## Adam Mark, Ryan Thompson, Chunlei Wu

#### October 7, 2014

#### **Contents**

1	Overview				
2	Gene Annotation Service  2.1 getGene				
3	Gene Query Service           3.1 query	<b>3</b> 3			
4	makeTranscriptDbFromMyGene	4			
5	Tutorial, ID mapping 5.1 Mapping gene symbols to Entrez gene ids 5.2 Mapping gene symbols to Ensembl gene ids 5.3 When an input has no matching gene 5.4 When input ids are not just symbols 5.5 When an input id has multiple matching genes 5.6 Can I convert a very large list of ids?	6 7 7 8			
6	References	10			

## 1 Overview

MyGene.Info provides simple-to-use REST web services to query/retrieve gene annotation data. It's designed with simplicity and performance emphasized. *mygene* is an easy-to-use R wrapper to access MyGene.Info services.

#### 2 Gene Annotation Service

#### 2.1 getGene

• Use getGene, the wrapper for GET query of "/gene/<geneid>" service, to return the gene object for the given geneid.

```
> gene=getGene("1017", fields="all")
> length(gene)
[1] 36
> gene$name
[1] "cyclin-dependent kinase 2"
> gene$taxid
[1] 9606
> gene$uniprot
$`Swiss-Prot`
[1] "P24941"
$TrEMBL
[1] "B4DDL9" "E7ESI2" "G3V317" "G3V5T9"
> gene$refseq
$genomic
[1] "AC_000144"
                   "NC_000012"
                                                  "NG_034014"
                                   "NC_018923"
                                                                  "NT_029419"
[6] "NW_001838059" "NW_004929384"
$rna
[1] "NM_001290230" "NM_001798"
                                   "NM_052827"
$protein
[1] "NP_001277159" "NP_001789"
                                   "NP_439892"
```

#### 2.2 getGenes

• Use getGenes, the wrapper for POST query of "/gene" service, to return the list of gene objects for the given character vector of geneids.

```
> getGenes(c("1017","1018","ENSG00000148795"))
chr "symbol,name,taxid,entrezgene"
DataFrame with 3 rows and 6 columns
```

				<character< th=""><th>r&gt;</th><th><character></character></th><th><numeric></numeric></th><th><numeric></numeric></th></character<>	r>	<character></character>	<numeric></numeric>	<numeric></numeric>
1			cyclin-depe	ndent kinase	2	CDK2	9606	1017
2			cyclin-depe	ndent kinase	3	CDK3	9606	1018
3	cytochrome P450,	family 17,	subfamily A,	polypeptide	1	CYP17A1	9606	1586
	query	_id						
	<character></character>	<character></character>						
1	1017	1017						
2	1018	1018						
3	ENSG00000148795	1586						

## 3 Gene Query Service

#### 3.1 query

• Use query, a wrapper for GET query of "/query?q=<query>" service, to return the query result.

```
> query(q="cdk2", size=5)
```

```
$hits
  entrezgene
                                                         symbol
                                                                   _id taxid
                                         name
                                                 _score
1
                    cyclin-dependent kinase 2 389.77063
                                                           CDK2
        1017
                                                                  1017 9606
2
       12566
                    cyclin-dependent kinase 2 348.08942
                                                           Cdk2 12566 10090
3
                    cyclin dependent kinase 2 276.61142
      362817
                                                           Cdk2 362817 10116
4
       52004
                    CDK2-associated protein 2 20.83584 Cdk2ap2 52004 10090
5
      78832 CDK2 associated, cullin domain 1 18.44532 Cacul1
                                                                 78832 10090
```

```
$max_score
[1] 389.7706

$took
[1] 7

$total
[1] 26
> query(q="NM_013993")
```

entrezgene name \_score symbol \_id taxid 1 780 discoidin domain receptor tyrosine kinase 1 0.524516 DDR1 780 9606

