

The biomaRt user's guide

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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()
```

	biomart	version
1	ensembl	ENSEMBL GENES 64 (SANGER UK)
2	snp	ENSEMBL VARIATION 64 (SANGER UK)
3	functional_genomics	ENSEMBL REGULATION 64 (SANGER UK)
4	vega	VEGA 44 (SANGER UK)
5	bacteria_mart_10	ENSEMBL BACTERIA 10 (EBI UK)
6	fungi_mart_10	ENSEMBL FUNGI 10 (EBI UK)
7	fungi_variations_10	ENSEMBL FUNGI VARIATION 10 (EBI UK)
8	metazoa_mart_10	ENSEMBL METAZOA 10 (EBI UK)
9	metazoa_variations_10	ENSEMBL METAZOA VARIATION 10 (EBI UK)
10	plants_mart_10	ENSEMBL PLANTS 10 (EBI UK)
11	plants_variations_10	ENSEMBL PLANTS VARIATION 10 (EBI UK)
12	protists_mart_10	ENSEMBL PROTISTS 10 (EBI UK)
13	protists_variations_10	ENSEMBL PROTISTS VARIATION 10 (EBI UK)
14	msd	MSD (EBI UK)
15	htgt	WTSI MOUSE GENETICS PROJECT (SANGER UK)
16	REACTOME	REACTOME (CSHL US)
17	WS220	WORMBASE 220 (CSHL US)
18	dicty	DICTYBASE (NORTHWESTERN US)
19	biomart	MGI (JACKSON LABORATORY US)
20	g4public	HGNC (EBI UK)
21	pride	PRIDE (EBI UK)
22	prod-intermart_1	INTERPRO (EBI UK)
23	unimart	UNIPROT (EBI UK)
24	biomartDB	PARAMECIUM GENOME (CNRS FRANCE)
25	Eurexpress Biomart	EUREXPRESS (MRC EDINBURGH UK)
26	pepseekerGOLD_mart06	PEPSEEKER GOLD (UNIVERSITY OF MANCHESTER UK)
27	pepseeker2_mart	PEPSEEKER 2 (UNIVERSITY OF MANCHESTER UK)
28	Potato_01	DB POTATO (INTERNATIONAL POTATO CENTER-CIP)
29	Sweetpotato_01	DB SWEETPOTATO (INTERNATIONAL POTATO CENTER-CIP)
30	phytozome_mart	PHYTOZOME (JGI/CIG US)
31	cyanobase_1	CYANOBASE 1 (KAZUSA JAPAN)
32	HapMap_rel127	HAPMAP 27 (NCBI US)
33	CosmicMart	COSMIC (SANGER UK)
34	cildb_all_v2	CILDB INPARANOID AND FILTERED BEST HIT (CNRS FRANCE)
35	cildb_inp_v2	CILDB INPARANOID (CNRS FRANCE)

36	experiments	INTOGEN EXPERIMENTS
37	combinations	INTOGEN COMBINATIONS
38	oncomodules	INTOGEN ONCOMODULES
39	gmap_japonica	RICE-MAP JAPONICA (PEKING UNIVESITY CHINA)
40	europheomeannotations	EUROPHEOME
41	emma_biomart	THE EUROPEAN MOUSE MUTANT ARCHIVE (EMMA)
42	ikmc	IKMC GENES AND PRODUCTS (IKMC)
43	EMAGE gene expression	EMAGE GENE EXPRESSION
44	EMAP anatomy ontology	EMAP ANATOMY ONTOLOGY
45	EMAGE browse repository	EMAGE BROWSE REPOSITORY
46	GermOnline	GERMONLINE
47	Sigenae_Oligo_Annotation_Ensembl_61	SIGENAE OLIGO ANNOTATION (ENSEMBL 61)
48	Sigenae Oligo Annotation (Ensembl 59)	SIGENAE OLIGO ANNOTATION (ENSEMBL 59)
49	Sigenae Oligo Annotation (Ensembl 56)	SIGENAE OLIGO ANNOTATION (ENSEMBL 56)
50	vectorbase_mart_11	VECTORBASE GENES
51	vectorbase_snp_mart_11	VECTORBASE VARIATION
52	expression	VECTORBASE EXPRESSION MART
53	UTRMart	AURA
54	gmap_indica	RICE-MAP INDICA (PEKING UNIVERSITY CHINA)
55	Ensembl56	PANCREATIC EXPRESSION DATABASE (INSTITUTE OF CANCER UK)
56	ENSEMBL_MART_PLANT	GRAMENE 30 ENSEMBL GENES (CSHL/CORNELL US)
57	ENSEMBL_MART_PLANT_SNP	GRAMENE 30 VARIATION (CSHL/CORNELL US)
58	GRAMENE_MARKER_30	GRAMENE 30 MARKERS (CSHL/CORNELL US)
59	GRAMENE_MAP_30	GRAMENE 30 MAPPINGS (CSHL/CORNELL US)
60	QTL_MART	GRAMENE 32 QTL DB (CSHL/CORNELL US)
61	salmosalar2_mart	UNIGENE SALMO SALAR DATABASE (CMM CHILE)
62	trucha_mart	UNIGENE ONCORHYNCHUS MYKISS DATABASE (CMM CHILE)

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 (taeGut3.2.4)	taeGut3.2.4
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 (loxAfr3)	loxAfr3
6	mlucifugus_gene_ensembl	Myotis lucifugus genes (myoLuc2)	myoLuc2
7	hsapiens_gene_ensembl	Homo sapiens genes (GRCh37.p5)	GRCh37.p5
8	choffmanni_gene_ensembl	Choloepus hoffmanni genes (choHof1)	choHof1
9	csavignyi_gene_ensembl	Ciona savignyi genes (CSAV2.0)	CSAV2.0
10	fcatus_gene_ensembl	Felis catus genes (CAT)	CAT
11	rnorvegicus_gene_ensembl	Rattus norvegicus genes (RGSC3.4)	RGSC3.4
12	ggallus_gene_ensembl	Gallus gallus genes (WASHUC2)	WASHUC2
13	tbelangeri_gene_ensembl	Tupaia belangeri genes (tupBel1)	tupBel1
14	xtropicalis_gene_ensembl	Xenopus tropicalis genes (JGI4.2)	JGI4.2
15	ecaballus_gene_ensembl	Equus caballus genes (EquCab2)	EquCab2
16	cjacchus_gene_ensembl	Callithrix jacchus genes (calJac3)	calJac3
17	pabelii_gene_ensembl	Pongo abelii genes (PPYG2)	PPYG2
18	drerio_gene_ensembl	Danio rerio genes (Zv9)	Zv9
19	stridecemlineatus_gene_ensembl	Spermophilus tridecemlineatus genes (speTri1)	speTri1
