliced transcript, that is exons + introns;'gene_exon_intron' gives the exons + introns of a gene;'coding' gives the coding sequence only;'coding_transcript_flank' gives the flanking region of the transcript including the UTRs, this must be accompanied with a given value for the upstream or downstream attribute;'coding_gene_flank' gives the flanking region of the gene including the UTRs, this must be accompanied with a given value for the upstream or downstream attribute; 'transcript_flank' gives the flanking region of the transcript excluding the UTRs, this must be accompanied with a given value for the upstream or downstream attribute; 'gene_flank' gives the flanking region of the gene excluding the UTRs, this must be accompanied with a given value for the upstream or downstream attribute.

In MySQL mode the `getSequence` function is more limited and the sequence that is returned is the 5' to 3'+ strand of the genomic sequence, given a chromosome, as start and an end position.

Task 4 requires us to retrieve 100bp upstream promoter sequences from a set of EnsemblGene identifiers. The type argument in `getSequence` can be thought of as the filter in this query and uses the same input names given by `listFilters`. In our query we use `entrezgene` for the type argument. Next we have to specify which type of sequences we want to retrieve, here we are interested in the sequences of the promoter region, starting right next to the coding start of the gene. Setting the `seqType` to `coding_gene_flank` will give us what we need. The `upstream` argument is used to specify how many bp of upstream sequence we want to retrieve, here we'll retrieve a rather short sequence of 100bp. Putting this all together in `getSequence` gives:

```
> entrez=c("673", "7157", "837")
> getSequence(id = entrez, type="entrezgene", seqType="coding_gene_flank", upstream=100, mart=ensembl)
```

4.8 Task 8: Retrieve all 5' UTR sequences of all genes that are located on chromosome 3 between the positions 185514033 and 185535839

As described in the previous task `getSequence` can also use chromosomal coordinates to retrieve sequences of all genes that lie in the given region. We also have to specify which type of identifier we want to retrieve together with the sequences, here we choose for entrezgene identifiers.

```
> utr5 = getSequence(chromosome=3, start=185514033, end=185535839,
+                      type="entrezgene", seqType="5utr", mart=ensembl)
> utr5
```

V1	V2
.....GAAGCGGTGGC	1981

4.9 Task 9: Retrieve protein sequences for a given list of EntrezGene identifiers

In this task the type argument specifies which type of identifiers we are using. To get an overview of other valid identifier types we refer to the `listFilters` function.

```
> protein = getSequence(id=c(100, 5728), type="entrezgene",
+                         seqType="peptide", mart=ensembl)
> protein
```

peptide	entrezgene
MAQTPAFDKPKVEL ...	100
MTAIKEIVSRNKRR ...	5728

4.10 Task 10: Retrieve known SNPs located on the human chromosome 8 between positions 148350 and 148612

For this example we'll first have to connect to a different BioMart database, namely `snp`.

```
> snpmart = useMart("snp", dataset="hsapiens_snp")
```

The `listAttributes` and `listFilters` functions give us an overview of the available attributes and filters. From these we need: `refsnp_id`, `allele`, `chrom_start` and `chrom_end` as attributes; and as filters we'll use: `chrom_start`, `chrom_end` and `chr_name`. Note that when a chromosome name, a start position and an end position are jointly used as filters, the BioMart webservice interprets this as return everything from the given chromosome between the given start and end positions. Putting our selected attributes and filters into `getBM` gives:

```
> getBM(c('refsnp_id', 'allele', 'chrom_start', 'chrom_strand'), filters = c('chr_name', 'chrom_start', 'chrom_end'), val
      refsnp_id allele chrom_start chrom_strand
1   rs1134195   G/T    148394      -1
2   rs4046274   C/A    148394       1
3   rs4046275   A/G    148411       1
4   rs13291    C/T    148462       1
5   rs1134192   G/A    148462      -1
6   rs4046276   C/T    148462       1
7   rs12019378  T/G    148471       1
8   rs1134191   C/T    148499      -1
9   rs4046277   G/A    148499       1
10  rs11136408  G/A    148525       1
11  rs1134190   C/T    148533      -1
12  rs4046278   G/A    148533       1
13  rs1134189   G/A    148535      -1
14  rs3965587   C/T    148535       1
15  rs1134187   G/A    148539      -1
16  rs1134186   T/C    148569       1
17  rs4378731   G/A    148601       1
```

4.11 Task 11: Given the human gene TP53, retrieve the human chromosomal location of this gene and also retrieve the chromosomal location and RefSeq id of it's homolog in mouse.

