

The biomaRt user's guide

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April 16, 2015

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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
1      ensembl
2              snp
3      regulation
4              vega
5      fungi_mart_26
6      fungi_variations_26
7      metazoa_mart_26
8      metazoa_variations_26
9      plants_mart_26
10     plants_variations_26
11     protists_mart_25
12     protists_variations_26
13             msd
14     cg_mart_02
15             htgt
16             WS220
17     parasite_mart
18             biomart
19             example
20             pride
21     prod-intermart_1
22             unimart
23             biomartDB
24             biblioDB
25     Eurexpress Biomart
26     phytozome_mart
27     metazome_mart
28     HapMap_rel27
29     oncomodules
30             ikmc
31     EMAGE gene expression
32     EMAP anatomy ontology
33     EMAGE browse repository
```

```

34                                     GermOnline
35   Sigenae_Oligo_Annotation_Ensembl_61
36 Sigenae Oligo Annotation (Ensembl 59)
37 Sigenae Oligo Annotation (Ensembl 56)
38       Breast_mart_69
39           K562_Gm12878
40           Hsmm_Hmec
41           Pancreas63
42       Public_OBIOMARTPUB
43           Public_VITIS
44       Public_VITIS_12x
45           Prod_WHEAT
46       Public_TAIRV10
47           Public_MAIZE
48           Prod_TOMATO
49           Prod_POPLAR
50       Prod_POPLAR_V2
51       Prod_BOTRYTISEDIT
52           Prod_BOFUB
53       Prod_LMACULANSEdit
54       vb_gene_mart_1502
55       vb_snp_mart_1502
56           expression
57       ENSEMBL_MART_PLANT
58       ENSEMBL_MART_PLANT_SNP

```

```

version
1                                     ENSEMBL GENES 79 (SANGER UK)
2                                     ENSEMBL VARIATION 79 (SANGER UK)
3                                     ENSEMBL REGULATION 79 (SANGER UK)
4                                     VEGA 59 (SANGER UK)
5                                     ENSEMBL FUNGI 26 (EBI UK)
6                                     ENSEMBL FUNGI VARIATION 26 (EBI UK)
7                                     ENSEMBL METAZOA 26 (EBI UK)
8                                     ENSEMBL METAZOA VARIATION 26 (EBI UK)
9                                     ENSEMBL PLANTS 26 (EBI UK)
10                                    ENSEMBL PLANTS VARIATION 26 (EBI UK)
11                                    ENSEMBL PROTISTS 25 (EBI UK)
12                                    ENSEMBL PROTISTS VARIATION 26 (EBI UK)
13                                    MSD (EBI UK)
14                                    PROTEOMICS (UNIVERSITY OF CAMBRIDGE - UK)
15                                    WTSI MOUSE GENETICS PROJECT (SANGER UK)
16                                    WORMBASE 220 (CSHL US)
17                                    ParaSite Mart (EBI UK)
18                                    MGI (JACKSON LABORATORY US)
19                                    FANTOM5 phase1.1 (RIKEN CSLST Japan)
20                                    PRIDE (EBI UK)
21                                    INTERPRO (EBI UK)
22                                    UNIPROT (EBI UK)
23                                    PARAMECIUM GENOME (CNRS FRANCE)
24                                    PARAMECIUM BIBLIOGRAPHY (CNRS FRANCE)
25                                    EUREXPRESS (MRC EDINBURGH UK)
26                                    Phytozome
27                                    Metazome
28                                    HAPMAP 27 (NCBI US)
29                                    INTOGEN ONCOMODULES
30                                    IKMC GENES AND PRODUCTS (IKMC)

```

