

# The biomaRt user's guide

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## Contents

<b>1</b>	<b>Introduction</b>	<b>2</b>
<b>2</b>	<b>Selecting a BioMart database and dataset</b>	<b>3</b>
<b>3</b>	<b>How to build a biomaRt query</b>	<b>5</b>
<b>4</b>	<b>Examples of biomaRt queries</b>	<b>7</b>
4.1	Task 1: Annotate a set of Affymetrix identifiers with HUGO symbol and chromosomal locations of corresponding genes . .	7
4.2	Task 2: Annotate a set of EntrezGene identifiers with GO annotation . . . . .	8
4.3	Task 3: Retrieve all HUGO gene symbols of genes that are located on chromosomes 1,2 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) . . . . .	8
4.4	Task 4: Annotate set of idenfiers with INTERPRO protein domain identifiers . . . . .	9
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. . . . .	9
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. . . . .	10

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4.7	Task 7: Given a set of EntrezGene identifiers, retrieve 100bp upstream promoter sequences . . . . .	11
4.8	Task 8: Retrieve all 5' UTR sequences of all genes that are located on chromosome 3 between the positions 185514033 and 185535839 . . . . .	12
4.9	Task 9: Retrieve protein sequences for a given list of EntrezGene identifiers . . . . .	12
4.10	Task 10: Retrieve known SNPs located on the human chromosome 8 between positions 148350 and 148612 . . . . .	13
4.10.1	getSNP . . . . .	13
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. . . . .	14
4.11.1	getHomolog . . . . .	14
<b>5</b>	<b>Using archived versions of Ensembl</b>	<b>15</b>
<b>6</b>	<b>Using a BioMart other than Ensembl</b>	<b>17</b>
<b>7</b>	<b>biomaRt helper functions</b>	<b>17</b>
7.1	exportFASTA . . . . .	18
7.2	Finding out more information on filters . . . . .	18
7.2.1	filterType . . . . .	19
7.2.2	filterOptions . . . . .	19
7.3	Attribute groups . . . . .	19
<b>8</b>	<b>Local BioMart databases</b>	<b>21</b>
8.1	Minimum requirements for local database installation . . . .	21
<b>9</b>	<b>Session Info</b>	<b>22</b>

# 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()
```

	biomart	version
1	ensembl	ENSEMBL 53 GENES (SANGER UK)
2	snp	ENSEMBL 53 VARIATION (SANGER UK)
3	vega	VEGA 34 (SANGER UK)
4	msd	MSD PROTOTYPE (EBI UK)
5	htgt	HIGH THROUGHPUT GENE TARGETING AND TRAPPING (SANGER UK)
6	QTL_MART	GRAMENE 29 QTL DB (CSHL US)
7	ENSEMBL_MART_ENSEMBL	GRAMENE 29 GENES (CSHL US)
8	ENSEMBL_MART_SNP	GRAMENE 29 SNPs (CSHL US)
9	GRAMENE_MARKER_29	GRAMENE 29 MARKERS (CSHL US)
10	GRAMENE_MAP_29	GRAMENE 29 MAPPINGS (CSHL US)
11	REACTOME	REACTOME (CSHL US)
12	wormbase_current	WORMBASE (CSHL US)
13	dicty	DICTYBASE (NORTHWESTERN US)
14	rgd__mart	RGD GENES (MCW US)
15	ipi_rat__mart	RGD IPI MART (MCW US)
16	SSLP__mart	RGD MICROSATELLITE MARKERS (MCW US)
17	g4public	HGNC (EBI UK)
18	pride	PRIDE (EBI UK)
19	uniprot_mart	UNIPROT (EBI UK)
20	ensembl_expressionmart_48	EURATMART (EBI UK)
21	biomartDB	PARAMECIUM GENOME (CNRS FRANCE)
22	Eurexpress Biomart	EUREXPRESS (MRC EDINBURGH UK)
23	pepseekerGOLD_mart06	PEPSEEKER (UNIVERSITY OF MANCHESTER UK)
24	Pancreatic_Expression	PANCREATIC EXPRESSION DATABASE (INSTITUTE OF CANCER UK)

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.putenv` command:

```
Sys.putenv("http\_proxy" = "http://my.proxy.org:9999")
```

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	tguttata_gene_ensembl	Taeniopygia guttata genes (ZEBRA_FINCH_1)	ZEBRA_FINCH_1
3	cporcellus_gene_ensembl	Cavia porcellus genes (cavPor3)	cavPor3
4	gaculeatus_gene_ensembl	Gasterosteus aculeatus genes (BROADS1)	BROADS1
5	lafricana_gene_ensembl	Loxodonta africana genes (loxAfr2)	loxAfr2
6	agambiae_gene_ensembl	Anopheles gambiae genes (AgamP3)	AgamP3
7	mlucifugus_gene_ensembl	Myotis lucifugus genes (MICROBAT1)	MICROBAT1
8	hsapiens_gene_ensembl	Homo sapiens genes (NCBI36)	NCBI36
9	choffmanni_gene_ensembl	Choloepus hoffmanni genes (SLOTH_1)	SLOTH_1
10	aaegypti_gene_ensembl	Aedes aegypti genes (AaegL1)	AaegL1
11	csavignyi_gene_ensembl	Ciona savignyi genes (CSAV2.0)	CSAV2.0
12	fcatus_gene_ensembl	Felis catus genes (CAT)	CAT
13	rnorvegicus_gene_ensembl	Rattus norvegicus genes (RGSC3.4)	RGSC3.4
14	ggallus_gene_ensembl	Gallus gallus genes (WASHUC2)	WASHUC2
15	tbelangeri_gene_ensembl	Tupaia belangeri genes (TREESHREW)	TREESHREW
16	xtropicalis_gene_ensembl	Xenopus tropicalis genes (JGI4.1)	JGI4.1
17	ecaballus_gene_ensembl	Equus caballus genes (EquCab2)	EquCab2
18	drerio_gene_ensembl	Danio rerio genes (ZFISH7)	ZFISH7
19	stridecemlineatus_gene_ensembl	Spermophilus tridecemlineatus genes (SQUIRREL)	SQUIRREL
20	tnigroviridis_gene_ensembl	Tetraodon nigroviridis genes (TETRAODON7)	TETRAODON7
21	ttruncatus_gene_ensembl	Tursiops truncatus genes (DOLPHIN1)	DOLPHIN1
22	scerevisiae_gene_ensembl	Saccharomyces cerevisiae genes (SGD1.01)	SGD1.01
23	celegans_gene_ensembl	Caenorhabditis elegans genes (WS190)	WS190
24	mmulatta_gene_ensembl	Macaca mulatta genes (MMUL_1)	MMUL_1
25	pvampyrus_gene_ensembl	Pteropus vampyrus genes (MEGABAT1)	MEGABAT1
26	mdomestica_gene_ensembl	Monodelphis domestica genes (BROAD03)	BROAD03
27	vpacos_gene_ensembl	Vicugna pacos genes (ALPACA1)	ALPACA1
28	acarolinensis_gene_ensembl	Anolis carolinensis genes (ANOLE_LIZARD_1)	ANOLE_LIZARD_1
29	tsyrichtha_gene_ensembl	Tarsius syrichta genes (TARSIER1)	TARSIER1
30	ogarnettii_gene_ensembl	Otolemur garnettii genes (BUSHBABY1)	BUSHBABY1
31	trubripes_gene_ensembl	Takifugu rubripes genes (FUGU4)	FUGU4
32	dmelanogaster_gene_ensembl	Drosophila melanogaster genes (BDGP5.4)	BDGP5.4
33	eeuropaeus_gene_ensembl	Erinaceus europaeus genes (HEDGEHOG)	HEDGEHOG
34	mmurinus_gene_ensembl	Microcebus murinus genes (micMur1)	micMur1
35	olatipes_gene_ensembl	Oryzias latipes genes (MEDAKA1)	MEDAKA1
36	etelfairi_gene_ensembl	Echinops telfairi genes (TENREC)	TENREC
37	cintestinalis_gene_ensembl	Ciona intestinalis genes (JGI2)	JGI2
38	ptroglodytes_gene_ensembl	Pan troglodytes genes (CHIMP2.1)	CHIMP2.1
39	oprinceps_gene_ensembl	Ochotona princeps genes (PIKA2)	PIKA2
40	ggorilla_gene_ensembl	Gorilla gorilla genes (GORILLA_1)	GORILLA_1
41	dordii_gene_ensembl	Dipodomys ordii genes (KANGAROO_RAT_1)	KANGAROO_RAT_1
42	ppygmaeus_gene_ensembl	Pongo pygmaeus abelii genes (PPYG2)	PPYG2
43	mmusculus_gene_ensembl	Mus musculus genes (NCBI37)	NCBI37
44	ocuniculus_gene_ensembl	Oryctolagus cuniculus genes (RABBIT)	RABBIT
45	saraneus_gene_ensembl	Sorex araneus genes (COMMON_SHREW1)	COMMON_SHREW1
46	dnovemcinctus_gene_ensembl	Dasypus novemcinctus genes (dasNov2)	dasNov2

47	pcapensis_gene_ensembl	Procavia capensis genes (ROCK_HYRAX_1)	ROCK_HYRAX_1
48	btaurus_gene_ensembl	Bos taurus genes (Btau_4)	Btau_4
49	cfamiliaris_gene_ensembl	Canis familiaris genes (BROADD2)	BROADD2

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	affy_hc_g110	Affy hc g110 ID(s)
2	affy_hg_focus	Affy hg focus ID(s)
3	affy_hg_u133_plus_2	Affy hg u133 plus 2 ID(s)
4	affy_hg_u133a	Affy hg u133a ID(s)
5	affy_hg_u133a_2	Affy hg u133a 2 ID(s)

*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
1	affy_hc_g110	Affy HC G110
2	affy_hg_focus	Affy HG FOCUS
3	affy_hg_u133_plus_2	Affy HG U133-PLUS-2
4	affy_hg_u133a	Affy HG U133A
5	affy_hg_u133a_2	Affy HG U133A_2

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 will use as input to the query.
- `values`: a vector of values for the filters. In case multiple 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 an object of class `Mart`, which is created by the `useMart` function.

Note: for some frequently used queries to Ensembl a set of wrapper are functions available as will be described in the sections below. These wrapper functions are: `getGene`, `getSequence`, `getGO`, `getHomolog`, `getSNP`. All 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 = ensembl)
```