The `getLDS` (Get Linked Dataset) function provides functionality to link 2 BioMart datasets which each other and construct a query over the two datasets. In Ensembl, linking two datasets translates to retrieving homology data across species. The usage of `getLDS` is very similar to `getBM`. The linked dataset is provided by a separate `Mart` object and one has to specify filters and attributes for the linked dataset. Filters can either be applied to both datasets or to one of the datasets. Use the `listFilters` and `listAttributes` functions on both `Mart` objects to find the filters and attributes for each dataset (species in Ensembl). The attributes and filters of the linked dataset can be specified with the `attributesL` and `filtersL` arguments. Entering all this information into `getLDS` gives:

```
human = useMart("ensembl", dataset = "hsapiens_gene_ensembl")
mouse = useMart("ensembl", dataset = "mmusculus_gene_ensembl")
getLDS(attributes = c("hgnc_symbol", "chromosome_name", "start_position"),
       filters = "hgnc_symbol", values = "TP53", mart = human,
       attributesL = c("refseq_mrna", "chromosome_name", "start_position"), martL = mouse)

V1 V2      V3      V4 V5      V6
1 TP53 17 7512464 NM_011640 11 69396600
```

5 Using archived versions of Ensembl

It is possible to query archived versions of Ensembl through *biomaRt*. There are currently two ways to access archived versions.

5.1 Using the archive=TRUE

First we list the available Ensembl archives by using the `listMarts` function and setting the archive attribute to TRUE. Note that not all archives are available this way and it seems that recently this only gives access to few archives if you don't see the version of the archive you need please look at the 2nd way to access archives.

```
> listMarts(archive=TRUE)

      biomart          version
1       ensembl_mart_51      Ensembl 51
2           snp_mart_51        SNP 51
3           vega_mart_51        Vega 32
4       ensembl_mart_50      Ensembl 50
5           snp_mart_50        SNP 50
6           vega_mart_50        Vega 32
7       ensembl_mart_49  ENSEMBL GENES 49 (SANGER)
8   genomic_features_mart_49  Genomic Features
9           snp_mart_49        SNP
10          vega_mart_49        Vega
11          ensembl_mart_48  ENSEMBL GENES 48 (SANGER)
12  genomic_features_mart_48  Genomic Features
13          snp_mart_48        SNP
14          vega_mart_48        Vega
15          ensembl_mart_47  ENSEMBL GENES 47 (SANGER)
16  genomic_features_mart_47  Genomic Features
17          snp_mart_47        SNP
18          vega_mart_47        Vega
19  compara_mart_homology_47  Compara homology
20 compara_mart_multiple_ga_47  Compara multiple alignments
21 compara_mart_pairwise_ga_47  Compara pairwise alignments
22          ensembl_mart_46  ENSEMBL GENES 46 (SANGER)
23  genomic_features_mart_46  Genomic Features
24          snp_mart_46        SNP
25          vega_mart_46        Vega
26  compara_mart_homology_46  Compara homology
27 compara_mart_multiple_ga_46  Compara multiple alignments
28 compara_mart_pairwise_ga_46  Compara pairwise alignments
29          ensembl_mart_45  ENSEMBL GENES 45 (SANGER)
30          snp_mart_45        SNP
31          vega_mart_45        Vega
32  compara_mart_homology_45  Compara homology
33 compara_mart_multiple_ga_45  Compara multiple alignments
34 compara_mart_pairwise_ga_45  Compara pairwise alignments
35          ensembl_mart_44  ENSEMBL GENES 44 (SANGER)
36          snp_mart_44        SNP
37          vega_mart_44        Vega
38  compara_mart_homology_44  Compara homology
39 compara_mart_pairwise_ga_44  Compara pairwise alignments
40          ensembl_mart_43  ENSEMBL GENES 43 (SANGER)
41          snp_mart_43        SNP
42          vega_mart_43        Vega
43  compara_mart_homology_43  Compara homology
44 compara_mart_pairwise_ga_43  Compara pairwise alignments
```

