

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.

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

GRanges object with 240 ranges and 5 metadata columns:
      seqnames      ranges strand | paramRangeID      REF
      <Rle>        <IRanges> <Rle> | <factor> <DNAStringSet>
rs376643643      1 [10019, 10020]   * | <NA>      TA
rs373328635      1 [10055, 10055]   * | <NA>      T
rs62651026       1 [10108, 10108]   * | <NA>      C
rs376007522      1 [10109, 10109]   * | <NA>      A
rs368469931      1 [10139, 10139]   * | <NA>      A
...             ...             ...   ...   ...
rs544020171      1 [17654, 17654]   * | <NA>      T
rs563880190      1 [17694, 17694]   * | <NA>      C
rs574335987      1 [17695, 17695]   * | <NA>      G
rs374995955      1 [17697, 17697]   * | <NA>      G
rs543363182      1 [17709, 17709]   * | <NA>      T
      ALT      QUAL      FILTER
      <DNAStringSetList> <numeric> <character>
rs376643643      T      <NA>      .
rs373328635      TA     <NA>      .
rs62651026       T      <NA>      .
rs376007522      T      <NA>      .
rs368469931      T      <NA>      .
...             ...             ...
rs544020171      C      <NA>      .
rs563880190      T      <NA>      .
rs574335987      A      <NA>      .
rs374995955      C      <NA>      .
rs543363182      G      <NA>      .
-----
seqinfo: 1 sequence from hg19 genome; no seqlengths
```

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- You can then use `formatHgvs` to extract HGVS IDs from the Vcf object.

```
> hgvs <- formatHgvs(vcf, variant_type="snp")
> head(hgvs)

[1] "chr1:g.10108C>T" "chr1:g.10109A>T" "chr1:g.10139A>T" "chr1:g.10150C>T"
[5] "chr1:g.10177A>C" "chr1: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.

```
> 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.

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

DataFrame with 2 rows and 4 columns
      query                X_id          cadd._license cadd.consequence
  <character>          <character>          <character>          <character>
1 chr1:g.35367G>A    chr1:g.35367G>A http://goo.gl/bkpNhq NONCODING_CHANGE
2 chr16:g.28883241A>G chr16:g.28883241A>G http://goo.gl/bkpNhq  NON_SYNONYMOUS
```

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 gene name (dbNSFP) is MLL2.

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

$max_score
NULL

$took
[1] 2

$total
[1] 0

$hits
list()
```

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

```
> queryVariant(q="rs58991260", fields="dbSNP.flags")$hits
      _id  _score  dbSNP._license
1 chr1:g.218631822G>A 10.36088 https://goo.gl/Ztr5r1
      dbSNP.flags
1 ASP, G5, GN0, KGPhase1, KGPhase3, SLO
```

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

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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 Welllderly study.

```
> 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.

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

 [1] "rs367896724" "rs140194106" "rs145427775" "rs147093981" "rs56289060"
 [6] "rs112766696" "rs55998931" "rs199606420" "rs58108140" "rs62635284"
[11] "rs62635286" "rs531730856" "rs527952245" "rs201747181" "rs546169444"
[16] "rs201055865" "rs62635298" "rs199856693" "rs201855936" "rs71252251"
[21] "rs201045431" "rs201635489" "rs533630043" "rs2691315" "rs572465511"
[26] "rs372319358" "rs11489794" "rs113141985" "rs148220436" "rs150723783"
[31] "rs62636367" "rs62636368" "rs199745162" "rs200658479" "rs201833382"
[36] "rs199740902" "rs200978805" "rs201535981" "rs192890528"
```

4 References

MyVariant.info help@myvariant.info