	affy_hg_u133_plus_2	entrezgene
1	202763_at	836
2	202763_at	NA
3	207500_at	838
4	209310_s_at	837

## 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 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	202763_at	CASP3	4	185785844	185807623	q35.1
2	207500_at	CASP5	11	104370180	104384957	q22.3
3	209310_s_at	CASP4	11	104318804	104344535	q22.3

As this is a frequently used query to Ensembl, a wrapper function `getGene` is provided that retrieves a standard set of information based for a given list of identifiers:

```
> getGene(id = affyids, type = "affy_hg_u133_plus_2", mart = ensembl)
```

```

      affy_hg_u133_plus_2 hgnc_symbol
1      202763_at      CASP3
2      207500_at      CASP5
3      209310_s_at      CASP4

1 Caspase-3 Precursor (CASP-3)(EC 3.4.22.56)(Apopain)(Cysteine protease CPP32)(CPP-32)(Yama protein)(SREBP cleavage
2      Caspase-5 Precursor (CASP-5)(EC 3.4.22.58)(ICH-3 protease)(TY prot
3      Caspase-4 Precursor (CASP-4)(EC 3.4.22.57)(ICH-2 protease)(TX
      chromosome_name  band strand start_position end_position ensembl_gene_id
1      4 q35.1      -1      185785844      185807623 ENSG00000164305
2      11 q22.3      -1      104370180      104384957 ENSG00000137757
3      11 q22.3      -1      104318804      104344535 ENSG00000196954

```

## 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")
> getBM(attributes = c("entrezgene", "go_biological_process_id"), filters = "entrezgene", values = entrez,
+      mart = ensembl)

```

```

      entrezgene go_biological_process_id
1      673      GO:0006468
2      673      GO:0006916
3      673      GO:0007264
4      673      GO:0009887
5      673      GO:0007242
6      673      GO:0007165
7      837      GO:0006508
8      837      GO:0006915
9      837      GO:0006917
10     837      GO:0042981

```

## 4.3 Task 3: Retrieve all HUGO gene symbols of genes that are located on chromosomes 1,2 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("G0:0051330","G0:0000080","G0:0000114"chrom=c(1,2,"Y")
getBM(attributes= "hgnc_symbol",
      filters=c("go","chromosome_name"),
      values=list(go,chrom), mart=ensembl)
```

```
hgnc_symbol
1      PPP1CB
2      SPDYA
3      ACVR1
4      CUL3
5      RCC1
6      CDC7
7      RHOU
```

#### 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_dna", "interpro", "interpro_description"), filters = c("refseqids"),
+ values = refseqids, mart = ensembl)
```