Next we select the archive we want to use using the `useMart` function, again setting the archive attribute to TRUE and giving the full name of the BioMart e.g. `ensembl_mart_46`.

```
> ensembl = useMart("ensembl_mart_46", dataset="hsapiens_gene_ensembl", archive = TRUE)
```

If you don't know the dataset you want to use could first connect to the BioMart using `useMart` and then use the `listDatasets` function on this object. After you selected the BioMart database and dataset, queries can be performed in the same way as when using the current BioMart versions.

5.2 Accessing archives through specifying the archive host

Use the <http://www.ensembl.org> website and go down the bottom of the page. Click on 'view in Archive' and select the archive you need. Copy the url and use that url as shown below to connect to the specified BioMart database. The example below shows how to query Ensembl 54.

```
> listMarts(host='may2009.archive.ensembl.org')
> ensembl54=useMart(host='may2009.archive.ensembl.org', biomart='ENSEMBL_MART_ENSEMBL')
> ensembl54=useMart(host='may2009.archive.ensembl.org', biomart='ENSEMBL_MART_ENSEMBL', dataset='hsapiens_gene_ensembl')
```

6 Using a BioMart other than Ensembl

To demonstrate the use of the `biomaRt` package with non-Ensembl databases the next query is performed using the Wormbase BioMart (WormMart). We connect to Wormbase, select the gene dataset to use and have a look at the available attributes and filters. Then we use a list of gene names as filter and retrieve associated RNAi identifiers together with a description of the RNAi phenotype.

```
> wormbase=useMart("WS220",dataset="wormbase_gene")
> listFilters(wormbase)
> listAttributes(wormbase)
> getBM(attributes = c("public_name", "rnai", "rnai_phenotype_phenotype_label"),
+       filters="gene_name", values=c("unc-26", "his-33"),
+       mart=wormbase)
>

  public_name      rnai  rnai_phenotype_phenotype_label
1   his-33 WBRNAi00082060          GRO slow growth
2   his-33 WBRNAi00082060 postembryonic development variant
3   his-33 WBRNAi00082060          EMB embryonic lethal
4   his-33 WBRNAi00082060          LVL larval lethal
5   his-33 WBRNAi00082060          LVA larval arrest
6   his-33 WBRNAi00082060 accumulated cell corpses
```

7 biomaRt helper functions

This section describes a set of biomaRt helper functions that can be used to export FASTA format sequences, retrieve values for certain filters and exploring the available filters and attributes in a more systematic manner.

7.1 exportFASTA

The data.frames obtained by the `getSequence` function can be exported to FASTA files using the `exportFASTA` function. One has to specify the data.frame to export and the filename using the `file` argument.

7.2 Finding out more information on filters

7.2.1 filterType

Boolean filters need a value TRUE or FALSE in biomaRt. Setting the value TRUE will include all information that fulfill the filter requirement. Setting FALSE will exclude the information that fulfills the filter requirement and will return all values that don't fulfill the filter. For most of the filters, their name indicates if the type is a boolean or not and they will usually start with "with". However this is not a rule and to make sure you got the type right you can use the function `filterType` to investigate the type of the filter you want to use.

```
> filterType("with_affy_hg_u133_plus_2",ensembl)
```

```
[1] "boolean_list"
```