```

31                                                     EMAGE GENE EXPRESSION
32                                                     EMAP ANATOMY ONTOLOGY
33                                                     EMAGE BROWSE REPOSITORY
34                                                     GERMONLINE
35                                                     SIGENAE OLIGO ANNOTATION (ENSEMBL 61)
36                                                     SIGENAE OLIGO ANNOTATION (ENSEMBL 59)
37                                                     SIGENAE OLIGO ANNOTATION (ENSEMBL 56)
38                                                     BCCTB Bioinformatics Portal (UK and Ireland)
39 Predictive models of gene regulation from processed high-throughput epigenomics data: K562 vs. Gm12878
40 Predictive models of gene regulation from processed high-throughput epigenomics data: Hsmm vs. Hmec
41                                                     PANCREATIC EXPRESSION DATABASE (BARTS CANCER INSTITUTE UK)
42 Multi-species: marker, QTL, SNP, gene, germplasm, phenotype, association, with Gene annotations
43 Grapevine 8x, stuctural annotation with Genetic maps (genetic markers..)
44 Grapevine 12x.0, stuctural and functional annotation with Genetic maps (genetic markers..)
45 Wheat, stuctural annotation with Genetic maps (genetic markers..)
46 Arabidopsis Thaliana TAIRV10, genes functional annotation
47 Zea mays ZmB73, genes functional annotation
48 Tomato, stuctural and functional annotation
49 Populus trichocarpa, genes functional annotation
50 Populus trichocarpa, genes functional annotation V2.0
51 Botrytis cinerea T4, genes functional annotation
52 Botrytis cinerea B0510, genes functional annotation
53 Leptosphaeria maculans, genes functional annotation
54 VectorBase Genes
55 VectorBase Variation
56 VectorBase Expression
57 GRAMENE 40 ENSEMBL GENES (CSHL/CORNELL US)
58 GRAMENE 40 VARIATION (CSHL/CORNELL US)

```

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")
```