20	tnigroviridis_gene_ensembl	Tetraodon nigroviridis genes (TETRAODON8.0)	TETRAODON8.0
21	ttruncatus_gene_ensembl	Tursiops truncatus genes (turTru1)	turTru1
22	scerevisiae_gene_ensembl	Saccharomyces cerevisiae genes (EF3)	EF3
23	amelanoleuca_gene_ensembl	Ailuropoda melanoleuca genes (ailMel1)	ailMel1
24	celegans_gene_ensembl	Caenorhabditis elegans genes (WS220)	WS220
25	mmulatta_gene_ensembl	Macaca mulatta genes (MMUL_1.0)	MMUL_1.0
26	pvampyrus_gene_ensembl	Pteropus vampyrus genes (pteVam1)	pteVam1
27	mdomestica_gene_ensembl	Monodelphis domestica genes (monDom5)	monDom5
28	vpacos_gene_ensembl	Vicugna pacos genes (vicPac1)	vicPac1
29	acarolinensis_gene_ensembl	Anolis carolinensis genes (AnoCar2.0)	AnoCar2.0
30	tsyrichta_gene_ensembl	Tarsius syrichta genes (tarSyr1)	tarSyr1
31	ogarnettii_gene_ensembl	Otolemur garnettii genes (otoGar1)	otoGar1
32	trubripes_gene_ensembl	Takifugu rubripes genes (FUGU4.0)	FUGU4.0
33	dmelanogaster_gene_ensembl	Drosophila melanogaster genes (BDGP5.25)	BDGP5.25
34	pmarinus_gene_ensembl	Petromyzon marinus genes (Petromyzon_marinus_7.0)	Petromyzon_marinus_7.0
35	eeuropaeus_gene_ensembl	Erinaceus europaeus genes (eriEur1)	eriEur1
36	mmurinus_gene_ensembl	Microcebus murinus genes (micMur1)	micMur1
37	olatipes_gene_ensembl	Oryzias latipes genes (HdrR)	HdrR
38	etelfairi_gene_ensembl	Echinops telfairi genes (TENREC)	TENREC
39	cintestinalis_gene_ensembl	Ciona intestinalis genes (JGI2)	JGI2
40	ptroglodytes_gene_ensembl	Pan troglodytes genes (CHIMP2.1)	CHIMP2.1
41	oprinceps_gene_ensembl	Ochotona princeps genes (OchPri2.0)	OchPri2.0
42	ggorilla_gene_ensembl	Gorilla gorilla genes (gorGor3.1)	gorGor3.1
43	dordii_gene_ensembl	Dipodomys ordii genes (dipOrd1)	dipOrd1
44	nleucogenys_gene_ensembl	Nomascus leucogenys genes (Nleu1.0)	Nleu1.0
45	sscrofa_gene_ensembl	Sus scrofa genes (Sscrofa9)	Sscrofa9
46	mmusculus_gene_ensembl	Mus musculus genes (NCBIM37)	NCBIM37
47	ocuniculus_gene_ensembl	Oryctolagus cuniculus genes (oryCum2.0)	oryCum2.0
48	mgallopavo_gene_ensembl	Meleagris gallopavo genes (UMD2)	UMD2
49	saraneus_gene_ensembl	Sorex araneus genes (sorAra1)	sorAra1
50	dnovemcinctus_gene_ensembl	Dasypus novemcinctus genes (dasNov2)	dasNov2
51	pcapensis_gene_ensembl	Procavia capensis genes (proCap1)	proCap1
52	btaurus_gene_ensembl	Bos taurus genes (UMD3.1)	UMD3.1
53	meugenii_gene_ensembl	Macropus eugenii genes (Meug_1.0)	Meug_1.0
54	sharrisii_gene_ensembl	Sarcophilus harrisii genes (DEVIL7.0)	DEVIL7.0
55	cfamiliaris_gene_ensembl	Canis familiaris genes (CanFam_2.0)	CanFam_2.0

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
1	ensembl_gene_id	Ensembl Gene ID
2	ensembl_transcript_id	Ensembl Transcript ID
3	ensembl_peptide_id	Ensembl Protein ID
4	canonical_transcript_stable_id	Canonical transcript stable ID(s)
5	description	Description

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, 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 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("GO:0051330", "GO:0000080", "GO: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=
```

```
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, Dwarfina-type
7  NM_005359  IPR003619          MAD homology 1, Dwarfina-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                   ENSG00000181791  
2           214555_at ENSG00000162009  
3                   ENSG00000162009  
4                   ENSG00000184471  
5           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           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 EntrezGene 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
MTAIIKEIVSRNKRR ... 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'), 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_dna","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("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

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] " [IG_C_gene,IG_C_pseudogene,IG_D_gene,IG_J_gene,IG_J_pseudogene,IG_V_gene,IG_V_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 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"          "transcript_event"  "homologs"          "snp"
```