```
ipro
  refseq_dna  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, Dwarfing-type
7 NM_005359 IPR003619      MAD homology 1, Dwarfing-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 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.

```
> 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		ENSG00000196364
2	214568_at	ENSG00000095917
3	207741_x_at	ENSG00000172236
4	210084_x_at	ENSG00000172236
5	216474_x_at	ENSG00000172236
6	207134_x_at	ENSG00000172236
7	205683_x_at	ENSG00000172236
8	215382_x_at	ENSG00000172236
9	217023_x_at	ENSG00000172236
10	217023_x_at	ENSG00000197253
11	215382_x_at	ENSG00000197253
12	207134_x_at	ENSG00000197253
13	207741_x_at	ENSG00000197253
14	205683_x_at	ENSG00000197253
15	210084_x_at	ENSG00000197253
16	216474_x_at	ENSG00000197253
17	220339_s_at	ENSG00000116176
18	205845_at	ENSG00000196557

#### 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	984	
2	NA	
3	100133692	CDC2L2
4	100133692	CDC2L1
5	728642	CDC2L2
6	728642	CDC2L1
7	984	CDC2L2
8	984	CDC2L1
9	5594	MAPK1
10	5596	MAPK4
11	5597	MAPK6
12	8621	CDC2L5
13	NA	CDC2L5
14	5598	MAPK7
15	NA	MAPK8
16	5599	MAPK8
17	51701	NLK
18	NA	NLK
19	5602	MAPK10
20	NA	MAPK10
21	5595	MAPK3
22	NA	MAPK3
23	6300	MAPK12

24	NA	MAPK12
25	5600	MAPK11
26	1017	CDK2
27	51755	CRKRS
28	NA	MAPK15
29	225689	MAPK15
30	1432	MAPK14
31	NA	MAPK13
32	5603	MAPK13
33	5601	MAPK9

#### 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 EntrezGene identifiers. The type argument in `getSequence` can be

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

	V1	V2
1 CCTCCGCTCCGCTCCGCTCCGCTCCCCAGCTCTCGCCTCCCTTCCCCTCCCGCCGCACAGCGGCCGTGGGCCCCGGCTCTCGGTTATAAG	673	
2 TCCTTCTCTGCAGGCCAGGTGACCCAGGGTTGGAAGTGTCATGCTGGATCCCCACTTTTCTCTTGAGCAGCCAGACTGCCTCCGGGTCACTGCC	7157	
3 CACGTTTCGGCCCTTTGCAATAAGGAAATACATAGTTTACTTTTATTTTTGACTCTGAGGCTCTTTCCAACGCTGTAAAAAAGGACAGAGGCTGTTCCCT	837	

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

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_strand` 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"), values = list(8, 148350, 148612), mart = snpmart)
```

	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

##### 4.10.1 getSNP

`getSNP` is a wrapper function for retrieving SNP data given a region on the genome.

```
> snp = getSNP(chromosome = 8, start = 148350, end = 148612, mart = snpmart)  
> snp
```

	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_dna", "chromosome_name", "start_position"), martL = mouse)
```

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

##### 4.11.1 getHomolog

The `getHomolog` is a wrapper function for mapping identifiers from one species to another. As described above this can also be done with the more general `getLDS` function. Similar as the `getGene` function, we have to specify the identifier we start from using either the `from.array` argument if the identifier comes from an affy array or else the `from.type` argument if we use an other identifier. The identifier we want to retrieve has to be specified by

using the *to.array* or *to.type* arguments.

A generalized version of the `getHomolog` function is the `getLDS` function (see Advanced Queries section). `getLDS` enables one to combine two datasets (=species in Ensembl) and query any field from one dataset based on the other.

In a first example we start from a affy identifier of a human chip and we want to retrieve the identifiers of the corresponding homolog on a mouse chip.

```
> human = useMart("ensembl", "hsapiens_gene_ensembl")
> mouse = useMart("ensembl", "mmusculus_gene_ensembl")
> homolog = getHomolog( id = "1939_at", to.type = "affy_mouse430_2", from.type =
                        "affy_hg_u95av2", from.mart = human, to.mart = mouse )

> homolog
      V1      V2
1 1939_at 1427739_a_at
2 1939_at 1426538_a_at
```

An other example starts from a human RefSeq id and we want to retrieve the corresponding affy ids on the affy mouse430\_2 chip.

```
> homolog = getHomolog( id = "NM_007294", to.type = "affy_mouse430_2",
                        from.type = "refseq_dna", from.mart = human,
                        to.mart = mouse )

> homolog
      V1      V2
1 NM_007294 1424629_at
2 NM_007294 1451417_at
3 NM_007294 1424630_a_at
```

## 5 Using archived versions of Ensembl

It is possible to query archived versions of Ensembl through *biomaRt*. The steps below show how to do this. First we list the available Ensembl archives by using the `listMarts` function and setting the archive attribute to `TRUE`.