7.2.2 filterOptions

Some filters have a limited set of values that can be given to them. To know which values these are one can use the `filterOptions` function to retrieve the predetermined values of the respective filter.

```
> filterOptions("biotype",ensembl)
```

```
[1] "[3prime_overlapping_ncrna,antisense,bidirectional_promoter_lncrna,IG_C_gene,IG_C_pseudog...
```

If there are no predetermined values e.g. for the entrezgene filter, then `filterOptions` will return the type of filter it is. And most of the times the filter name or its description will suggest what values one case use for the respective filter (e.g. entrezgene filter will work with enterzgene identifiers as values)

7.3 Attribute Pages

For large BioMart databases such as Ensembl, the number of attributes displayed by the `listAttributes` function can be very large. In BioMart databases, attributes are put together in pages, such as sequences, features, homologs for Ensembl. An overview of the attributes pages present in the respective BioMart dataset can be obtained with the `attributePages` function.

```
> pages = attributePages(ensembl)
> pages
[1] "feature_page" "structure"     "homologs"      "snp"          "snp_somatic"  "sequences"
```

To show us a smaller list of attributes which belong to a specific page, we can now specify this in the `listAttributes` function as follows:

```
> listAttributes(ensembl, page="feature_page")
   name                                     description
1  ensembl_gene_id                         Ensembl Gene ID fe
2  ensembl_transcript_id                   Ensembl Transcript ID fe
3  ensembl_peptide_id                     Ensembl Protein ID fe
4  ensembl_exon_id                        Ensembl Exon ID fe
5  description                            Description fe
6  chromosome_name                        Chromosome Name fe
7  start_position                         Gene Start (bp) fe
8  end_position                           Gene End (bp) fe
9  strand                                 Strand fe
10 band                                   Band fe
11 transcript_start                      Transcript Start (bp) fe
12 transcript_end                         Transcript End (bp) fe
13 transcription_start_site              Transcription Start Site (TSS) fe
14 transcript_length                      Transcript length (including UTRs and CDS) fe
15 transcript tsl                         Transcript Support Level (TSL) fe
16 transcript_gencode_basic              GENCODE basic annotation fe
17 transcript_appris                      APPRIS annotation fe
18 external_gene_name                     Associated Gene Name fe
19 external_gene_source                   Associated Gene Source fe
20 external_transcript_name              Associated Transcript Name fe
21 external_transcript_source_name       Associated Transcript Source fe
22 transcript_count                       Transcript count fe
23 percentage_gc_content                 % GC content fe
24 gene_biotype                          Gene type fe
25 transcript_biotype                    Transcript type fe
26 source                                Source (gene) fe
```

```

27      transcript_source          Source (transcript) fe
28          status                 Status (gene) fe
29          transcript_status      Status (transcript) fe
30          version                Version (gene) fe
31          transcript_version     Version (transcript) fe
32          phenotype_description  Phenotype description fe
33              source_name         Source name fe
34          study_external_id       Study External Reference fe
35              go_id                GO Term Accession fe
36                  name_1006          GO Term Name fe
37          definition_1006         GO Term Definition fe
38          go_linkage_type        GO Term Evidence Code fe
39          namespace_1003          GO domain fe
40          goslim_goa_accession    GOSlim GOA Accession(s) fe
41          goslim_goa_description  GOSlim GOA Description fe
42              arrayexpress         ArrayExpress fe
43              chembl               ChEMBL ID(s) fe
44          clone_based_ensembl_gene_name Clone based Ensembl gene name fe
45          clone_based_ensembl_transcript_name Clone based Ensembl transcript name fe
46          clone_based_vega_gene_name  Clone based VEGA gene name fe
47          clone_based_vega_transcript_name Clone based VEGA transcript name fe
48              ccds                 CCDS ID fe
49          dbass3_id               Database of Aberrant 3' Splice Sites (DBASS3) IDs fe
50          dbass3_name             DBASS3 Gene Name fe
51          dbass5_id               Database of Aberrant 5' Splice Sites (DBASS5) IDs fe
52          dbass5_name             DBASS5 Gene Name fe
53          embl                  EMBL (Genbank) ID fe
54          ens_hs_transcript       Ensembl Human Transcript IDs fe
55          ens_hs_translation      Ensembl Human Translation IDs fe
56          ens_lrg_gene            LRG to Ensembl link gene fe
57          ens_lrg_transcript      LRG to Ensembl link transcript fe
58          entrezgene             EntrezGene ID fe
59          entrezgene_transcript_name EntrezGene transcript name ID fe
60          hpa                   Human Protein Atlas Antibody ID fe
61          ottg                  VEGA gene ID(s) (OTTG) fe
62          ottt                  VEGA transcript ID(s) (OTTT) fe
63          otpp                  VEGA protein ID(s) (OTTP) fe
64          hgnc_id                HGNC ID(s) fe
65          hgnc_symbol            HGNC symbol fe
66          hgnc_transcript_name   HGNC transcript name fe
67          merops                 MEROPS ID fe
68          mim_gene_accession    MIM Gene Accession fe
69          mim_gene_description   MIM Gene Description fe
70          mirbase_accession     miRBase Accession(s) fe
71          mirbase_id             miRBase ID(s) fe

