# An R interface to the Ensembl REST API

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		11 0	14					
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		· ·	15					
		ı ,	15					
	2.8	1 , 0	16					
	0		16					
			16					

# 1 Introduction

This package uses the Ensembl REST  $API^1$  (currently in beta) to extract data from Ensembl into R. As the REST API is in Beta, this package should also be considered to be in flux and functions/parameters/etc are subject to change as things get finalized.

It could also do with your help. If you find a problem, something you think could be better, or a better way of doing things, please consider visiting the GitHub project at https://github.com/acbb/EnsemblRest and posting an issue or a Pull Request. Thanks!

# 2 Available Methods

To begin with (assuming you have installed this package), you need to load it into your R session:

> library( EnsemblRest )

The following subsections then list the methods available to you.

<sup>1</sup>http://beta.rest.ensembl.org/

#### 2.1 Information

#### 2.1.1 isAlive

Firstly, we can check to see that the REST API is accepting calls:

```
> isAlive()
[1] TRUE
```

## 2.1.2 infoSpecies

To get a list of available species on the server, you can use the infoSpecies call<sup>2</sup>.

```
> infoSpecies()[1:3] # Just the first 3
[[1]]
name
       : saccharomyces_cerevisiae
aliases: 4932, saccer, saccharomyces cerevisiae (baker's yeast), baker's yeast, scer, sacchar
groups : core, otherfeatures, variation, funcgen
release: 70
[[2]]
       : ciona_savignyi
aliases : ciosav, 51511, ciona savignyi, csavignyi, c.savignyi, csav, sea squirt ciona savigny
groups : core, otherfeatures
release: 70
[[3]]
        : myotis_lucifugus
aliases: little brown bat, mlucifugus, myoluc, mluc, 59463, myotis lucifugus, myotis_lucifugu
groups : core, otherfeatures
release: 70
```

## 2.1.3 infoAssembly

The infoAssembly call<sup>3</sup> returns information about the currently available assemblies in the given species.

```
> infoAssembly( 'human' )
assembly_name
                              : GRCh37.p10
assembly_date
                               : 2009-02
                             : , GRCh37, NCBI36, NCBI34, NCBI35
coord_system_versions
schema_build
                              : 70_37
                              : 2010-07-Ensembl
genebuild_start_date
genebuild_initial_release_date : 2011-04
genebuild_last_geneset_update : 2013-01
genebuild_method
                               : full_genebuild
                               : 1, 10, 11, 12, 13, 14, 15, 16, 17, 18, 19, 2, 20, 21, 22, 3,
top_level_seq_region_names
```

 $<sup>{}^2</sup> h ttp://beta.rest.ensembl.org/documentation/info/species$ 

<sup>&</sup>lt;sup>3</sup>http://beta.rest.ensembl.org/documentation/info/assembly\_info

## 2.1.4 assemblyDetails

The assemblyDetails call<sup>4</sup> returns information about one of these assemblies.

```
> assemblyDetails('X', 'human')
is_chromosome : TRUE
length : 155270560
assembly_exception_type : REF
coordinate_system : chromosome
```

# 2.1.5 infoComparas

The infoComparas call<sup>5</sup> lists the available comparative genomics databases.

```
> infoComparas()
multi
  "70"
```

#### 2.1.6 infoData

The infoData call<sup>6</sup> shows the data releases available to the REST service

```
> infoData()
[1] 70
```

# 2.1.7 infoRest

infoRest shows the current version<sup>7</sup> of the REST service

```
> infoRest()
[1] "1.2.0"
```

#### 2.1.8 infoSoftware

And finally in the info section, infoSoftware shows<sup>8</sup> the current version of the Ensembl API.

```
> infoSoftware()
[1] 70
```

<sup>4</sup>http://beta.rest.ensembl.org/documentation/info/assembly\_stats
5http://beta.rest.ensembl.org/documentation/info/comparas
6http://beta.rest.ensembl.org/documentation/info/data
7http://beta.rest.ensembl.org/documentation/info/rest
8http://beta.rest.ensembl.org/documentation/info/software

# 2.2 Comparative Genomics

#### 2.2.1 geneTree

This method<sup>9</sup> fetches the gene tree in New Hampshire format for a given Ensembl gene tree identifier.

