

# **GeneCruiser 3.2**

## **Web Service Specification**

<http://genecruiser.broad.mit.edu/>

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# 1 Changes from previous versions

If you are a new user of GeneCruiser Web Services, you can skip this section. For old users, this section describes changes that might affect your usage of GeneCruiser Web Services.

## 1.1 Changes from version 3.1

### 1.1.1 Added new service class: General

General-services can be used to fetch information about GeneCruiser properties such as version, result size limits, data source version information and available id types. See corresponding section in this documentation for further information.

## 1.2 Changes from version 3.0.x

### 1.2.1 Updated from Ensembl version 43 to version 47

Update causes many changes to the actual data content of the GeneCruiser service, and also introduces new genomic identifier types.

### 1.2.2 HUGO genomic identifier type is replaced by HGNC\_GENE

You should replace your idType parameters from HUGO to HGNC\_GENE. We have created a patch that automatically converts HUGO queries to HGNC\_GENE, but it is unlikely to be supported in the future. The patch does not alter the returned response from the service, so if you do a query using the deprecated HUGO idType, your response will contain HGNC\_GENE IdType element, possibly causing problems with your parser e.g.:

```
<vgc:GenomicId>
  <vgc:IdType>HGNC_GENE</vgc:IdType>
  <vgc:Accession>1100</vgc:Accession>
  <vgc:DisplayName>BRCA1</vgc:DisplayName>
</vgc:GenomicId>
```

### 1.2.3 Added new Gene-services: byLocation and byLocationOutputIdTypes

Added new services that can be used to retrieve information about genes based on their genomic locations. See corresponding section in this documentation for further information.

## 2 Login/Registration

The web services require that the client specify an e-mail address that has already been registered using the web-application. The registration page can be found at:

*<http://genecruiser.broad.mit.edu/genecruiser3/pages/register.jsf>*

In this document, the example e-mail address `joe@example.com` is used. Replace it with your registered e-mail.

### 3 Quick reference

This quick reference provides examples for common use cases for GeneCruiser Web Services. To use GeneCruiser Web Services, 1) point your application/browser to the GeneCruiser Web Services URL, including parameters of your query, and 2) parse the returned XML response and use it to your needs.

More detailed information about different query parameters and how to retrieve large result sets can be found from the rest of this documentation.

#### How to retrieve information about GeneCruiser properties?

Point your application to the following URL (replace *joe@example.com* with your registered GeneCruiser e-mail):

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/general/properties?email=joe@example.com](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/general/properties?email=joe@example.com)*

#### How to retrieve information about a gene?

Point your application to the following URL (replace BRCA1 with desired HGNC Gene Symbol and *joe@example.com* with your registered GeneCruiser e-mail):

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/gene/byGenomicId?idType=HGNC\\_GENE&id=BRCA1&firstResult=0&email=joe@example.com](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/gene/byGenomicId?idType=HGNC_GENE&id=BRCA1&firstResult=0&email=joe@example.com)*

#### How to retrieve information about a SNP?

Point your application to the following URL (replace rs2856753 with desired SNP rs-number and *joe@example.com* with your registered GeneCruiser e-mail):

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/variation/byName?name=rs2856753&firstResult=0&email=joe@example.com](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/variation/byName?name=rs2856753&firstResult=0&email=joe@example.com)*

#### How to retrieve information about SNPs located in a gene?

Point your application to the following URL (replace BRCA1 with desired HGNC Gene Symbol and *joe@example.com* with your registered GeneCruiser e-mail):

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/variation/byGenomicId?idType=HGNC\\_GENE&id=BRCA1&firstResult=0&email=joe@example.com](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/variation/byGenomicId?idType=HGNC_GENE&id=BRCA1&firstResult=0&email=joe@example.com)*

## 4 General

General service can be used to retrieve information about GeneCruiser properties.

### 4.1 General properties

#### 4.1.1 General properties

This web service returns information about GeneCruiser properties, such as version, result size limits data source version information and available id types.

URL:

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/general/properties?email=<email\\_address>](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/general/properties?email=<email_address>)*

Where:

- <email\_address> is the email address of a registered GeneCruiser user.

Returned properties are key/value pairs of:

- **Version:** GeneCruiser version
- **BuildNumber:** GeneCruiser build number
- **BuildTimestamp:** Date and time when this build was created
- **EnsemblSchemaVersion:** Ensembl schema version in use
- **AssemblyName:** Genome assembly name
- **AssemblyDate:** Date of genome assembly
- **GeneBuildVersion:** Version of Ensembl genebuild
- **dnSNPVersion:** Version of dbSNP from Ensembl
- Maximum resultset sizes for various services:
  - **GeneServiceByIdMaxResults**
  - **VariationServiceByIdMaxResults**
  - **VariationServiceByLocationMaxResults**
  - **GeneServiceByLocationMaxResults**
  - **GeneServiceByVariationIdMaxResults:**

#### 4.1.1.1 Examples

Retrieving information about GeneCruiser properties:

[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/general/properties?email=joe@example.com](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/general/properties?email=joe@example.com)