\$max\_score [1] 0.524516

\$took

\$hits

[1] 6

\$total

[1] 1

#### 3.2 queryMany

• Use queryMany, a wrapper for POST query of "/query" service, to return the batch query result.

```
> queryMany(c('1053_at', '117_at', '121_at', '1255_g_at', '1294_at'),
+ scopes="reporter", species="human")
```

#### Finished

DataFrame with 5 rows and 6 columns

יב	Datarrame with 6 rows and 6 coramins						
			name	symbol	taxid	entrezgene	
		<char< td=""><td>acter&gt;</td><td><character></character></td><td><numeric></numeric></td><td><numeric></numeric></td></char<>	acter>	<character></character>	<numeric></numeric>	<numeric></numeric>	
1	replication	factor C (activator 1) 2,	40kDa	RFC2	9606	5982	
2	heat	shock 70kDa protein 6 (HS	P70B')	HSPA6	9606	3310	
3		paired	box 8	PAX8	9606	7849	
4	guanylat	ce cyclase activator 1A (re	etina)	GUCA1A	9606	2978	
5	ubiquitin-l:	ke modifier activating en	zyme 7	UBA7	9606	7318	
	query	_id					
	<character></character>	<character></character>					
1	1053_at	5982					
2	117_at	3310					
3	121_at	7849					
4	1255_g_at	2978					
5	1294_at	7318					

## 4 makeTranscriptDbFromMyGene

TranscriptDb is a container for storing transcript annotations. makeTranscriptDbFromMyGene allows the user to make a TranscriptDb object in the Genomic Features package from a mygene "exons" query.

```
> xli<-c('DDX26B',
+ 'CCDC83',
+ 'MAST3',
+ 'RPL11',
+ 'ZDHHC20',
+ 'LUC7L3',
+ 'SNORD49A',
+ 'CTSH',
+ 'ACOT8')
> txdb<-makeTranscriptDbFromMyGene(xli,</pre>
```

```
+ scopes="symbol", species="human")
> transcripts(txdb)
```

GRanges with 15 ranges and 2 metadata columns:

	seqnames		ranges	strand		${\sf tx\_id}$	<pre>tx_name</pre>
	<rle></rle>		<pre><iranges></iranges></pre>	<rle></rle>		<integer></integer>	<character></character>
[1]	1	[24018268,	24022915]	+	- 1	5	NM_001199802
[2]	1	[24018268,	24022915]	+	- 1	6	NM_000975
[3]	11	[85566143,	85631063]	+	- 1	2	NM_001286159
[4]	11	[85566143,	85631063]	+	- 1	3	NM_173556
[5]	13	[21946709,	22033508]	-	- 1	10	NM_153251
[11]	17	[ 48796925,	48830072]	+	- 1	11	NM_016424
[12]	17	[ 48796925,	48830072]	+	- 1	12	NM_006107
[13]	19	[ 18208602,	18262499]	+	- 1	4	NM_015016
[14]	20	[ 44470359,	44486048]	-	- 1	15	NM_005469
[15]	Х	[134654554,	134716460]	+	- 1	1	NM_182540

seqlengths:

X 11 19 1 13 17 15 20 NA NA NA NA NA NA NA

makeTranscriptDbFromMyGene invokes either the query or queryMany method and passes the response to construct a TranscriptDb object. See ?TranscriptDb for methods to utilize and access transcript annotations.

## 5 Tutorial, ID mapping

ID mapping is a very common, often not fun, task for every bioinformatician. Supposedly you have a list of gene symbols or reporter ids from an upstream analysis, and then your next analysis requires to use gene ids (e.g. Entrez gene ids or Ensembl gene ids). So you want to convert that list of gene symbols or reporter ids to corresponding gene ids.

Here we want to show you how to do ID mapping quickly and easily.