## 2.2.2 homologyById

When given an Ensembl Gene ID, returns the homology<sup>10</sup> information for it.

```
> hResponse = homologyById( 'ENSG00000170037' )
> hResponse
                            # The response object
$ENSG00000170037
$id: ENSG00000170037 $homologies: 17 homologies
> hResponse[[1]]$homologies[1:2,] # Just the top 2 homologies to save room
  source.perc_pos
1
               99
2
               99
1 MATSADSPSSPLGAEDLLSDSSEPPGLNQVSSEVTSQLYASLRLSRQAEATARAQLYLPSTSPPHEGLDGFAQELSRSLSVGLEKNLKKKDG
2\ {\tt MATSADSPSSPLGAEDLLSDSSEPPGLNQVSSEVTSQLYASLRLSRQAEATARAQLYLPSTSPPHEGLDGFAQELSRSLSVGLEKNLKKKDG}
  source.protein_id source.perc_id source.cigar_line source.species
                                                 925M homo_sapiens
   ENSP00000369614
                                99
1
   ENSP00000369614
                                98
                                                 925M
                                                        homo_sapiens
        source.id dn_ds target.perc_pos
1 ENSG00000170037 0.29412
                                       100
2 ENSG00000170037
                                        99
1 MATSADSPSSPLGAEDLLSDSSEPPGLNQVSSEVTSQLYASLRLSRQAEATARAQLYLPSTSPPHEGLDGLAQELSRSLSVGLENNLKKKDG
2\ \mathsf{MATSADSPSSPLGAEDLLSDSSEPPGLNQVSSEVTSQLYASLRLSRQAEATARAQLYLPSTSPPHEGLDGLAQELSRSLSVGLENNLKKKDG
   target.protein_id target.perc_id target.cigar_line target.species
1 ENSPTRP00000014861
                                 100
                                              850MD74M pan_troglodytes
2 ENSGGOP0000006314
                                                  925M gorilla_gorilla
           target.id
                       subtype
1 ENSPTRG00000008719 Homininae ortholog_one2one
```

#### 2.2.3 homologyBySymbol

You can also retrieve homology information<sup>11</sup> given a symbol and a species;

2 ENSGGOGOOOOOO6451 Homininae ortholog\_one2one

<sup>9</sup>http://beta.rest.ensembl.org/documentation/info/rest

<sup>10</sup>http://beta.rest.ensembl.org/documentation/info/homology\_ensemblgene