This returns GeneCruiser properties as key/value pairs:

```
<ns:propertiesResponse">
  <ns:return>
    <vgc:Properties">
      <vgc:Property>
        <vgc:Key>Version</vgc:Key>
        <vgc:Value>3.2</vgc:Value>
      </vgc:Property>
      <vgc:Property>
        <vgc:Key>BuildNumber</vgc:Key>
        <vgc:Value>459</vgc:Value>
      </vgc:Property>
      <vgc:Property>
        <vgc:Key>BuildTimestamp</vgc:Key>
        <vgc:Value>08/07/2008 11:36 AM</vgc:Value>
      </vgc:Property>
      <vgc:Property>
        <vgc:Key>EnsemblSchemaVersion</vgc:Key>
        <vgc:Value>47</vgc:Value></vgc:Property>
      <vgc:Property>
        <vgc:Key>AssemblyName</vgc:Key>
        <vgc:Value>NCBI_36</vgc:Value>
      </vgc:Property>
      <vgc:Property>
        <vgc:Key>AssemblyDate</vgc:Key>
        <vgc:Value>2005-10</vgc:Value>
      </vgc:Property>
      <vgc:Property>
        <vgc:Key>GenebuildVersion</vgc:Key>
        <vgc:Value>2006-12-Ensembl</vgc:Value>
      </vgc:Property>
      <vgc:Property>
        <vgc:Key>dbSNPVersion</vgc:Key>
        <vgc:Value>127</vgc:Value>
      </vgc:Property>
      <vgc:Property>
        <vgc:Key>GeneServiceByIdMaxResults</vgc:Key>
        <vgc:Value>200</vgc:Value>
      </vgc:Property>
      <vgc:Property>
        <vgc:Key>VariationServiceByIdMaxResults</vgc:Key>
        <vgc:Value>200</vgc:Value>
      </vgc:Property>
      <vgc:Property>
        <vgc:Key>VariationServiceByLocationMaxResults</vgc:Key>
        <vgc:Value>200</vgc:Value>
      </vgc:Property>
      <vgc:Property>
        <vgc:Key>GeneServiceByLocationMaxResults</vgc:Key>
```

```

        <vgc:Value>200</vgc:Value>
      </vgc:Property>
    </vgc:Property>
    <vgc:Key>GeneServiceByVariationIdMaxResults</vgc:Key>
    <vgc:Value>200</vgc:Value>
  </vgc:Property>
</vgc:Properties>
</ns:return>
</ns:propertiesResponse>

```

## 4.2 General idTypes

### 4.2.1 General idTypes

This web service returns list of available genomic identifier types that can be used in other GeneCruiser services.

URL:

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/general/idTypes?email=<email\\_address>](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/general/idTypes?email=<email_address>)*

Where:

- <email\_address> is the email address of a registered GeneCruiser user.

#### 4.2.1.1 Examples

Retrieving information about available identifier types:

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/general/idTypes?email=joe@example.com](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/general/idTypes?email=joe@example.com)*

This returns information about available identifier types. Note that only part of the available identifier types are shown for brevity:

```

<ns:idTypesResponse>
  <ns:return>
    <vgc:Identifiers>
      <vgc:Identifier>
        <vgc:UrlName>HGNC_GENE</vgc:UrlName>
        <vgc:DisplayName>HGNC          Gene          Symbol
        (HGNC)</vgc:DisplayName>
        <vgc:DbName>HGNC</vgc:DbName>
        <vgc:Type>GENE</vgc:Type>

```

```

</vgc:Identifier>
<vgc:Identifier>
  <vgc:UrlName>HGNC_TRANSCRIPT</vgc:UrlName>
  <vgc:DisplayName>HGNC      Transcript      Symbol
  (HGNC)</vgc:DisplayName>
  <vgc:DbName>HGNC</vgc:DbName>
  <vgc:Type>TRANSCRIPT</vgc:Type>
</vgc:Identifier>
<vgc:Identifier>
  <vgc:UrlName>ensembl_gene_stable_id</vgc:UrlName>
  <vgc:DisplayName>Ensembl      Gene      Stable
  Id</vgc:DisplayName>
  <vgc:DbName>ensembl_gene_stable_id</vgc:DbName>
  <vgc:Type>GENE</vgc:Type>
</vgc:Identifier>
</vgc:Identifiers>
</ns:return>
</ns:propertiesResponse>

```

## 4.2.2 General outputIdTypes

This web service returns list of available genomic identifier output types that can be used in other GeneCruiser services. Note: The outputIdTypes contain effectively the same identifiers as idTypes, except that Ensembl Stable Identifiers for gene, transcript and translation are excluded. This is because when e.g., retrieving a gene, the Ensembl stable id for that gene will always be automatically returned.

URL:

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/general/outputIdTypes?email=<email\\_address>](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/general/outputIdTypes?email=<email_address>)*

Where:

- <email\_address> is the email address of a registered GeneCruiser user.

### 4.2.2.1 Examples

Retrieving information about available identifier types:

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/general/outputIdTypes?email=joe@example.com](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/general/outputIdTypes?email=joe@example.com)*

This returns information about available output identifier types. Note that only part of the available identifier types are shown for brevity:

```
<ns:idTypesResponse>
  <ns:return>
    <vgc:Identifiers>
      <vgc:Identifier>
        <vgc:UrlName>HGNC_GENE</vgc:UrlName>
        <vgc:DisplayName>HGNC          Gene          Symbol
        (HGNC)</vgc:DisplayName>
        <vgc:DbName>HGNC</vgc:DbName>
        <vgc:Type>GENE</vgc:Type>
      </vgc:Identifier>
      <vgc:Identifier>
        <vgc:UrlName>HGNC_TRANSCRIPT</vgc:UrlName>
        <vgc:DisplayName>HGNC          Transcript      Symbol
        (HGNC)</vgc:DisplayName>
        <vgc:DbName>HGNC</vgc:DbName>
        <vgc:Type>TRANSCRIPT</vgc:Type>
      </vgc:Identifier>
    </vgc:Identifiers>
  </ns:return>
</ns:propertiesResponse>
```

## 5 Genes

There are three ways to retrieve genes: by identifier, genomic location or by Variation identifier. These methods are described below in more detail.