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	lafricana_gene_ensembl	Loxodonta africana genes (loxAfr3)	loxAfr3
5	itridecemlineatus_gene_ensembl	Ictidomys tridecemlineatus genes (spetri2)	spetri2
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_5.0)	Rnor_5.0
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	trubripes_gene_ensembl	Takifugu rubripes genes (FUGU4.0)	FUGU4.0
18	amexicanus_gene_ensembl	Astyanax mexicanus genes (AstMex102)	AstMex102
19	pmarinus_gene_ensembl	Petromyzon marinus genes (Pmarinus_7.0)	Pmarinus_7.0
20	eeuropaeus_gene_ensembl	Erinaceus europaeus genes (eriEur1)	eriEur1
21	falbicollis_gene_ensembl	Ficedula albicollis genes (FicAlb_1.4)	FicAlb_1.4
22	ptroglodytes_gene_ensembl	Pan troglodytes genes (CHIMP2.1.4)	CHIMP2.1.4
23	etelfairi_gene_ensembl	Echinops telfairi genes (TENREC)	TENREC
24	cintestinalis_gene_ensembl	Ciona intestinalis genes (KH)	KH
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	Dasypus novemcinctus genes (Dasnov3.0)	Dasnov3.0
29	pcapensis_gene_ensembl	Procapia 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.p2)	GRCh38.p2
33	pformosa_gene_ensembl	Poecilia formosa genes (PoeFor_5.1.2)	PoeFor_5.1.2
34	mfuro_gene_ensembl	Mustela putorius furo genes (MusPutFur1.0)	MusPutFur1.0
35	tbelangeri_gene_ensembl	Tupaia belangeri genes (tupBel1)	tupBel1
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	xmaculatus_gene_ensembl	Xiphophorus maculatus genes (Xipmac4.4.2)	Xipmac4.4.2
41	drerio_gene_ensembl	Danio rerio genes (Zv9)	Zv9
42	lchalumnae_gene_ensembl	Latimeria chalumnae genes (LatChal1)	LatChal1
43	tnigroviridis_gene_ensembl	Tetraodon nigroviridis genes (TETRAODON8.0)	TETRAODON8.0
44	amelanoleuca_gene_ensembl	Ailuropoda melanoleuca genes (ailMel1)	ailMel1
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	tsyricha_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	ggorilla_gene_ensembl	Gorilla gorilla genes (gorGor3.1)	gorGor3.1
58	oprinceps_gene_ensembl	Ochotona princeps genes (OchPri2.0)	OchPri2.0
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 (GRCm38.p3)	GRCm38.p3
62	mgallopavo_gene_ensembl	Meleagris gallopavo genes (UMD2)	UMD2
63	gmorhua_gene_ensembl	Gadus morhua genes (gadMor1)	gadMor1
64	aplatyrhynchos_gene_ensembl	Anas platyrhynchos genes (BGI_duck_1.0)	BGI_duck_1.0
65	saraneus_gene_ensembl	Sorex araneus genes (sorAra1)	sorAra1
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
1	ensembl_gene_id	Ensembl Gene ID
2	ensembl_transcript_id	Ensembl Transcript ID
3	ensembl_peptide_id	Ensembl Protein ID
4	ensembl_exon_id	Ensembl Exon ID
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 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_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, 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                    ENSG00000260702
2          215502_at  ENSG00000260532
3                    ENSG00000273551
4          217023_x_at  ENSG00000172236
5          210084_x_at  ENSG00000172236
6          215382_x_at  ENSG00000172236
7          207741_x_at  ENSG00000172236
8          216474_x_at  ENSG00000172236
9          207134_x_at  ENSG00000172236
10         205683_x_at  ENSG00000172236
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                    ENSG00000261294
18                    ENSG00000259910
19         220339_s_at  ENSG00000116176
20                    ENSG00000260403
21                    ENSG00000277010
22         205845_at  ENSG00000196557
23                    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 EntrzGene 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_47  ENSEMBL GENES 47 (SANGER)
2 genomic_features_mart_47  Genomic Features
3      snp_mart_47      SNP
4      vega_mart_47      Vega
5 compara_mart_homology_47  Compara homology
6 compara_mart_multiple_ga_47  Compara multiple alignments
7 compara_mart_pairwise_ga_47  Compara pairwise alignments
8      ensembl_mart_46  ENSEMBL GENES 46 (SANGER)
9 genomic_features_mart_46  Genomic Features
10      snp_mart_46      SNP
11      vega_mart_46      Vega
12 compara_mart_homology_46  Compara homology
13 compara_mart_multiple_ga_46  Compara multiple alignments
14 compara_mart_pairwise_ga_46  Compara pairwise alignments
15      ensembl_mart_45  ENSEMBL GENES 45 (SANGER)
16      snp_mart_45      SNP
```