<sup>11</sup>http://beta.rest.ensembl.org/documentation/info/homology\_symbol

```
> hResponse = homologyBySymbol( 'BRCA2', 'human' )
> hResponse
                             # The response object
$BRCA2
$id: ENSG00000139618 $homologies: 17 homologies
> hResponse[[1]] $homologies[1:2,] # Again, just the top 2 homologies to save room
  source.perc_pos
1
                99
2
                98
1 MPIGSKERPTFFEIFKTRCNKADLGPISLNWFEELSSEAPPYNSEPAEESEHKNNNYEPNLFKTPQRKPSYNQLASTPIIFKEQGLTLPLYQ
2 MPIGSKERPTFFEIFKTRCNKADLGPISLNWFEELSSEAPPYNSEPAEESEHKNNNYEPNLFKTPQRKPSYNQLASTPIIFKEQGLTLPLYQ
  source.protein_id source.perc_id source.cigar_line source.species
1
    ENSP00000439902
                                 99
                                                3418M
                                                        homo_sapiens
    ENSP00000439902
                                                3418M homo_sapiens
                                 96
         source.id dn_ds target.perc_pos
 1 ENSG00000139618 0.29371
 2 ENSG00000139618 0.61742
1 MPIGSKERPTFFEIFKTRCNKADLGPISLNWFEELSSEAPPYNSEPAEESEHKNNNYEPNLFKTPQRKPSYNQLASTPIIFKEQGLTLPLYQ
 2 MPVGSKERPTFFEIFKTRCNKADLGPISLHWFEELSSEAPPYNSEPAEESEHKNNNYEPNLFKTPQRKPSYNQLASTPIIFKEQGLTLPLYQ
                                        target.cigar_line target.species
    target.protein_id target.perc_id
1 ENSPTRP00000009812
                                  99
                                                    3418M pan_troglodytes
2 ENSPPYP00000005997
                                  96 975MD302MD442MD1696M
                                                             pongo_abelii
            target.id
                        subtype
                                            type
1 ENSPTRG00000005766 Homininae ortholog_one2one
 2 ENSPPYG00000005264 Hominidae ortholog_one2one
And using format='condensed', you can get a more condensed result:
> hResponse = homologyBySymbol( 'BRCA2', 'human', format='condensed' )
> hResponse
                             # The response object
$id: ENSG00000139618 $homologies: 4 homologies
> hResponse[[1]]$homologies[1:10,]
                        protein_id
     Homininae ENSPTRP00000009812 ENSPTRG00000005766 ortholog_one2one
1
2
     Hominidae ENSPPYP00000005997 ENSPPYG00000005264 ortholog_one2one
3
   Hominoidea ENSNLEP00000001277 ENSNLEG00000001048 ortholog_one2one
4
    Homininae ENSGGDP00000015446 ENSGGDG00000015808 ortholog_one2one
    Catarrhini ENSMMUP00000009432 ENSMMUG00000007197 ortholog_one2one
5
6 Simiiformes ENSCJAP00000034250 ENSCJAG00000018462 ortholog_one2one
      Primates ENSMICP00000010933 ENSMICG00000011994 ortholog_one2one
7
8 Haplorrhini ENSTSYP00000000441 ENSTSYG00000000478 ortholog_one2one
9
      Eutheria ENSECAP00000013146 ENSECAG00000014890 ortholog_one2one
      Primates ENSOGAP00000009477 ENSOGAG00000010588 ortholog_one2one
10
```

# 2.3 Cross References

Cross references are links to other data about the object of interest. It should be noted that these other data hold different fields and datatypes, so the results are returned in a data.frame containing a superset of column names, with non-applicable columns for a given result filled with <NA>.

As a single object may have multiple synonyms, this will cause the object to exist in multiple rows, one for each synonym.

# 2.3.1 xrefsByld

4

5

6

LIP8

PP1221

Firstly we can get all external references<sup>12</sup> for a given Ensembl ID:

> 2	xrefsById( 'ENSGOO	0000170037')					
			display_id		primary_id	version	
1			MG00000172932			2	
2		EN	ISG00000170037	ENSG	00000170037	0	
3			CNTROB		116840	0	
4			CNTROB		29616	0	
5			CNTROB		29616	0	
6	CENTROSOMAL BRCA	A2-INTERACTING P			611425	0	
7			Hs.348012		Hs.348012	0	
8			Hs.732863		Hs.732863	0	
9			CNTROB		CNTROB	0	
10			CNTROB		116840	0	
	dbname	info_type	info	_text	db_display		
1	OTTG	NONE			Havana	•	
2	ArrayExpress	DIRECT			ArrayEx	-	
3	EntrezGene	DEPENDENT				ezGene	
4	HGNC		Generated via		HGNC S	•	
5	HGNC		Generated via	ccds	HGNC S	•	
6	MIM_GENE	DEPENDENT				I gene	
7		SEQUENCE_MATCH				niGene	
8		SEQUENCE_MATCH				niGene	
9	Uniprot_genename	DEPENDENT		Un	iProtKB Gene		
10	WikiGene	DEPENDENT			Wik	riGene	
1							descripti <n2< td=""></n2<>
2							
3			ce	ntrobin,	centrosomal	BRCA2	interacting prote
4			ce	ntrobin,	centrosomal	BRCA2	interacting prote
5			ce				interacting prote
6				CENTROS	OMAL BRCA2-I	NTERACT	ING PROTEIN; CNTR
7			Ce	ntrobin,	centrosomal	BRCA2	interacting prote
8	Transcribed locus	s, moderately si	milar to NP_4	44279.2	centrobin is	soform al	lpha [Homo sapien:
9							
10	armanima angombi	identity engemb					interacting prote
1	synonyms ensembl_	_identity ensemb NA	NA	NA	rei_end ensem NA	NA NA	
2	<na></na>	NA NA	NA NA	NA NA	NA NA	NA NA	
3	LIP8	NA NA	NA NA	NA NA	NA NA	NA NA	
J	LIFO	IVA	IVA	IVA	IVA	IVA	