### 5.1 Genes by GenomicId

#### 5.1.1 Genes by GenomicId

This web service returns genes with the given identifiers. The id can be specified by accession number or by display name. The identifiers can be gene, transcript, or translation identifiers.

URL:

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/gene/byGenomicId?idType=<id\\_type>&id=<id\\_list>&firstResult=<first\\_result>&email=<email\\_address>](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/gene/byGenomicId?idType=<id_type>&id=<id_list>&firstResult=<first_result>&email=<email_address>)*

Where:

- <id\_type> is the type of genomic identifier. The possible values for id\_type can be retrieved using General/idTypes service.
- <id\_list> is a list of identifiers. Identifiers are separated by “+” symbols. For example: ENSG00000141510+ENSG00000067646
- <first\_result> indicates the index of the first row to be returned. The web service will only return a set number of rows and the client is responsible for requesting additional results. The first row number starts with 0.
- <email\_address> is the email address of a registered GeneCruiser user.

#### 5.1.1.1 Examples

Searching for a single id, using the HGNC Gene Symbol as identifier:

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/gene/byGenomicId?idType=HGNC\\_GENE&id=brca1&firstResult=0&email=joe@example.com](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/gene/byGenomicId?idType=HGNC_GENE&id=brca1&firstResult=0&email=joe@example.com)*

This returns a single gene:

```
<ns:byGenomicIdResponse>
  <ns:return>
    <vgc:GeneByGenomicIdResults>
      <vgc:GeneByGenomicIdResult>
        <vgc:GeneByGenomicIdParameter>
          <vgc:Accession>1100</vgc:Accession>
          <vgc:DisplayName>BRCA1</vgc:DisplayName>
        </vgc:GeneByGenomicIdParameter>
      </vgc:GeneByGenomicIdResult>
    </vgc:GeneByGenomicIdResults>
  </ns:return>
</ns:byGenomicIdResponse>
```

```

        <vgc:GeneId>56352</vgc:GeneId>
        <vgc:Description>
        Breast cancer type 1 susceptibility protein (RING finger protein
        53). [Source:Uniprot/SWISSPROT;Acc:P38398]
        </vgc:Description>
        <vgc:Source>ensembl</vgc:Source>
        <vgc:Biotype>protein_coding</vgc:Biotype>
        <vgc:Chromosome>17</vgc:Chromosome>
        <vgc:Start>38449840</vgc:Start>
        <vgc:End>38530994</vgc:End>
        <vgc:Strand>-1</vgc:Strand>
        <vgc:StableId>ENSG00000012048</vgc:StableId>
    </vgc:Gene>
  </vgc:GeneByGenomicIdResult>
</vgc:GeneByGenomicIdResults>
</ns:return>
</ns:byGenomicIdResponse>

```

### 5.1.2 Genes byGenomicIdOutputIdTypes

This web service returns genes with the given identifiers. The id can be specified by accession number or by display name. This service also allows the caller to specify which external database accessions should be displayed. Note that retrieving the external accessions is costly, so unneeded accessions should not be retrieved for best performance.

URL:

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/gene/byGenomicIdOutputIdTypes?idType=<id\\_type>&id=<id\\_list>&outputIdTypes=<output\\_id\\_types>&firstResult=<first\\_result>&email=<email\\_address>](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/gene/byGenomicIdOutputIdTypes?idType=<id_type>&id=<id_list>&outputIdTypes=<output_id_types>&firstResult=<first_result>&email=<email_address>)*

Where:

- <id\_type> is the type of genomic identifier. The possible values for id\_type can be retrieved using General/idTypes service.
- <id\_list> is a list of identifiers. Identifiers are separated by “+” symbols. For example: ENSG00000141510+ENSG00000067646
- <output\_id\_types> is a list of output identifier types. The possible values for output\_id\_type can be retrieved using General/outputIdTypes service.
- <first\_result> indicates the index of the first row to be returned. The web service will only return a set number of rows and the client is responsible for requesting additional results. The first row number starts with 0.
- <email\_address> is the email address of a registered GeneCruiser user.

#### 5.1.2.1 Examples:

Searching for a single id, using the HGNC Gene Symbol as identifier, and specifying that the output should include EntrezGene, UniGene, and EMBL identifiers:

*http://genecruiser.broad.mit.edu/genecruiser3\_services/rest/gene/byGenomicIdOutputIdTypes?idType=HGNC\_GENE&id=brca1&outputIdTypes=EntrezGene+UniGene+EMBL&firstResult=0&email=joe@example.com*