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	
1	ensembl_gene_id	Ensembl Gene ID
2	ensembl_transcript_id	Ensembl Transcript ID
3	ensembl_peptide_id	Ensembl Peptide ID
4	canonical_transcript_stable_id	Canonical transcript stable ID
5	description	
6	chromosome_name	Chromosome Name
7	start_position	Gene Start Position
8	end_position	Gene End Position
9	strand	Gene Strand

10	band	
11	transcript_start	Transcrip
12	transcript_end	Transcr
13	external_gene_id	Associat
14	external_transcript_id	Associated Tra
15	external_gene_db	Associ
16	transcript_db_name	Associated T
17	transcript_count	Tran
18	percentage_gc_content	
19	gene_biotype	
20	transcript_biotype	Transc
21	source	
22	status	S
23	transcript_status	Status
24	go_id	GO Ter
25	name_1006	
26	definition_1006	GO Term
27	go_linkage_type	GO Term E
28	namespace_1003	
29	goslim_goa_accession	GOSlim GOA .
30	goslim_goa_description	GOSlim GOA
31	ox_pubmed_dm_dbprimary_acc_1074	
32	ucsc	
33	pdb	
34	clone_based_ensembl_gene_name	Clone based Ensem
35	clone_based_ensembl_transcript_name	Clone based Ensembl tra
36	clone_based_vega_gene_name	Clone based VEG
37	clone_based_vega_transcript_name	Clone based VEGA tra
38	ccds	
39	embl	EMBL
40	ens_hs_gene	Ensembl to LRG l
41	ens_hs_transcript	Ensembl to LRG link tr
42	ens_hs_translation	Ensembl to LRG link tra
43	ox_ens_lrg_transcript_dm_dbprimary_acc_1074	LRG to Ensembl lin
44	entrezgene	E
45	ottt	VEGA transcript
46	ottg	VEGA gene
47	shares_cds_with_ens	Ensembl transcript (where OTTT shares CD
48	shares_cds_with_ottt	HAVANA transcript (where ENST shares CD
49	shares_cds_and_utr_with_ottt	HAVANA transcript (where ENST identi
50	hgnc_id	
51	hgnc_symbol	
52	hgnc_transcript_name	HGNC tra
53	ipi	
54	merops	