```
> 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 = T
```

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.



## 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("wormbase_current", dataset = "wormbase_gene")
> listFilters(wormbase)
> listAttributes(wormbase)
> getBM(attributes = c("name", "rna_i", "rna_i_phenotype", "phenotype_desc"), filters = "gene_name",
+       values = c("unc-26", "his-33"), mart = wormbase)
```

	name	rna_i	rna_i_phenotype	phenotype_desc
1	his-33	WBRNAi00000104	Emb   Nmo	embryonic lethal   Nuclear morphology alteration in early embryo
2	his-33	WBRNAi00012233	WT	wild type morphology
3	his-33	WBRNAi00024356	Ste	sterile
4	his-33	WBRNAi00025036	Emb	embryonic lethal
5	his-33	WBRNAi00025128	Emb	embryonic lethal
6	his-33	WBRNAi00025393	Emb	embryonic lethal
7	his-33	WBRNAi00025515	Emb   Lva   Unc	embryonic lethal   larval arrest   uncoordinated
8	his-33	WBRNAi00025632	Gro   Ste	slow growth   sterile
9	his-33	WBRNAi00025686	Gro   Ste	slow growth   sterile
10	his-33	WBRNAi00025785	Gro   Ste	slow growth   sterile
11	his-33	WBRNAi00026259	Emb   Gro   Unc	embryonic lethal   slow growth   uncoordinated
12	his-33	WBRNAi00026375	Emb	embryonic lethal
13	his-33	WBRNAi00026376	Emb	embryonic lethal
14	his-33	WBRNAi00027053	Emb   Unc	embryonic lethal   uncoordinated
15	his-33	WBRNAi00030041	WT	wild type morphology
16	his-33	WBRNAi00031078	Emb	embryonic lethal
17	his-33	WBRNAi00032317	Emb	embryonic lethal
18	his-33	WBRNAi00032894	Emb	embryonic lethal
19	his-33	WBRNAi00033648	Emb	embryonic lethal
20	his-33	WBRNAi00035430	Emb	embryonic lethal
21	his-33	WBRNAi00035860	Egl   Emb	egg laying defect   embryonic lethal
22	his-33	WBRNAi00048335	Emb   Sister Chromatid Separation abnormal (Cross-eyed)	embryonic lethal
23	his-33	WBRNAi00049266	Emb   Sister Chromatid Separation abnormal (Cross-eyed)	embryonic lethal
24	his-33	WBRNAi00053026	Emb   Sister Chromatid Separation abnormal (Cross-eyed)	embryonic lethal
25	unc-26	WBRNAi00021278	WT	wild type morphology
26	unc-26	WBRNAi00026915	WT	wild type morphology
27	unc-26	WBRNAi00026916	WT	wild type morphology
28	unc-26	WBRNAi00027544	Unc	uncoordinated
29	unc-26	WBRNAi00049565	WT	wild type morphology
30	unc-26	WBRNAi00049566	WT	wild type morphology

## 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

In BioMart databases, filters can be grouped. Ensembl for example contains the filter groups GENE:, REGION:, ... An overview of the categories and groups for attributes present in the respective BioMart dataset can be obtained with the `filterSummary` function.

```
> summaryF = filterSummary(ensembl)
> summaryF[1:5, ]
```

	category	group
1	FILTERS	GENE:
2	FILTERS	REGION:
3	FILTERS	EXPRESSION:
4	FILTERS	PROTEIN DOMAINS:
5	FILTERS	MULTI SPECIES COMPARISONS:

To show us a smaller list of filters which belong to a specified group or category we can now specify this in the `listFilters` function as follows:

```
> listFilters(ensembl, group = "REGION:")
```

	name	description
1	band_end	<NA>
2	band_start	<NA>
3	chromosomal_region	Chromosome Regions
4	chromosome_name	Chromosome name
5	end	Gene End (bp)
6	hsapiens_encode.encode_region	<NA>
7	hsapiens_encode.type	<NA>
8	marker_end	<NA>
9	marker_start	<NA>
10	start	Gene Start (bp)
11	strand	Strand