## 5.1 Mapping gene symbols to Entrez gene ids

Suppose xli is a list of gene symbols you want to convert to entrez gene ids:

```
> xli<-c('DDX26B',
+ 'CCDC83',
+ 'MAST3',
+ 'FLOT1',
+ 'RPL11',</pre>
```

```
+ 'ZDHHC20',
+ 'LUC7L3',
+ 'SNORD49A',
+ 'CTSH',
+ 'ACOT8')
```

You can then call queryMany method, telling it your input is symbol, and you want entrezgene (Entrez gene ids) back.

> queryMany(xli, scopes="symbol", fields="entrezgene", species="human")

#### Finished

DataFrame with 10 rows and 3 columns

	query	${\tt entrezgene}$	_id
	<character></character>	<numeric></numeric>	<character></character>
1	DDX26B	203522	203522
2	CCDC83	220047	220047
3	MAST3	23031	23031
4	FLOT1	10211	10211
5	RPL11	6135	6135
6	ZDHHC20	253832	253832
7	LUC7L3	51747	51747
8	SNORD49A	26800	26800
9	CTSH	1512	1512
10	ACOT8	10005	10005

#### 5.2 Mapping gene symbols to Ensembl gene ids

Now if you want Ensembl gene ids back:

> out <-queryMany(xli, scopes="symbol", fields="ensembl.gene", species="human")

#### Finished

> out

DataFrame with 10 rows and 3 columns

	ensembl.gene	_id	query
	<characterlist></characterlist>	<character></character>	<character></character>
1	ENSG00000165359	203522	DDX26B
2	ENSG00000150676	220047	CCDC83
3	ENSG00000099308	23031	MAST3
4	ENSG00000137312, ENSG00000206379, ENSG00000206480,	10211	FLOT1
5	ENSG00000142676	6135	RPL11
6	ENSG00000180776	253832	ZDHHC20
7	ENSG00000108848	51747	LUC7L3
8	ENSG00000277370, ENSG00000175061	26800	SNORD49A

```
9
                                         ENSG00000103811
                                                                              CTSH
                                                                 1512
10
                                         ENSG00000101473
                                                                10005
                                                                             ACOT8
```

- > out\$ensembl.gene[[4]]
- [1] "ENSG00000137312" "ENSG00000206379" "ENSG00000206480" "ENSG00000223654"
- [5] "ENSG00000224740" "ENSG00000230143" "ENSG00000232280" "ENSG00000236271"

#### 5.3 When an input has no matching gene

In case that an input id has no matching gene, you will be notified from the output. The returned list for this query term contains notfound value as True.

```
> xli<-c('DDX26B',
         'CCDC83',
+
         'MAST3',
         'FLOT1',
         'RPL11',
         'Gm10494')
> queryMany(xli, scopes="symbol", fields="entrezgene", species="human")
```

#### Finished

Pass returnall=TRUE to return lists of duplicate or missing query terms. DataFrame with 6 rows and 4 columns

	query	${\tt entrezgene}$	_id	notfound
	<character></character>	<numeric></numeric>	<character></character>	<logical></logical>
1	DDX26B	203522	203522	NA
2	CCDC83	220047	220047	NA
3	MAST3	23031	23031	NA
4	FLOT1	10211	10211	NA
5	RPL11	6135	6135	NA
6	Gm10494	NA	NA	TRUE

#### When input ids are not just symbols 5.4

```
> xli<-c('DDX26B',
          'CCDC83',
          'MAST3',
+
          'FLOT1',
          'RPL11',
          'Gm10494',
          '1007_s_at',
          'AK125780')
```

Above id list contains symbols, reporters and accession numbers, and supposedly we want to get back both Entrez gene ids and uniprot ids. Parameters scopes, fields, species are all flexible enough to support multiple values, either a list or a comma-separated string:

```
> out<-queryMany(xli, scopes=c("symbol", "reporter", "accession"),
+ fields=c("entrezgene", "uniprot"), species="human")</pre>
```

#### Finished

Pass returnall=TRUE to return lists of duplicate or missing query terms.