 $<sup>^{12} \</sup>verb|http://beta.rest.ensembl.org/documentation/info/xref_id$ 

NA

7	<1	JA>	99	1	32	3794	3769
8	<1	IA>	99	1	6	752	752
9	L	[P8	NA	NA	NA	NA	NA
10	<1	1A>	NA	NA	NA	NA	NA
	score	cigar_line	${\tt xref\_identity}$				
1	NA	<na></na>	NA				
2	NA	<na></na>	NA				
3	NA	<na></na>	NA				
4	NA	<na></na>	NA				
5	NA	<na></na>	NA				
6	NA	<na></na>	NA				
7	18783	376M6D3387M	90				
8	3677	289M6D406M1I51M	99				
9	NA	<na></na>	NA				
10	NA	<na></na>	NA				

# 2.3.2 xrefsByName

Or, we can look for an external reference primary accession<sup>13</sup> (given a species):

# 2.3.3 xrefsBySymbol

And we can finally look up all Ensembl objects referenced by an external symbol for a given species 14:

 $<sup>^{13} {\</sup>tt http://beta.rest.ensembl.org/documentation/info/xref\_name}$ 

<sup>14</sup>http://beta.rest.ensembl.org/documentation/info/xref\_external

## 2.4 Features

# 2.4.1 featuresByRegion

We can also look for features along a given range<sup>15</sup> (by default this will just look for genes):

```
> featuresByRegion( '7:140424943-140624564', 'human' )
 GRanges with 2 ranges and 6 metadata columns:
       seqnames
                                 ranges strand |
                                                                ID
          <Rle>
                              <IRanges> <Rle> |
                                                         <factor>
   [1]
              7 [140424943, 140624564]
                                              - | ENSG00000157764
   [2]
              7 [140583872, 140583978]
                                              + | ENSG00000207040
                logic_name feature_type external_name
                                <factor>
                   <factor>
                                               <factor>
   [1] ensembl_havana_gene
                                                   BRAF
                                    gene
   [2]
                      ncrna
                                    gene
                                                     U6
                                                                           description
                                                                               <factor>
   [1] v-raf murine sarcoma viral oncogene homolog B1 [Source:HGNC Symbol;Acc:1097]
   [2]
                                       U6 spliceosomal RNA [Source:RFAM; Acc:RF00026]
              biotype
             <factor>
   [1] protein_coding
   [2]
                snRNA
   seqlengths:
     7
    NA
And by using the feature parameter, we can specify what we're looking for
 > featuresByRegion( '7:140424943-140624564', 'human', feature='transcript' )
 GRanges with 6 ranges and 5 metadata columns:
       seqnames
                                                                TD
                                 ranges strand |
          <Rle>
                              <IRanges> <Rle> |
                                                         <factor>
              7 [140424943, 140482957]
   [1]
                                              - | ENST00000496384
   [2]
              7 [140434279, 140624564]
                                              - | ENST00000288602
   [3]
              7 [140434321, 140454011]
                                              - | ENST00000479537
   [4]
              7 [140434397, 140624458]
                                              - | ENST00000497784
   [5]
              7 [140533861, 140624509]
                                              - | ENST0000469930
              7 [140583872, 140583978]
   [6]
                                              + | ENST00000384313
                       logic_name feature_type
                                                         Parent
                         <factor>
                                      <factor>
                                                       <factor>
   [1]
                           havana
                                    transcript ENSG00000157764
   [2] ensembl_havana_transcript
                                    transcript ENSG00000157764
   [3]
                                    transcript ENSG00000157764
                           havana
   [4]
                           havana
                                    transcript ENSG00000157764
   [5]
                                    transcript ENSG00000157764
                           havana
   [6]
                                    transcript ENSG00000207040
                            ncrna
                        biotype
                       <factor>
   [1]
                protein_coding
```

protein\_coding

[2]

 $<sup>^{15} \</sup>verb|http://beta.rest.ensembl.org/documentation/info/feature\_region$ 

```
[3] nonsense_mediated_decay
[4] nonsense_mediated_decay
[5] retained_intron
[6] snRNA
---
seqlengths:
7
NA
```

