MyVariant.info R Client

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1 Overview

MyVariant.info is a simple-to-use REST web service to query/retrieve genetic variant annotation from an aggregation of variant annotation resources. myvariant is an easy-to-use R wrapper to access MyVariant.info services and explore variant annotations.
2 Variant Annotation Service

2.1 Obtaining HGVS IDs from a VCF file.

- Use `readVcf` from the VariantAnnotation package to read a Vcf file in. The Vcf object can then be passed to `formatHgvs` to retrieve HGVS IDs. HGVS IDs are based on the GRCh38/hg19 reference genome. Support for hg38 is coming soon.

```r
> file.path <- system.file("extdata", "dbsnp_mini.vcf", package="myvariant")
> vcf <- readVcf(file.path, genome="hg19")
> rowRanges(vcf)
```

<table>
<thead>
<tr>
<th>seqnames</th>
<th>ranges</th>
<th>strand</th>
<th>paramRangeID</th>
<th>REF</th>
<th>ALT</th>
<th>QUAL</th>
<th>FILTER</th>
</tr>
</thead>
<tbody>
<tr>
<td>rs376643643</td>
<td>10019-10020</td>
<td>*</td>
<td>NA</td>
<td>TA</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>rs373328635</td>
<td>10055</td>
<td>*</td>
<td>NA</td>
<td>T</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>rs62651026</td>
<td>10108</td>
<td>*</td>
<td>NA</td>
<td>C</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>rs376007522</td>
<td>10109</td>
<td>*</td>
<td>NA</td>
<td>A</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>rs368469931</td>
<td>10139</td>
<td>*</td>
<td>NA</td>
<td>A</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>...</td>
<td>...</td>
<td>...</td>
<td>...</td>
<td>...</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>rs544020171</td>
<td>17654</td>
<td>*</td>
<td>NA</td>
<td>T</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>rs563880190</td>
<td>17694</td>
<td>*</td>
<td>NA</td>
<td>C</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>rs574335987</td>
<td>17695</td>
<td>*</td>
<td>NA</td>
<td>G</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>rs374995955</td>
<td>17697</td>
<td>*</td>
<td>NA</td>
<td>G</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>rs543363182</td>
<td>17709</td>
<td>*</td>
<td>NA</td>
<td>T</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

**seqinfo:** 1 sequence from hg19 genome; no seqlengths
You can then use `formatHgvs` to extract HGVS IDs from the Vcf object.

```r
> hgvs <- formatHgvs(vcf, variant_type="snp")
> head(hgvs)
[1] "1:g.10108C>T" "1:g.10109A>T" "1:g.10139A>T" "1:g.10150C>T" "1:g.10177A>C"
[6] "1:g.10180T>C"
```

2.2 `getVariant`

Use `getVariant`, the wrapper for GET query of "/v1/variant/<hgvsid>" service, to return the variant object for the given HGVS id.

```r
> variant <- getVariant("chr1:g.35367G>A")
> variant[[1]]$dbnsfp$genename
NULL
> variant[[1]]$cadd$phred
[1] 3.726
```

2.3 `getVariants`

Use `getVariants`, the wrapper for POST query of "/v1/variant" service, to return the list of variant objects for the given character vector of HGVS ids.

```r
> getVariants(c("chr1:g.35367G>A", "chr16:g.28883241A>G"),
+   fields="cadd.consequence")
```

<table>
<thead>
<tr>
<th>query</th>
<th>X_id</th>
<th>cadd_license</th>
</tr>
</thead>
<tbody>
<tr>
<td>cadd.consequence</td>
<td>&lt;character&gt;</td>
<td>&lt;character&gt;</td>
</tr>
<tr>
<td>NONCODING_CHANGE</td>
<td>1</td>
<td></td>
</tr>
<tr>
<td>NON_SYNONYMOUS</td>
<td>2</td>
<td></td>
</tr>
</tbody>
</table>
3 Variant Query Service

3.1 `queryVariant`

- `queryVariant` is a wrapper for GET query of "/v1/query?q=<query>" service, to return the query result. This function accepts wild card input terms and allows you to query for variants that contain a specific annotation. For example, the following query searches for the CADD phred score and consequence for all variants whose genename (dbNSFP) is MLL2.

```r
> queryVariant(q="dbnsfp.genename:MLL2", fields=c("cadd.phred", "cadd.consequence"))
```

```r
took
[1] 15

total
[1] 0

max_score
NULL

hits
list()
```

- You can also use `queryVariant` to retrieve all annotations that map to a specific rsID.

```r
> queryVariant(q="rs58991260", fields="dbsnp.flags")$hits
```

```r
     _id   _score
1 chr1:g.218631822G>A 21.25038
```

3.2 `queryVariants`

- `queryVariants` is a wrapper for POST query of "/v1/query?q=<query>" service, to return the query result. Query terms include any available field as long as scopes are defined. The following example reads the dbSNP rsIDs from a VCF and queries for all fields. The returned DataFrame can then be easily subsetted to include, for example, those that have not been documented in the Wellderly study.

```r
> rsids <- paste("rs", info(vcf)$RS, sep="")
> res <- queryVariants(q=rsids, scopes="dbsnp.rsid", fields="all")
```
Finished

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

```r
> subset(res, !is.na(wellderly.vartype))$query
```

```
[1] "rs62651026" "rs376007522" "rs368469931" "rs368469931" "rs371194064"
[6] "rs371194064" "rs201752861" "rs201752861" "rs201694901" "rs201694901"
[11] "rs201694901" "rs200279319" "rs200279319" "rs145599635" "rs148908337"
[16] "rs148908337" "rs199706086" "rs111200574" "rs111200574" "rs112155239"
[21] "rs112155239" "rs528916756" "rs565971701" "rs55998931" "rs199606420"
[26] "rs62636508" "rs537182016" "rs58108140" "rs189107123" "rs10218527"
[31] "rs540538026" "rs62635286" "rs62635286" "rs531730856" "rs548333521"
[36] "rs538791886" "rs527952245" "rs558318514" "rs574697788" "rs199896944"
[41] "rs554008981" "rs113004249" "rs546169444" "rs28503599" "rs62635297"
[46] "rs62635297" "rs201055865" "rs201327123" "rs62635298" "rs571121669"
[51] "rs533499096" "rs199856693" "rs201855936" "rs71252251" "rs71252251"
[56] "rs201045431" "rs368345873" "rs576044687" "rs201635489" "rs533630043"
[61] "rs533630043" "rs564003018" "rs374029747" "rs2691315" "rs112448831"
[66] "rs372319358" "rs541172944" "rs529651976" "rs548165136" "rs11489794"
[71] "rs113141985" "rs148220436" "rs373516660" "rs150723783" "rs62636367"
[76] "rs62636367" "rs201330479" "rs62636368" "rs201563295" "rs199745162"
[81] "rs200658479" "rs200658479" "rs201833382" "rs199740902" "rs9651250"
[86] "rs9651250" "rs200978805" "rs555297131" "rs369606208" "rs201535981"
[91] "rs200784459" "rs111588939" "rs372841554" "rs200503540" "rs192890528"
[96] "rs201578576" "rs374545136" "rs377698370" "rs201072270" "rs544020171"
[101] "rs563880190" "rs574335987" "rs543363182"
```

4 References

MyVariant.info help@myvariant.info