This returns a single gene. Note that some of the returned EMBL ids are not shown for brevity:

```
<ns:byGenomicIdOutputIdTypesResponse>
  <ns:return>
    <vgc:GeneByGenomicIdResults>
      <vgc:GeneByGenomicIdResult>
        <vgc:GeneByIdParameter>
          <vgc:Accession>1100</vgc:Accession>
          <vgc:DisplayName>BRCA1</vgc:DisplayName>
        </vgc:GeneByIdParameter>
        <vgc:Gene>
          <vgc:GeneId>56352</vgc:GeneId>
          <vgc:Description>
            Breast cancer type 1 susceptibility protein
            (RING finger protein 53).
            [Source:Uniprot/SWISSPROT;Acc:P38398]
          </vgc:Description>
          <vgc:Source>ensembl</vgc:Source>
          <vgc:Biotype>protein_coding</vgc:Biotype>
          <vgc:Chromosome>17</vgc:Chromosome>
          <vgc:Start>38449840</vgc:Start>
          <vgc:End>38530994</vgc:End>
          <vgc:Strand>-1</vgc:Strand>
          <vgc:StableId>ENSG00000012048</vgc:StableId>
          <vgc:GenomicIds>
            <vgc:GenomicId>
              <vgc:IdType>EMBL</vgc:IdType>
              <vgc:Accession>DQ299317</vgc:Accession>
            </vgc:GenomicId>
            <vgc:GenomicId>
              <vgc:IdType>EMBL</vgc:IdType>
              <vgc:Accession>DQ145825</vgc:Accession>
            </vgc:GenomicId>
            <vgc:GenomicId>
              <vgc:IdType>EMBL</vgc:IdType>
              <vgc:Accession>DQ299325</vgc:Accession>
            </vgc:GenomicId>
            ...
          </vgc:GenomicIds>
          <vgc:GenomicId>
            <vgc:IdType>UniGene</vgc:IdType>
            <vgc:Accession>Hs.194143</vgc:Accession>
          </vgc:GenomicId>
        </vgc:Gene>
      </vgc:GeneByGenomicIdResult>
    </vgc:GeneByGenomicIdResults>
  </ns:return>
</ns:byGenomicIdOutputIdTypesResponse>
```

```

        <vgc:DisplayName>Hs.194143</vgc:Dis
        playName>
    </vgc:GenomicId>
    vgc:GenomicId>
        <vgc:IdType>UniGene</vgc:IdType>
        <vgc:Accession>Hs.622171</vgc:Acces
        sion>
        <vgc:DisplayName>Hs.622171</vgc:Dis
        playName>
    </vgc:GenomicId>
    vgc:GenomicId>
        <vgc:IdType>EntrezGene</vgc:IdType>
        <vgc:Accession>672</vgc:Accession>
        <vgc:DisplayName>BRCA1</vgc:Display
        Name>
    </vgc:GenomicId>
    </vgc:GenomicIds>
    </vgc:Gene>
    </vgc:GeneByIdResult>
    </vgc:GeneByIdResults>
    </ns:return>
</ns:byIdOutputIdTypesResponse>

```

## 5.2 Genes byLocation

This web service returns the genes within a range of genomic locations. The genomic location range can be specified in a number of formats. Notice that the service will also return genes that are partially located in the defined locations (i.e. if your location only contains part of the gene).

URL:

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/gene/byLocation?locations=<genomic\\_location\\_list>&<first\\_result>=0&email=<email\\_address>](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/gene/byLocation?locations=<genomic_location_list>&<first_result>=0&email=<email_address>)*

Where:

- <genomic\_location\_list> is a list of genomic locations. Locations are separated by “+” symbols. For example: 6:1234-5678+7:1234-5678. See Appendix for more details.
- <first\_result> indicates the index of the first row to be returned. The web service will only return a set number of rows and the client is responsible for requesting additional results. The first row number starts with 0.
- <email\_address> is the email address of a registered GeneCruiser user.

### 5.2.1 Examples

Searching for genes in the location 17:7512445 and range 2:196772222-196773000.

[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/gene/byLocation?locations=17:7512445+2:196772222-196773000&firstResult=0&email=joe@example.com](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/gene/byLocation?locations=17:7512445+2:196772222-196773000&firstResult=0&email=joe@example.com)

This returns two genes located in the provided genomic locations.

```
<ns:byLocationResponse xmlns:ns="http://service.genecruiser.org/xsd">
  <ns:return>
    <vgc:Genes xmlns:vgc="service.genecruiser.org">
      <vgc:Gene>
        <vgc:GeneId>11294</vgc:GeneId>
        <vgc:Description>Cellular tumor antigen p53 (Tumor suppressor p53) (Phosphoprotein p53) (Antigen NY-CO-13). [Source:Uniprot/SWISSPROT;Acc:P04637]</vgc:Description>
        <vgc:Source>ensembl</vgc:Source>
        <vgc:Biotype>protein_coding</vgc:Biotype>
        <vgc:Chromosome>17</vgc:Chromosome>
        <vgc:Start>7512445</vgc:Start>
        <vgc:End>7531642</vgc:End>
        <vgc:Strand>-1</vgc:Strand>
        <vgc:StableId>ENSG00000141510</vgc:StableId>
      </vgc:Gene>
      <vgc:Gene>
        <vgc:GeneId>23010</vgc:GeneId>
        <vgc:Description>E3 ubiquitin-protein ligase HECW2 (EC 6.3.2.-) (HECT, C2 and WW domain-containing protein 2) (NEDD4-like ubiquitin-protein ligase 2). [Source:Uniprot/SWISSPROT;Acc:Q9P2P5]</vgc:Description>
        <vgc:Source>ensembl</vgc:Source>
        <vgc:Biotype>protein_coding</vgc:Biotype>
        <vgc:Chromosome>2</vgc:Chromosome>
        <vgc:Start>196772222</vgc:Start>
        <vgc:End>197165580</vgc:End>
        <vgc:Strand>-1</vgc:Strand>
        <vgc:StableId>ENSG00000138411</vgc:StableId>
      </vgc:Gene>
    </vgc:Genes>
  </ns:return>
</ns:byLocationResponse>
```

### 5.3 Genes byLocationOutputIdTypes

This web service returns the genes with the given identifiers within a range of genomic locations. The genomic location range can be specified in a number of formats. Notice that the service will also return genes that are partially located in the defined locations (i.e. if your location only contains part of the gene). This service also allows the caller to specify which external database accessions should be displayed. Note that retrieving the external accessions is costly, so unneeded accessions should not be retrieved for best performance.