```

17          vega_mart_45                      Vega
18  compara_mart_homology_45                  Compara homology
19  compara_mart_multiple_ga_45              Compara multiple alignments
20  compara_mart_pairwise_ga_45              Compara pairwise alignments
21          ensembl_mart_44                  ENSEMBL GENES 44 (SANGER)
22          snp_mart_44                      SNP
23          vega_mart_44                      Vega
24  compara_mart_homology_44                  Compara homology
25  compara_mart_pairwise_ga_44              Compara pairwise alignments
26          ensembl_mart_43                  ENSEMBL GENES 43 (SANGER)
27          snp_mart_43                      SNP
28          vega_mart_43                      Vega
29  compara_mart_homology_43                  Compara homology
30  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	GR0 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,IG_C_gene,IG_C_pseudogene,IG_D_gene,IG_J_gene,IG_J_p
```

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"      "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
2	ensembl_transcript_id	Ensembl Transcript ID
3	ensembl_peptide_id	Ensembl Protein ID
4	ensembl_exon_id	Ensembl Exon ID
5	description	Description
6	chromosome_name	Chromosome Name
7	start_position	Gene Start (bp)
8	end_position	Gene End (bp)
9	strand	Strand

10	band	Band
11	transcript_start	Transcript Start (bp)
12	transcript_end	Transcript End (bp)
13	transcription_start_site	Transcription Start Site (TSS)
14	transcript_length	Transcript length
15	transcript_tsl	Transcript Support Level (TSL)
16	transcript_gencode_basic	GENCODE basic annotation
17	external_gene_name	Associated Gene Name
18	external_gene_source	Associated Gene Source
19	external_transcript_name	Associated Transcript Name
20	external_transcript_source_name	Associated Transcript Source
21	transcript_count	Transcript count
22	translation_count	Translation count
23	percentage_gc_content	% GC content
24	gene_biotype	Gene type
25	transcript_biotype	Transcript type
26	source	Source (gene)
27	transcript_source	Source (transcript)
28	status	Status (gene)
29	transcript_status	Status (transcript)
30	version	Version (gene)
31	transcript_version	Version (transcript)
32	phenotype_description	Phenotype description
33	source_name	Source name
34	study_external_id	Study External Reference
35	go_id	GO Term Accession
36	name_1006	GO Term Name
37	definition_1006	GO Term Definition
38	go_linkage_type	GO Term Evidence Code
39	namespace_1003	GO domain
40	goslim_goa_accession	GOSlim GOA Accession(s)
41	goslim_goa_description	GOSlim GOA Description
42	arrayexpress	ArrayExpress
43	chembl	ChEMBL ID(s)
44	clone_based_ensembl_gene_name	Clone based Ensembl gene name
45	clone_based_ensembl_transcript_name	Clone based Ensembl transcript name
46	clone_based_vega_gene_name	Clone based VEGA gene name
47	clone_based_vega_transcript_name	Clone based VEGA transcript name
48	ccds	CCDS ID
49	dbass3_id	Database of Aberrant 3' Splice Sites (DBASS3) IDs
50	dbass3_name	DBASS3 Gene Name
51	dbass5_id	Database of Aberrant 5' Splice Sites (DBASS5) IDs
52	dbass5_name	DBASS5 Gene Name
53	embl	EMBL (Genbank) ID
54	ens_hs_gene	Ensembl Human Gene IDs

55	ens_hs_transcript	Ensembl Human Transcript IDs
56	ens_hs_translation	Ensembl Human Translation IDs
57	ens_lrg_gene	LRG to Ensembl link gene
58	ens_lrg_transcript	LRG to Ensembl link transcript
59	entrezgene	EntrezGene ID
60	entrezgene_transcript_name	EntrezGene transcript name ID
61	hpa	Human Protein Atlas Antibody ID
62	ottg	VEGA gene ID(s) (OTTG)
63	ottd	VEGA transcript ID(s) (OTTT)
64	ottp	VEGA protein ID(s) (OTTP)
65	hgnc_id	HGNC ID(s)
66	hgnc_symbol	HGNC symbol
67	hgnc_transcript_name	HGNC transcript name
68	merops	MEROPS ID
69	mim_morbid_accession	MIM Morbid Accession
70	mim_morbid_description	MIM Morbid Description
71	mim_gene_accession	MIM Gene Accession
72	mim_gene_description	MIM Gene Description
73	mirbase_accession	miRBase Accession(s)