You can specify multiple features (columns which don't exist for a given type of result will be filled with NA)

```
> featuresByRegion( '7:140424943-140624564', 'human', feature=c('gene','transcript') )
GRanges with 8 ranges and 7 metadata columns:
      seqnames
                                                               ID
                                ranges strand |
         <Rle>
                             <IRanges> <Rle> |
                                                        <factor>
             7 [140424943, 140624564]
  [1]
                                            - | ENSG00000157764
  [2]
             7 [140583872, 140583978]
                                             + | ENSG00000207040
  [3]
             7 [140424943, 140482957]
                                             - | ENST00000496384
  [4]
             7 [140434279, 140624564]
                                             - | ENST00000288602
  [5]
             7 [140434321, 140454011]
                                             - | ENST00000479537
             7 [140434397, 140624458]
  [6]
                                             - | ENST00000497784
             7 [140533861, 140624509]
                                             - | ENST00000469930
  [7]
             7 [140583872, 140583978]
  [8]
                                            + | ENST00000384313
                      logic_name feature_type external_name
                        <factor>
                                     <factor>
                                                    <factor>
  [1]
                                                        BRAF
            ensembl_havana_gene
                                         gene
  [2]
                           ncrna
                                                          U6
                                          gene
  [3]
                          havana
                                   transcript
                                                        <NA>
                                                        <NA>
      ensembl_havana_transcript
                                   transcript
  [5]
                          havana
                                   transcript
                                                        <NA>
  [6]
                          havana
                                   transcript
                                                        <NA>
  [7]
                          havana
                                   transcript
                                                        <NA>
  [8]
                                                        <NA>
                           ncrna
                                   transcript
                                                                          description
                                                                              <factor>
  [1] v-raf murine sarcoma viral oncogene homolog B1 [Source:HGNC Symbol;Acc:1097]
  [2]
                                      U6 spliceosomal RNA [Source:RFAM; Acc:RF00026]
  [3]
                                                                                  <NA>
  [4]
                                                                                  <NA>
  [5]
                                                                                  <NA>
  [6]
                                                                                  <NA>
  [7]
                                                                                  <NA>
  [8]
                                                                                  <NA>
                       biotype
                                        Parent
                      <factor>
                                       <factor>
  [1]
               protein_coding
                                           <NA>
  [2]
                         {\tt snRNA}
               protein_coding ENSG00000157764
  [3]
  [4]
               protein_coding ENSG00000157764
  [5] nonsense_mediated_decay ENSG00000157764
  [6] nonsense_mediated_decay ENSG00000157764
  [7]
              retained_intron ENSG00000157764
  [8]
                         snRNA ENSG00000207040
```

seqlengths:

7 NA

# 2.5 Lookup

# 2.5.1 lookupld

To find the database and species containing a known Ensembl id, you can use the lookup function  $^{16}$  like so:

 $<sup>^{16} \</sup>mathtt{http://beta.rest.ensembl.org/documentation/info/lookup}$ 

# 2.6 Mapping

The mapping functions are used to convert co-ordinates between systems or databases.

There is currently an issue with mappingCdna, mappingCds and mappingTranslation in that the seq\_region\_name is not returned from the REST interface. This has been reported to Ensembl and should be fixed in the next release.

#### 2.6.1 mapping

The mapping function<sup>17</sup> converts the co-ordinates in one assembly into another, ie:

```
> mapping( 'NCBI36', 'X:1..10000:1', 'GRCh37', 'human' )
[[1]]
GRanges with 2 ranges and 3 metadata columns:
                       ranges strand |
                                           assembly coordinate_system
     seqnames
         <Rle>
                    <IRanges> <Rle> | <character>
                                                         <character>
                    1, 10000]
                                   + |
  [1]
             X [
                                             NCBI36
                                                           chromosome
  [2]
             X [60001, 70000]
                                   + |
                                             GRCh37
                                                           chromosome
             type
      <character>
  [1]
         original
  [2]
          mapped
 seqlengths:
   X
```

As you can see, it returns one GRanges object per result, with an original row and a mapped row.