URL:

*http://genecruiser.broad.mit.edu/genecruiser3\_services/rest/gene/byLocation?locations=<genomic\_location\_list>&outputIdTypes=<output\_id\_types>&first\_result=0&email=<email\_address>*

Where:

- <genomic\_location\_list> is a list of genomic locations. Locations are separated by “+” symbols. For example: 6:1234-5678+7:1234-5678. See Appendix for more details.
- <output\_id\_types> is a list of output identifier types. The possible values for output\_id\_type can be retrieved using General/outputIdTypes service.
- <first\_result> indicates the index of the first row to be returned. The web service will only return a set number of rows and the client is responsible for requesting additional results. The first row number starts with 0.
- <email\_address> is the email address of a registered GeneCruiser user.

### 5.3.1 Examples

Searching for genes in the location 17:7512445 and range 2:196772222-196773000.

*http://genecruiser.broad.mit.edu/genecruiser3\_services/rest/gene/byLocationOutputIdTypes?locations=17:7512445&outputIdTypes=HGNC\_GENE&firstResult=0&email=joe@example.com*

This returns a single gene with HGNC\_GENE identifier:

```
<ns:byLocationOutputIdTypesResponse>
  <ns:return>
    <vgc:Genes xmlns:vgc="service.genecruiser.org">
      <vgc:Gene>
        <vgc:GeneId>11294</vgc:GeneId>
        <vgc:Description>Cellular tumor antigen p53 (Tumor suppressor p53) (Phosphoprotein p53) (Antigen NY-CO-13). [Source:Uniprot/SWISSPROT;Acc:P04637]</vgc:Description>
        <vgc:Source>ensembl</vgc:Source>
        <vgc:Biotype>protein_coding</vgc:Biotype>
        <vgc:Chromosome>17</vgc:Chromosome>
        <vgc:Start>7512445</vgc:Start>
        <vgc:End>7531642</vgc:End>
        <vgc:Strand>-1</vgc:Strand>
        <vgc:StableId>ENSG00000141510</vgc:StableId>
        <vgc:GenomicIds>
          <vgc:GenomicId>
```

```

        <vgc:IdType>HGNC_GENE</vgc:IdType>
        <vgc:Accession>11998</vgc:Accession>
        <vgc:DisplayName>TP53</vgc:DisplayName>
      </vgc:GenomicId>
    </vgc:GenomicIds>
  </vgc:Gene>
</vgc:Genes>
</ns:return>
</ns:byLocationOutputIdTypesResponse>

```

## 5.4 Genes byVariationId

There are two services for retrieving genes: byVariationID – a simple version (byVariationId) and an extended version that also retrieves accession numbers from external databases (byVariationIdOutputIdTypes).

These services return genes for which the variations identified by the input variation ids are in the gene.

These services are described below in more detail.

### 5.4.1 Searching in the Gene

#### 5.4.1.1 Genes byVariationId

This web service returns genes for which the variations specified by the given variation identifiers are within the gene. The variation identifier is the “rs number” of the variation.

URL:

*http://genecruiser.broad.mit.edu/genecruiser3\_services/rest/gene/byVariationId?id=<id\_list>&firstResult=<first\_result>&email=<email\_address>*

Where:

- <id\_list> is a list of variation identifiers. Identifiers are separated by “+” symbols. For example: rs858528+rs2856753
- <first\_result> indicates the index of the first row to be returned. The web service will only return a set number of rows and the client is responsible for requesting additional results. The first row number starts with 0.
- <email\_address> is the email address of a registered GeneCruiser user.

#### **Examples**

Searching for a gene with the single variation identifier:

*http://genecruiser.broad.mit.edu/genecruiser3\_services/rest/gene/byVariationId?id=rs858528&firstResult=0&email=joe@example.com*

This returns a single gene:

```

<ns:byVariationIdResponse>
  <ns:return>
    <vgc:GeneByVariationIdResults>
      <vgc:GeneByVariationIdResult>
        <vgc:QueryParameter>
          <vgc:VariationName>rs858528</vgc:VariationName>
        </vgc:QueryParameter>
        <vgc:Gene>
          <vgc:GeneId>67394</vgc:GeneId>
          <vgc:Description>
            Cellular tumor antigen p53 (Tumor suppressor
            p53) (Phosphoprotein p53) (Antigen NY-CO-13).
            [Source:Uniprot/SWISSPROT;Acc:P04637]
          </vgc:Description>
          <vgc:Source>ensembl</vgc:Source>
          <vgc:Biotype>protein_coding</vgc:Biotype>
          <vgc:Chromosome>17</vgc:Chromosome>
          <vgc:Start>7512464</vgc:Start>
          <vgc:End>7531642</vgc:End>
          <vgc:Strand>-1</vgc:Strand>
          <vgc:StableId>ENSG00000141510</vgc:StableId>
        </vgc:Gene>
      </vgc:GeneByVariationIdResult>
    </vgc:GeneByVariationIdResults>
  </ns:return>
</ns:byVariationIdResponse>

```

### 5.4.1.2 Genes byVariationIdOutputIdTypes

This web service returns genes for which the variations specified by the given variation identifiers are within the gene. The The identifier is the variation “rs number.”