74	mirbase_id	miRBase ID(s)
75	mirbase_transcript_name	miRBase transcript name
76	pdb	PDB ID
77	protein_id	Protein (Genbank) ID
78	reactome	Reactome ID
79	refseq_mrna	RefSeq mRNA [e.g. NM_001195597]
80	refseq_mrna_predicted	RefSeq mRNA predicted [e.g. XM_001125684]
81	refseq_ncrna	RefSeq ncRNA [e.g. NR_002834]
82	refseq_ncrna_predicted	RefSeq ncRNA predicted [e.g. XR_108264]
83	refseq_peptide	RefSeq Protein ID [e.g. NP_001005353]
84	refseq_peptide_predicted	RefSeq Predicted Protein ID [e.g. XP_001720922]
85	rfam	Rfam ID
86	rfam_transcript_name	Rfam transcript name
87	rnacentral	RNACentral ID
88	ucsc	UCSC ID
89	unigene	Unigene ID
90	uniparc	UniParc
91	uniprot_sptrembl	UniProt/TrEMBL ID
92	uniprot_swissprot	UniProt/SwissProt ID
93	uniprot_genename	UniProt Gene Name
94	uniprot_genename_transcript_name	UniProt Genename Transcript Name
95	wikigene_name	WikiGene Name
96	wikigene_id	WikiGene ID
97	wikigene_description	WikiGene Description
98	efg_agilent_sureprint_g3_ge_8x60k	Agilent SurePrint G3 GE 8x60k probe
99	efg_agilent_sureprint_g3_ge_8x60k_v2	Agilent SurePrint G3 GE 8x60k v2 probe

100	efg_agilent_wholegenome_4x44k_v1	Agilent WholeGenome 4x44k v1 probe
101	efg_agilent_wholegenome_4x44k_v2	Agilent WholeGenome 4x44k v2 probe
102	affy_hc_g110	Affy HC G110 probeset
103	affy_hg_focus	Affy HG FOCUS probeset
104	affy_hg_u133_plus_2	Affy HG U133-PLUS-2 probeset
105	affy_hg_u133a_2	Affy HG U133A_2 probeset
106	affy_hg_u133a	Affy HG U133A probeset
107	affy_hg_u133b	Affy HG U133B probeset
108	affy_hg_u95av2	Affy HG U95AV2 probeset
109	affy_hg_u95b	Affy HG U95B probeset
110	affy_hg_u95c	Affy HG U95C probeset
111	affy_hg_u95d	Affy HG U95D probeset
112	affy_hg_u95e	Affy HG U95E probeset
113	affy_hg_u95a	Affy HG U95A probeset
114	affy_hugenefl	Affy HuGene FL probeset
115	affy_primeview	Affy primeview
116	affy_u133_x3p	Affy U133 X3P probeset
117	agilent_cgh_44b	Agilent CGH 44b probe
118	codelink	Codelink probe
119	illumina_humanwg_6_v1	Illumina HumanWG 6 v1 probe
120	illumina_humanwg_6_v2	Illumina HumanWG 6 v2 probe
121	illumina_humanwg_6_v3	Illumina HumanWG 6 v3 probe
122	illumina_humanht_12_v3	Illumina Human HT 12 V3 probe
123	illumina_humanht_12_v4	Illumina Human HT 12 V4 probe
124	illumina_humanref_8_v3	Illumina Human Ref 8 V3 probe
125	phalanx_onearray	Phalanx OneArray probe
126	family	Ensembl Protein Family ID(s)
127	family_description	Ensembl Family Description
128	pirsf	PIRSF ID
129	superfamily	SUPERFAMILY ID
130	smart	SMART ID
131	profile	Pfscan ID
132	prints	PRINTS ID
133	pfam	Pfam ID
134	tigrfam	TIGRFAM ID
135	interpro	Interpro ID
136	interpro_short_description	Interpro Short Description
137	interpro_description	Interpro Description
138	low_complexity	low complexity (SEG)
139	transmembrane_domain	Transmembrane domain (tmhmm)
140	signal_domain	signal peptide
141	ncoils	coiled coil (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 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] "chromosome_name" "start"          "end"

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

[1] "ensembl_gene_id"      "ensembl_transcript_id" "ensembl_peptide_id"
```

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)

[1] "LRG_1"   "LRG_10"  "LRG_100"
```

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          202763_at          836
2          207500_at          838
3          209310_s_at          837
```

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 3.2.0 (2015-04-16)
```

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

```
Running under: Ubuntu 14.04.2 LTS
```

```
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=en_US.UTF-8
[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_2.24.0
```

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

[1] IRanges_2.2.0	DBI_0.3.1	parallel_3.2.0	tools_3.2.0
[6] Biobase_2.28.0	AnnotationDbi_1.30.0	RSQLite_1.0.0	S4Vectors_0.6.0
[11] GenomeInfoDb_1.4.0	stats4_3.2.0	bitops_1.0-6	XML_3.98-1.1

```
> warnings()
```

```
NULL
```