```

```

72     mirbase_transcript_name          miRBase transcript name fe
73             pdb                      PDB ID fe
74             protein_id               Protein (Genbank) ID [e.g. AAA02487] fe
75             reactome                Reactome ID fe
76             reactome_gene            Reactome gene ID fe
77             reactome_transcript      Reactome transcript ID fe
78             refseq_mrna              RefSeq mRNA [e.g. NM_001195597] fe
79             refseq_mrna_predicted     RefSeq mRNA predicted [e.g. XM_001125684] fe
80             refseq_ncrna              RefSeq ncRNA [e.g. NR_002834] fe
81             refseq_ncrna_predicted    RefSeq ncRNA predicted [e.g. XR_108264] fe
82             refseq_peptide            RefSeq Protein ID [e.g. NP_001005353] fe
83             refseq_peptide_predicted  RefSeq Predicted Protein ID [e.g. XP_001720922] fe
84             rfam                     Rfam ID fe
85             rfam_transcript_name      Rfam transcript name fe
86             rnacentral               RNACentral ID fe
87             ucsc                     UCSC ID fe
88             unigene                  Unigene ID fe
89             uniparc                  UniParc fe
90             uniprot_sptrembl         UniProt/TrEMBL Accession fe
91             uniprot_swissprot          UniProt/SwissProt Accession fe
92             uniprot_genename          UniProt Gene Name fe
93             wikigene_name            WikiGene Name fe
94             wikigene_id               WikiGene ID fe
95             wikigene_description       WikiGene Description fe
96     efg_agilent_sureprint_g3_ge_8x60k Agilent SurePrint G3 GE 8x60k probe fe
97     efg_agilent_sureprint_g3_ge_8x60k_v2 Agilent SurePrint G3 GE 8x60k v2 probe fe
98     efg_agilent_wholegenome_4x44k_v1   Agilent WholeGenome 4x44k v1 probe fe
99     efg_agilent_wholegenome_4x44k_v2   Agilent WholeGenome 4x44k v2 probe fe
100            affy_hc_g110           Affy HC G110 probeset fe
101            affy_hg_focus          Affy HG FOCUS probeset fe
102            affy_hg_u133_plus_2    Affy HG U133-PLUS-2 probeset fe
103            affy_hg_u133a_2        Affy HG U133A_2 probeset fe
104            affy_hg_u133a          Affy HG U133A probeset fe
105            affy_hg_u133b          Affy HG U133B probeset fe
106            affy_hg_u95av2         Affy HG U95AV2 probeset fe
107            affy_hg_u95b           Affy HG U95B probeset fe
108            affy_hg_u95c           Affy HG U95C probeset fe
109            affy_hg_u95d           Affy HG U95D probeset fe
110            affy_hg_u95e           Affy HG U95E probeset fe
111            affy_hg_u95a           Affy HG U95A probeset fe
112            affy_hugeneFL          Affy HuGene FL probeset fe
113            affy_huex_1_0_st_v2    Affy HuEx 1_0 st v2 probeset fe
114            affy_hugene_1_0_st_v1  Affy HuGene 1_0 st v1 probeset fe
115            affy_hugene_2_0_st_v1  Affy HuGene 2_0 st v1 probeset fe
116            affy_primeview         Affy primeview fe