This service also allows the caller to specify which external database accessions should be displayed. Note that retrieving the external accessions is costly, so unneeded accessions should not be retrieved for best performance.

URL:

*http://genecruiser.broad.mit.edu/genecruiser3\_services/rest/gene/byVariationIdOutputIdTypes?id=<id\_list>&outputIdTypes=<output\_id\_types>&firstResult=<first\_result>&email=<email\_address>*

Where:

- <id\_list> is a list of identifiers. Identifiers are separated by “+” symbols. For example: rs858528+rs2856753
- <output\_id\_types> is a list of output identifier types. The possible values for output\_id\_type can be retrieved using General/outputIdTypes service.
- <first\_result> indicates the index of the first row to be returned. The web service will only return a set number of rows and the client is responsible for requesting additional results. The first row number starts with 0.

- <email\_address> is the email address of a registered GeneCruiser user.

## Examples

Searching for a gene with the single variation identifier:

*http://genecruiser.broad.mit.edu/genecruiser3\_services/rest/gene/byVariationIdOutputIdTypes?id=rs858528&outputIdTypes=EntrezGene+UniGene+EMBL&firstResult=0&email=joe@example.com*

This returns a single gene. Some of the EMBL accessions have been removed for brevity.

```
<ns:byVariationIdOutputIdTypesResponse>
  <ns:return>
    <vgc:GeneByVariationIdResults>
      <vgc:GeneByVariationIdResult>
        <vgc:QueryParameter>
          <vgc:VariationName>rs858528</vgc:VariationName>
        </vgc:QueryParameter>
        <vgc:Gene>
          <vgc:GeneId>67394</vgc:GeneId>
          <vgc:Description>
            Cellular tumor antigen p53 (Tumor suppressor
            p53) (Phosphoprotein p53) (Antigen NY-CO-13).
            [Source:Uniprot/SWISSPROT;Acc:P04637]
          </vgc:Description>
          <vgc:Source>ensembl</vgc:Source>
          <vgc:Biotype>protein_coding</vgc:Biotype>
          <vgc:Chromosome>17</vgc:Chromosome>
          <vgc:Start>7512464</vgc:Start>
          <vgc:End>7531642</vgc:End>
          <vgc:Strand>-1</vgc:Strand>
          <vgc:StableId>ENSG00000141510</vgc:StableId>
          <vgc:GenomicIds>
            <vgc:GenomicId>
              <vgc:IdType>EMBL</vgc:IdType>
            </vgc:GenomicId>
            <vgc:Accession>X60014</vgc:Accession>
          </vgc:GenomicIds>
          <vgc:DisplayName>X60014</vgc:DisplayName>
          </vgc:GenomicId>
          <vgc:GenomicId>
            <vgc:IdType>EMBL</vgc:IdType>
          </vgc:GenomicId>
          <vgc:Accession>AF210308</vgc:Accession>
          <vgc:DisplayName>AF210308</vgc:DisplayName>
          </vgc:GenomicId>
          <vgc:GenomicId>
            <vgc:IdType>EntrezGene</vgc:IdType>
            <vgc:Accession>7157</vgc:Accession>
          </vgc:GenomicId>
        </vgc:Gene>
      </vgc:GeneByVariationIdResult>
    </vgc:GeneByVariationIdResults>
  </ns:return>
</ns:byVariationIdOutputIdTypesResponse>
```

```

        <vgc:DisplayName>TP53</vgc:DisplayName>
        </vgc:GenomicId>
    </vgc:GenomicIds>
</vgc:Gene>
</vgc:GeneByVariationIdResult>
</vgc:GeneByVariationIdResults>
</ns:return>
</ns:byVariationIdOutputIdTypesResponse>

```

## 5.4.2 Searching in the 5' flanking region

This web service returns genes for which the variations specified by the given variation identifiers are in the 5' flanking region of the gene. The variation identifier is the “rs number” of the variation.

URL:

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/gene/byVariationIdFivePrime?id=<id\\_list>&fivePrimeSize=<five\\_prime\\_window\\_size>&firstResult=<first\\_result>&email=<email\\_address>](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/gene/byVariationIdFivePrime?id=<id_list>&fivePrimeSize=<five_prime_window_size>&firstResult=<first_result>&email=<email_address>)*

Where:

- <id\_list> is a list of variation identifiers. Identifiers are separated by “+” symbols. For example: rs858528+rs2856753
- <five\_prime\_window\_size> is an integer number of base pairs to search in the 5' direction from the end of the gene.
- <first\_result> indicates the index of the first row to be returned. The web service will only return a set number of rows and the client is responsible for requesting additional results. The first row number starts with 0.
- <email\_address> is the email address of a registered GeneCruiser user.