# 2.6.2 mappingCdna

This function<sup>18</sup> converts CDNA co-ordinates for a given Ensembl Transcript to genomic co-ordinates.

```
> mappingCdna( 'ENST00000288602', '100..300')
GRanges with 2 ranges and 2 metadata columns:
      seqnames
                               ranges strand |
                                                      gap
         <Rle>
                            <IRanges> <Rle> | <numeric> <numeric>
  [1]
             7 [140624366, 140624465]
                                                        0
                                                                   0
  [2]
             7 [140549912, 140550012]
                                                        0
                                                                   0
  seqlengths:
   7
  NA
```

## 2.6.3 mappingCds

Or you can convert CDS co-ordinates<sup>19</sup> instead of CDNA ones:

```
> mappingCds( 'ENST00000288602', '100..300')
```

<sup>17</sup>http://beta.rest.ensembl.org/documentation/info/assembly\_map

<sup>18</sup>http://beta.rest.ensembl.org/documentation/info/assembly\_cdna

<sup>19</sup>http://beta.rest.ensembl.org/documentation/info/assembly\_cds

```
GRanges with 3 ranges and 2 metadata columns:
                            ranges strand | gap
     seqnames
                                                         rank
        <Rle>
                         <IRanges> <Rle> | <numeric> <numeric>
  [1]
           7 [140624366, 140624404] - | 0
  [2]
           7 [140549911, 140550012]
                                       - |
                                                            0
                                                 0
 [3]
           7 [140534613, 140534672]
                                       - |
                                                            0
 seqlengths:
   7
  NA
```

## 2.6.4 mappingTranslation

And finally, it is possible to convert from protein co-ordinates to genomic ones using the mapping- $Translation\ method^{20}$ :

```
> mappingTranslation( 'ENSP00000288602', '100..300')
GRanges with 5 ranges and 2 metadata columns:
     seqnames
                            ranges strand |
                                           gap
        <Rle>
                         <IRanges> <Rle> | <numeric> <numeric>
 [1]
           7 [140534409, 140534615]
                                      - |
                                             0
                                                          0
 [2]
           7 [140508692, 140508795]
                                                           0
                                                 0
                                     - |
                                               0
                                                          0
 [3]
           7 [140507760, 140507862]
 [4]
           7 [140501212, 140501360]
                                     - |
                                                0
                                                          0
                                     - |
 [5]
          7 [140500242, 140500281]
                                                0
                                                          0
 seqlengths:
   7
  NA
```

 $<sup>^{20}</sup> http://beta.rest.ensembl.org/documentation/info/assembly\_translation$ 

# 2.7 Sequences

## 2.7.1 sequenceById

List of 4 \$ desc

Fetch a sequence based on the stable id of an Ensembl feature<sup>21</sup> (I'm using str here to avoid overflowing the pdf too much):

```
> str( sequenceById( 'ENSG00000157764' ), give.head=F, strict.width='cut' )
List of 4
$ desc : "chromosome:GRCh37:7:140424943:140624564:-1"
$ id : "ENSG00000157764"
$ seq : "CGCCTCCCTTCCCCCTCCCCGCCCGACAGCGGCCCCGGCCCCGGCTCTCGGTTATAAGATGG...
$ molecule: "dna"
```

You can also get different types of sequence, here is an example for the spliced CDNA sequence of a transcript:

```
$ desc : NULL
$ id : "ENST00000408384"
$ seq : "GGATGCCCAGCTAGTTTGAATTTTAGATAAACAACGAATAATTTCGTAGCATAAATATGTCCCAA..
$ molecule: "dna"

And again, for the protein coding
> str( sequenceById( 'ENSP00000334393', type='protein'), give.head=F, strict.width='cut')
List of 4
$ desc : NULL
$ id : "ENSP00000334393"
```

: "MVTEFIFLGLSDSQELQTFLFMLFFVFYGGIVFGNLLIVITVVSDSHLHSPMYFLLANLSLIDLS...