```

```

117      affy_u133_x3p
118      agilent_cgh_44b
119          codelink
120      illumina_humanwg_6_v1
121      illumina_humanwg_6_v2
122      illumina_humanwg_6_v3
123      illumina_humanht_12_v3
124      illumina_humanht_12_v4
125      illumina_humanref_8_v3
126          phalanx_onearray
127              family
128          family_description
129              pirsf
130          pirsf_start
131              pirsf_end
132          superfamily
133          superfamily_start
134              superfamily_end
135              smart
136          smart_start
137              smart_end
138          hamap
139          hamap_start
140              hamap_end
141          profile
142          profile_start
143              profile_end
144          prosite
145          prosite_start
146              prosite_end
147          prints
148          prints_start
149              prints_end
150          pfam
151          pfam_start
152              pfam_end
153          tigrfam
154          tigrfam_start
155              tigrfam_end
156          gene3d
157          gene3d_start
158              gene3d_end
159          hmmpanther
160          hmmpanther_start
161          hmmpanther_end

```

Affy U133 X3P probeset feature
Agilent CGH 44b probe feature
Codelink probe feature
Illumina HumanWG 6 v1 probe feature
Illumina HumanWG 6 v2 probe feature
Illumina HumanWG 6 v3 probe feature
Illumina Human HT 12 V3 probe feature
Illumina Human HT 12 V4 probe feature
Illumina Human Ref 8 V3 probe feature
Phalanx OneArray probe feature
Ensembl Protein Family ID(s) feature
Ensembl Family Description feature
PIRSF ID feature
PIRSF start feature
PIRSF end feature
SUPERFAMILY ID feature
SUPERFAMILY start feature
SUPERFAMILY end feature
SMART ID feature
SMART start feature
SMART end feature
HAMAP Accession ID feature
HAMAP start feature
HAMAP end feature
Pfscan ID feature
Pfscan start feature
Pfscan end feature
ScanProsite ID feature
ScanProsite start feature
ScanProsite end feature
PRINTS ID feature
PRINTS start feature
PRINTS end feature
Pfam ID feature
Pfam start feature
Pfam end feature
TIGRFAM ID feature
TIGRFAM start feature
TIGRFAM end feature
Gene3D ID feature
Gene3D start feature
Gene3D end feature
HMMPanther ID feature
HMMPanther start feature
HMMPanther end feature

```

162          interpro
163      interpro_short_description
164          interpro_description
165          interpro_start
166          interpro_end
167          low_complexity
168          low_complexity_start
169          low_complexity_end
170          transmembrane_domain
171          transmembrane_domain_start
172          transmembrane_domain_end
173          signal_domain
174          signal_domain_start
175          signal_domain_end
176          ncoils
177          ncoils_start
178          ncoils_end

```

Interpro ID fe
Interpro Short Description fe
Interpro Description fe
Interpro start fe
Interpro end fe
low complexity (SEG) fe
low complexity (SEG) start fe
low complexity (SEG) end fe
Transmembrane domain (tmhmm) fe
Transmembrane domain (tmhmm) start fe
Transmembrane domain (tmhmm) end fe
signal peptide fe
signal peptide start fe
signal peptide end fe
coiled coil (ncoils) fe
coiled coil (ncoils) start fe
coiled coil (ncoils) end fe

We now get a short list of attributes related to the region where the genes are located.