### Examples

Searching for a gene with the single variation identifier:

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/gene/byVariationIdFivePrime?id=rs17879312&fivePrimeSize=1000&firstResult=0&email=joe@example.com](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/gene/byVariationIdFivePrime?id=rs17879312&fivePrimeSize=1000&firstResult=0&email=joe@example.com)*

This returns a single gene:

```

<ns:byVariationIdFivePrimeResponse>
  <ns:return>
    <vgc:GeneByVariationIdResults>
      <vgc:GeneByVariationIdResult>
        <vgc:QueryParameter>

        <vgc:VariationName>rs17879312</vgc:VariationName>
        </vgc:QueryParameter>

```

```

    <vgc:Gene>
      <vgc:GeneId>67394</vgc:GeneId>
      <vgc:Description>
        Cellular tumor antigen p53 (Tumor suppressor
        p53) (Phosphoprotein p53) (Antigen NY-CO-13).
        [Source:Uniprot/SWISSPROT;Acc:P04637]
      </vgc:Description>
      <vgc:Source>ensembl</vgc:Source>
      <vgc:Biotype>protein_coding</vgc:Biotype>
      <vgc:Chromosome>17</vgc:Chromosome>
      <vgc:Start>7512464</vgc:Start>
      <vgc:End>7531642</vgc:End>
      <vgc:Strand>-1</vgc:Strand>
      <vgc:StableId>ENSG00000141510</vgc:StableId>
    </vgc:Gene>
  </vgc:GeneByVariationIdResult>
</vgc:GeneByVariationIdResults>
</ns:return>
</ns:byVariationIdFivePrimeResponse>

```

### 5.4.3 Searching in the 5' flanking region OutputIdTypes

This web service returns genes for which the variations specified by the given variation identifiers are in the 5' flanking region of the gene. The variation identifier is the “rs number” of the variation.

This service also allows the caller to specify which external database accessions should be displayed. Note that retrieving the external accessions is costly, so unneeded accessions should not be retrieved for best performance.

URL:

*http://genecruiser.broad.mit.edu/genecruiser3\_services/rest/gene/byVariationIdFivePrime?id=<id\_list>&fivePrimeSize=<five\_prime\_window\_size>&outputIdTypes=<output\_id\_types>&firstResult=<first\_result>&email=<email\_address>*

Where:

- <id\_list> is a list of variation identifiers. Identifiers are separated by “+” symbols. For example: rs858528+rs2856753
- <five\_prime\_window\_size> is an integer number of base pairs to search in the 5' direction from the end of the gene.
- <output\_id\_types> is a list of output identifier types. The possible values for output\_id\_type can be retrieved using General/outputIdTypes service.
- <first\_result> indicates the index of the first row to be returned. The web service will only return a set number of rows and the client is responsible for requesting additional results. The first row number starts with 0.
- <email\_address> is the email address of a registered GeneCruiser user.

### Examples

Searching for a gene with the single variation identifier:

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/gene/byVariationIdFivePrimeOutputIdTypes?id=rs17879312&fivePrimeSize=1000&outputIdTypes=EntrezGene+UniGene+EMBL&firstResult=0&email=joe@example.com](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/gene/byVariationIdFivePrimeOutputIdTypes?id=rs17879312&fivePrimeSize=1000&outputIdTypes=EntrezGene+UniGene+EMBL&firstResult=0&email=joe@example.com)*

This returns a single gene:

```
<ns:byVariationIdFivePrimeOutputIdTypesResponse>
  <ns:return>
    <vgc:GeneByVariationIdResults>
      <vgc:GeneByVariationIdResult>
        <vgc:QueryParameter>

        <vgc:VariationName>rs17879312</vgc:VariationName>
        </vgc:QueryParameter>
        <vgc:Gene>
          <vgc:GeneId>67394</vgc:GeneId>
          <vgc:Description>
            Cellular tumor antigen p53 (Tumor suppressor
            p53) (Phosphoprotein p53) (Antigen NY-CO-13).
            [Source:Uniprot/SWISSPROT;Acc:P04637]
          </vgc:Description>
          <vgc:Source>ensembl</vgc:Source>
          <vgc:Biotype>protein_coding</vgc:Biotype>
          <vgc:Chromosome>17</vgc:Chromosome>
          <vgc:Start>7512464</vgc:Start>
          <vgc:End>7531642</vgc:End>
          <vgc:Strand>-1</vgc:Strand>
          <vgc:StableId>ENSG00000141510</vgc:StableId>
          <vgc:GenomicIds>
            <vgc:GenomicId>
              <vgc:IdType>EntrezGene</vgc:IdType>
              <vgc:Accession>7157</vgc:Accession>

            <vgc:GenomicId>
              <vgc:IdType>EMBL</vgc:IdType>
              <vgc:Accession>X60011</vgc:Accession>

            <vgc:GenomicId>
              <vgc:IdType>EMBL</vgc:IdType>
              <vgc:Accession>AB082923</vgc:Accession>

            <vgc:GenomicId>
              <vgc:IdType>EMBL</vgc:IdType>
              <vgc:Accession>AB082923</vgc:Accession>
          </vgc:GenomicIds>
        </vgc:Gene>
      </vgc:GeneByVariationIdResult>
    </vgc:GeneByVariationIdResults>
  </ns:return>
</ns:byVariationIdFivePrimeOutputIdTypesResponse>
```

```

        </vgc:GeneByVariationIdResult>
      </vgc:GeneByVariationIdResults>
    </ns:return>
  </ns:byVariationIdFivePrimeOutputIdTypesResponse>

```

#### 5.4.4 Searching in the 3' flanking region

This web service returns genes for which the variations specified by the given variation identifiers are in the 3' flanking region of the gene. The variation identifier is the “rs number” of the variation.

URL:

*http://genecruiser.broad.mit.edu/genecruiser3\_services/rest/gene/byVariationIdThreePrime?id=<id\_list>&threePrimeSize=<three\_prime\_window\_size>&firstResult=<first\_result>&email=<email\_address>*

Where:

- <id\_list> is a list of variation identifiers. Identifiers are separated by “+” symbols. For example: rs858528+rs2856753
- <three\_prime\_window\_size> is an integer number of base pairs to search in the 3' direction from the end of the gene.
- <first\_result> indicates the index of the first row to be returned. The web service will only return a set number of rows and the client is responsible for requesting additional results. The first row number starts with 0.
- <email\_address> is the email address of a registered GeneCruiser user.