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

### 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"
```

### 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] "IG_C_gene"	"IG_D_gene"	"IG_J_gene"	"IG_V_gene"
[6] "miRNA"	"miRNA_pseudogene"	"misc_RNA"	"misc_RNA_pseudogene"
[11] "Mt_tRNA"	"Mt_tRNA_pseudogene"	"protein_coding"	"pseudogene"
[16] "rRNA"	"rRNA_pseudogene"	"scRNA"	"scRNA_pseudogene"
[21] "snoRNA_pseudogene"	"snRNA"	"snRNA_pseudogene"	"tRNA_pseudogene"

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 can use for the respective filter (e.g. `entrezgene` filter will work with `entrezgene` identifiers as values)

## 7.3 Attribute groups

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 categories, such as Sequences, Features, Homologs for Ensembl, and within these categories, attributes can be grouped. The Features category of Ensembl for example contains the attribute groups GENE:, PROTEIN:, ... An overview of the categories and

groups for attributes present in the respective BioMart dataset can be obtained with the `attributeSummary` function.

```
> summaryA = attributeSummary(ensembl)
> summaryA[1:10, ]
```

	category	group
1	Features	EXTERNAL:
2	Features	EXPRESSION:
3	Features	GENE:
4	Features	PROTEIN DOMAINS:
5	Homologs	AEDES ORTHOLOGS:
6	Homologs	ALPACA ORTHOLOGS:
7	Homologs	ANOLE LIZARD ORTHOLOGS:
8	Homologs	ANOPHELES ORTHOLOGS:
9	Homologs	ARMADILLO ORTHOLOGS:
10	Homologs	BUSHBABY ORTHOLOGS:

To show us a smaller list of attributes which belong to a specified group or category we can now specify this in the `listAttributes` function as follows:

```
> listAttributes(ensembl, category = "Features", group = "GENE:")
```

	name	description
1	band	Band
2	canonical_transcript_stable_id	Canonical transcript stable ID(s)
3	chromosome_name	Chromosome Name
4	description	Description
5	end_position	Gene End (bp)
6	ensembl_gene_id	Ensembl Gene ID
7	ensembl_peptide_id	Ensembl Protein ID
8	ensembl_transcript_id	Ensembl Transcript ID
9	external_gene_db	Associated Gene DB
10	external_gene_id	Associated Gene Name
11	external_transcript_id	Associated Transcript Name
12	gene_biotype	Gene Biotype
13	percentage_gc_content	% GC content
14	source	Source
15	start_position	Gene Start (bp)
16	status	Status (gene)
17	strand	Strand
18	transcript_biotype	Transcript Biotype
19	transcript_count	Transcript count
20	transcript_db_name	Associated Transcript DB
21	transcript_end	Transcript End (bp)
22	transcript_start	Transcript Start (bp)
23	transcript_status	Status (transcript)

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. 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

One needs to first download the SQL code to generate the database. For `ensembl_mart_42` this was in the file `ensembl_mart_42.sql.gz`. Then run this SQL code to generate the tables of your local database:

```
mysql -D ensembl_mart_42 -u username -p < ensembl_mart_42.sql
```

Once the tables are created you need to fill the following tables with the downloaded data:

Essential tables:

```
meta_conf__dataset__main.txt.table  
meta_conf__xml__dm.txt.table
```

You can install them from your MySQL command line with:

```
LOAD DATA INFILE 'meta_conf__dataset__main.txt.table' INTO TABLE meta_conf__dataset__main;  
LOAD DATA INFILE 'meta_conf__xml__dm.txt.table' INTO TABLE meta_conf__xml__dm;
```

Next you load all the tables that have the name of your species of interest with with the corresponding table data. Once the local database is installed you can use biomaRt on this database by:

```
mart=useMart("ensembl_mart_42", mysql=TRUE, host="localhost", user="****", password="****",  
            local=TRUE, dataset="hsapiens_gene_ensembl")
```

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

## 9 Session Info

```
> sessionInfo()
```

```
R version 2.8.1 Patched (2009-03-18 r48200)  
i386-apple-darwin9.6.0
```

```
locale:  
C
```

```
attached base packages:
```

```
[1] tools      stats      graphics  grDevices  utils      datasets  methods    base
```

```
other attached packages:
```

```
[1] biomaRt_1.16.0      annotate_1.20.1      xtable_1.5-5        AnnotationDbi_1.4.3 B
```

```
loaded via a namespace (and not attached):
```

```
[1] DBI_0.2-4      Rcurl_0.94-1  RSQLite_0.7-1 XML_2.3-0
```

```
> warnings()
```

```
NULL
```