8 Local BioMart databases

The biomaRt package can be used with a local install of a public BioMart database or a locally developed BioMart database and web service. In order for biomaRt to recognize the database as a BioMart, make sure that the local database you create has a name conform with

`database_mart_version`

where database is the name of the database and version is a version number. No more underscores than the ones showed should be present in this name. A possible name is for example

`ensemblLocal_mart_46`

8.1 Minimum requirements for local database installation

More information on installing a local copy of a BioMart database or develop your own BioMart database and webservice can be found on <http://www.biomart.org>. Once the local database is installed you can use biomaRt on this database by:

```
listMarts(host="www.myLocalHost.org", path="/myPathToWebservice/martservice")
mart=useMart("nameOfMyMart",dataset="nameOfMyDataset",host="www.myLocalHost.org", path="/myPathToWebservice/martser
```

For more information on how to install a public BioMart database see:
<http://www.biomart.org/install.html> and follow link databases.

9 Using select

In order to provide a more consistent interface to all annotations in Bioconductor the `select`, `columns`, `keytypes` and `keys` have been implemented to wrap some of the existing functionality above. These methods can be called in the same manner that they are used in other parts of the project except that instead of taking a `AnnotationDb` derived class they take instead a `Mart` derived class as their 1st argument. Otherwise usage should be essentially the same. You still use `columns` to discover things that can be extracted from a `Mart`, and `keytypes` to discover which things can be used as keys with `select`.

```
> mart<-useMart(dataset="hsapiens_gene_ensembl",biomart='ensembl')
> head(keytypes(mart), n=3)

[1] "affy_hc_g110"          "affy_hg_focus"        "affy_hg_u133_plus_2"

> head(columns(mart), n=3)

[1] "3_utr_end"    "3_utr_end"    "3_utr_start"
```

And you still can use `keys` to extract potential keys, for a particular key type.

```
> k = keys(mart, keytype="chromosome_name")
> head(k, n=3)

[1] "1" "2" "3"
```

When using `keys`, you can even take advantage of the extra arguments that are available for others keys methods.

```
> k = keys(mart, keytype="chromosome_name", pattern="LRG")
> head(k, n=3)

character(0)
```

Unfortunately the `keys` method will not work with all key types because they are not all supported.

But you can still use `select` here to extract columns of data that match a particular set of keys (this is basically a wrapper for `getBM`).

```
> affy=c("202763_at","209310_s_at","207500_at")
> select(mart, keys=affy, columns=c('affy_hg_u133_plus_2','entrezgene'),
+   keytype='affy_hg_u133_plus_2')

affy_hg_u133_plus_2 entrezgene
1      209310_s_at      837
2      202763_at      836
3      207500_at      838
```

So why would we want to do this when we already have functions like `getBM`? For two reasons: 1) for people who are familiar with `select` and its helper methods, they can now proceed to use `biomaRt` making the same kinds of calls that are already familiar to them and 2) because the `select` method is implemented in many places elsewhere, the fact that these methods are shared allows for more convenient programmatic access of all these resources. An example of a package that takes advantage of this is the *OrganismDbi* package. Where several packages can be accessed as if they were one resource.

10 Session Info

```
> sessionInfo()

R version 3.3.0 RC (2016-04-26 r70550)
Platform: x86_64-apple-darwin13.4.0 (64-bit)
Running under: OS X 10.9.5 (Mavericks)

locale:
[1] C/en_US.UTF-8/en_US.UTF-8/C/en_US.UTF-8/en_US.UTF-8

attached base packages:
[1] stats      graphics   grDevices utils      datasets   methods    base

other attached packages:
[1] biomaRt_2.28.0
```

```
loaded via a namespace (and not attached):
[1] IRanges_2.6.0          parallel_3.3.0        DBI_0.4
[6] Biobase_2.32.0         AnnotationDbi_1.34.0 RSQLite_1.0.0
[11] stats4_3.3.0          bitops_1.0-6          XML_3.98-1.4
[14] grid_3.3.0             gridBase_0.4.1       tools_3.3.0
[15] lattice_0.20-30        latticeExtra_0.6-2  S4Vectors_0.10.0
```

> *warnings()*

NULL