> str( sequenceById( 'ENST00000408384', type='cdna' ), give.head=F, strict.width='cut' )

# 2.7.2 sequenceByRegion

\$ molecule:"protein"

You can also just query for a region of a given species<sup>22</sup>:

```
> str( sequenceByRegion( 'X:1_000_000..1_000_100:1', 'human' ), give.head=F, strict.width='cut
List of 3
$ id :"chromosome:GRCh37:X:1000000:1000100:1"
$ seq :"GAAACAGCTACTTGGAAGGCTGAAGCAGGAGGATTGTTTGAGTCTAGGAGTTTGAGGCTGCAGTG..
$ molecule:"dna"
```

If you pass format='fasta' to the above method, it will just return you a character vector containing a FastA formatted sequence.

<sup>&</sup>lt;sup>21</sup>http://beta.rest.ensembl.org/documentation/info/sequence\_id

<sup>&</sup>lt;sup>22</sup>http://beta.rest.ensembl.org/documentation/info/sequence\_region

## 2.8 Variation

#### 2.8.1 variationAllele

```
> var = variationAllele( 'C', '9:22125503-22125502:1', 'human' )
                           # The response object
> var
[[1]]
hgvs
 C = 9:g.22125502_22125503insC
transcripts : 10 in total
> var[[1]] $transcripts[1:2] # Just the top 2 transcripts of the first response to save room
[[1]]
data
                    gene_id transcript_id biotype cdna_allele_string
1 CDKN2B-AS1 ENSG00000240498 ENST00000585267 antisense
 is_canonical
       FALSE
alleles
       consequence_terms
1 downstream_gene_variant
[[2]]
data
                    gene_id transcript_id biotype cdna_allele_string
1 CDKN2B-AS1 ENSG00000240498 ENST00000580576 antisense
 is_canonical
    FALSE
alleles
       consequence_terms
1 downstream_gene_variant
```

## 2.8.2 variationId

```
> var = variationId( 'COSM476', 'human' )
> var
                           # The response object
[[1]]
          : COSM476
is_somatic : TRUE
hgvs
 T = 7:g.140453136A>T
transcripts: 4 in total
> var[[1]]$transcripts[1:2] # Again, just the top 2 transcripts to save room
[[1]]
data
                :
              gene_id transcript_id
1 BRAF ENSG00000157764 ENST00000479537 nonsense_mediated_decay
  cdna_allele_string codon_position translation_stable_id translation_start
                T/A
                                2
                                        ENSP00000418033
 translation_end exon_number cdna_start cdna_end cds_start cds_end
```

```
2/6 83
              28
                                         83
                                                    83
                                                             83
 is_canonical
       FALSE
 display_codon_allele_string pep_allele_string codon_allele_string
                                         V/E
                    gTg/gAg
           hgvs_transcript
                                         hgvs_protein polyphen_score
1 ENST00000479537.1:c.83T>A ENSP00000418033.1:p.Val28Glu
                                                           0.946
 polyphen_prediction sift_score sift_prediction
1 probably damaging
                          0.12 tolerated missense_variant
      consequence_terms
1 NMD_transcript_variant
protein_features :
     name
                          db
1 PF00069
                 Pfam domain
2 PF07714
                 Pfam domain
3 SSF56112 Superfamily domains
4 PS50011 PROSITE profiles
[[2]]
data
             gene_id transcript_id biotype
1 BRAF ENSG00000157764 ENST00000288602 protein_coding CCDS5863.1
 cdna_allele_string codon_position translation_stable_id translation_start
               T/A
                               2 ENSP00000288602
 translation_end exon_number cdna_start cdna_end cds_start cds_end
                     15/18
                               1860
 is_canonical
      TRUE
alleles
 display_codon_allele_string pep_allele_string codon_allele_string
                    gTg/gAg
                                         V/E
                                                       GTG/GAG
             hgvs_transcript
                                            hgvs_protein polyphen_score
1 ENST00000288602.6:c.1799T>A ENSP00000288602.6:p.Val600Glu
 \verb"polyphen_prediction" sift_score sift_prediction consequence\_terms"
1 probably damaging
                        0.02 deleterious missense_variant
protein_features :
     name
1 PF07714
                 Pfam domain
2 PF00069
                 Pfam domain
3 SSF56112 Superfamily domains
               SMART domains
4 SM00220
5 SM00219
                SMART domains
6 PS50011
          PROSITE profiles
```