> out

DataFrame with 9 rows and 6 columns

	query	${\tt entrezgene}$	${\tt uniprot.Swiss-Prot}$	uniprot.TrEMBL	_id
	<character></character>	<numeric></numeric>	<character></character>	<characterlist></characterlist>	<character></character>
1	DDX26B	203522	Q5JSJ4		203522
2	CCDC83	220047	Q8IWF9	HOYDV3	220047
3	MAST3	23031	060307	V9GYV0	23031
4	FLOT1	10211	075955	A2AB09,A2AB10,A2AB11,	10211
5	RPL11	6135	P62913	Q5VVC8,Q5VVC9,Q5VVD0	6135
6	Gm10494	NA	NA		NA
7	1007_s_at	780	Q08345	A2ABLO,A2ABL2,A2ABM8,	780
8	1007_s_at	100616237	NA		100616237
9	AK125780	2978	P43080	A6PVH5	2978
	notfound				
	<logical></logical>				
1	NA				
2	NA				
3	NA				
4	NA				
5	NA				
6	TRUE				
7	NA				
8	NA				
9	NA				

> out\$`uniprot.Swiss-Prot`[[5]]

[1] "P62913"

#### 5.5 When an input id has multiple matching genes

From the previous result, you may have noticed that query term 1007\_s\_at matches two genes. In that case, you will be notified from the output, and the returned result will include both matching genes.

By passing returnall=TRUE, you will get both duplicate or missing query terms

```
> queryMany(xli, scopes=c("symbol", "reporter", "accession"),
             fields=c("entrezgene", "uniprot"), species='human', returnall=TRUE)
Finished
$response
DataFrame with 9 rows and 6 columns
        query entrezgene uniprot. Swiss-Prot
                                                          uniprot.TrEMBL
                                                                                  _id
                                  <character>
                                                         <CharacterList> <character>
  <character>
                <numeric>
1
       DDX26B
                   203522
                                       Q5JSJ4
                                                                               203522
2
       CCDC83
                   220047
                                       Q8IWF9
                                                                  HOYDV3
                                                                               220047
3
        MAST3
                                       060307
                    23031
                                                                  V9GYV0
                                                                                23031
4
        FLOT1
                    10211
                                       075955 A2AB09,A2AB10,A2AB11,...
                                                                                10211
5
        RPL11
                                                   Q5VVC8,Q5VVC9,Q5VVD0
                     6135
                                       P62913
                                                                                 6135
6
      Gm10494
                       NA
                                            NA
                                                                                   NA
7
    1007_s_at
                      780
                                       Q08345 A2ABL0, A2ABL2, A2ABM8, ...
                                                                                  780
8
    1007_s_at
                100616237
                                            NA
                                                                            100616237
9
     AK125780
                                       P43080
                     2978
                                                                  A6PVH5
                                                                                 2978
   notfound
  <logical>
1
         NA
2
         NA
3
         NA
4
         NA
5
         NA
6
       TRUE
7
         NA
8
         NA
9
         NA
$duplicates
  X1007_s_at
1
           2
$missing
```

The returned result above contains out for mapping output, missing for missing query terms (a list), and dup for query terms with multiple matches (including the number of matches).

## 5.6 Can I convert a very large list of ids?

[1] "Gm10494"

Yes, you can. If you pass an id list (i.e., xli above) larger than 1000 ids, we will do the id mapping in-batch with 1000 ids at a time, and then concatenate the results all together for you. So, from the user-end, it's exactly the same as passing a shorter list. You don't need to worry about saturating our backend servers. Large lists, however, may take a while longer to query, so please wait patiently.

# **6** References

Wu C, MacLeod I, Su AI (2013) BioGPS and MyGene.info: organizing online, gene-centric information. Nucl. Acids Res. 41(D1): D561-D565. help@mygene.info