55	mim_morbid_accession	MIM Morb
56	mim_morbid_description	MIM Morbid
57	mim_gene_accession	MIM Ge
58	mim_gene_description	MIM Gene
59	mirbase_accession	miRBase .
60	mirbase_id	m
61	mirbase_gene_name	miRBA
62	mirbase_transcript_name	miRBase tra
63	protein_id	Protein
64	refseq_peptide	RefSe
65	refseq_peptide_predicted	RefSeq Predicted
66	refseq_genomic	RefSeq G
67	rfam	
68	rfam_gene_name	Rf
69	rfam_transcript_name	Rfam tra
70	unigene	
71	uniprot_sptrembl	UniProt/TrEM
72	uniprot_swissprot	UniProt/
73	uniprot_swissprot_accession	UniProt/SwissPr
74	uniprot_genename	UniPr
75	wikigene_name	W
76	wikigene_description	WikiGene
77	hpa	Human Protein Atlas
78	dbass3_id	Database of Aberrant 3' Splice Sites
79	dbass3_name	DBAS
80	dbass5_id	Database of Aberrant 5' Splice Sites
81	dbass5_name	DBAS
82	ox_refseq_mrna__dm_dbprimary_acc_1074	
83	ox_refseq_mrna_predicted__dm_dbprimary_acc_1074	RefSeq mR
84	ox_refseq_ncrna__dm_dbprimary_acc_1074	.
85	ox_refseq_ncrna_predicted__dm_dbprimary_acc_1074	RefSeq ncr
86	affy_hc_g110	.
87	affy_hg_focus	A
88	affy_hg_u133_plus_2	Affy HG
89	affy_hg_u133a_2	Aff
90	affy_hg_u133a	A
91	affy_hg_u133b	A
92	affy_hg_u95av2	Af
93	affy_hg_u95b	.
94	affy_hg_u95c	.
95	affy_hg_u95d	.
96	affy_hg_u95e	.
97	affy_hg_u95a	.
98	affy_hugenefl	Af
99	affy_huex_1_0_st_v2	Affy Hu

100	affy_hugene_1_0_st_v1	Affy HuGe
101	affy_u133_x3p	A
102	agilent_cgh_44b	Agi
103	agilent_wholegenome	Agilent
104	codelink	
105	illumina_humanwg_6_v1	Illumina
106	illumina_humanwg_6_v2	Illumina
107	illumina_humanwg_6_v3	Illumina
108	illumina_humanht_12	Illumina
109	phalanx_onearray	Phal
110	anatomical_system	Anatomical System
111	development_stage	Development Stage
112	cell_type	Cell Type
113	pathology	Pathology
114	atlas_celltype	GNF/Atl
115	atlas_diseasestate	GNF/Atlas d
116	atlas_organismpart	GNF/Atlas o
117	family_description	Ensembl Family
118	family	Ensembl Protein
119	pirsf	PIRSF Su
120	superfamily	Su
121	smart	
122	profile	
123	prints	
124	pfam	
125	tigrfam	
126	protein_feature_seg__dm_hit_name_1048	
127	interpro	
128	interpro_short_description	Interpro Short
129	interpro_description	Interpro
130	transmembrane_domain	Transmem
131	signal_domain	S
132	ncoils	

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 Session Info

```
> sessionInfo()
```

```
R version 2.14.0 (2011-10-31)
```

```
Platform: x86_64-unknown-linux-gnu (64-bit)
```

```
locale:
```

```
[1] LC_CTYPE=en_US.UTF-8      LC_NUMERIC=C               LC_TIME=en_US.UTF-8
```

```
[5] LC_MONETARY=en_US.UTF-8   LC_MESSAGES=en_US.UTF-8   LC_PAPER=C
```

```
[9] LC_ADDRESS=C              LC_TELEPHONE=C            LC_MEASUREMENT=en_US.UTF-8
```

```
attached base packages:
```

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

```
other attached packages:
```

```
[1] biomaRt_2.10.0
```

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

```
[1] Rcurl_1.6-10 XML_3.4-3    tools_2.14.0
```

```
> warnings()
```

```
NULL
```