#### Examples

Searching for a gene with the single variation identifier:

*http://genecruiser.broad.mit.edu/genecruiser3\_services/rest/gene/byVariationIdThreePrime?id=rs17885780&threePrimeSize=1000&firstResult=0&email=joe@example.com*

This returns a single gene:

```

<ns:byVariationIdThreePrimeResponse>
  <ns:return>
    <vgc:GeneByVariationIdResults>
      <vgc:GeneByVariationIdResult>
        <vgc:QueryParameter>
          <vgc:VariationName>rs17885780</vgc:VariationName>
        </vgc:QueryParameter>
        <vgc:Gene>
          <vgc:GeneId>67394</vgc:GeneId>
          <vgc:Description>
            Cellular tumor antigen p53 (Tumor suppressor
            p53) (Phosphoprotein p53) (Antigen NY-CO-13).
            [Source:Uniprot/SWISSPROT;Acc:P04637]
          </vgc:Description>
        </vgc:Gene>
      </vgc:GeneByVariationIdResult>
    </vgc:GeneByVariationIdResults>
  </ns:return>
</ns:byVariationIdThreePrimeResponse>

```

```

        </vgc:Description>
        <vgc:Source>ensembl</vgc:Source>
        <vgc:Biotype>protein_coding</vgc:Biotype>
        <vgc:Chromosome>17</vgc:Chromosome>
        <vgc:Start>7512464</vgc:Start>
        <vgc:End>7531642</vgc:End>
        <vgc:Strand>-1</vgc:Strand>
        <vgc:StableId>ENSG00000141510</vgc:StableId>
    </vgc:Gene>
  </vgc:GeneByVariationIdResult>
</vgc:GeneByVariationIdResults>
</ns:return>
</ns:byVariationIdThreePrimeResponse>

```

### 5.4.5 Searching in the 3' flanking region OutputIdTypes

This web service returns genes for which the variations specified by the given variation identifiers are in the 3' flanking region of the gene. The variation identifier is the “rs number” of the variation.

This service also allows the caller to specify which external database accessions should be displayed. Note that retrieving the external accessions is costly, so unneeded accessions should not be retrieved for best performance.

URL:

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/gene/byVariationIdThreePrime?id=<id\\_list>&threePrimeSize=<three\\_prime\\_window\\_size>&outputIdTypes=<output\\_id\\_types>&firstResult=<first\\_result>&email=<email\\_address>](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/gene/byVariationIdThreePrime?id=<id_list>&threePrimeSize=<three_prime_window_size>&outputIdTypes=<output_id_types>&firstResult=<first_result>&email=<email_address>)*

Where:

- <id\_list> is a list of variation identifiers. Identifiers are separated by “+” symbols. For example: rs858528+rs2856753
- <three\_prime\_window\_size> is an integer number of base pairs to search in the 3' direction from the end of the gene.
- <output\_id\_types> is a list of output identifier types. The possible values for output\_id\_type can be retrieved using General/outputIdTypes service.
- <first\_result> indicates the index of the first row to be returned. The web service will only return a set number of rows and the client is responsible for requesting additional results. The first row number starts with 0.
- <email\_address> is the email address of a registered GeneCruiser user.

### Examples

Searching for a gene with the single variation identifier:

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/gene/byVariationIdThreePrimeOutputIdTypes?id=rs17885780&threePrimeSize=1000&outputIdTypes=EntrezGene+UniGene+EMBL&firstResult=0&email=joe@example.com](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/gene/byVariationIdThreePrimeOutputIdTypes?id=rs17885780&threePrimeSize=1000&outputIdTypes=EntrezGene+UniGene+EMBL&firstResult=0&email=joe@example.com)*

This returns a single gene:

```
<ns:byVariationIdThreePrimeOutputIdTypesResponse>
  <ns:return>
    <vgc:GeneByVariationIdResults>
      <vgc:GeneByVariationIdResult>
        <vgc:QueryParameter>

        <vgc:VariationName>rs17885780</vgc:VariationName>
        </vgc:QueryParameter>
        <vgc:Gene>
          <vgc:GeneId>67394</vgc:GeneId>
          <vgc:Description>
            Cellular tumor antigen p53 (Tumor suppressor
            p53) (Phosphoprotein p53) (Antigen NY-CO-13).
            [Source:Uniprot/SWISSPROT;Acc:P04637]
          </vgc:Description>
          <vgc:Source>ensembl</vgc:Source>
          <vgc:Biotype>protein_coding</vgc:Biotype>
          <vgc:Chromosome>17</vgc:Chromosome>
          <vgc:Start>7512464</vgc:Start>
          <vgc:End>7531642</vgc:End>
          <vgc:Strand>-1</vgc:Strand>
          <vgc:StableId>ENSG00000141510</vgc:StableId>
          <vgc:GenomicIds>
            <vgc:GenomicId>
              <vgc:IdType>EntrezGene</vgc:IdType>
              <vgc:Accession>7157</vgc:Accession>

            <vgc:GenomicId>
              <vgc:IdType>EMBL</vgc:IdType>
              <vgc:Accession>DQ486039</vgc:Accession>

            <vgc:GenomicId>
              <vgc:IdType>EMBL</vgc:IdType>
              <vgc:Accession>M14694</vgc:Accession>

            <vgc:GenomicId>
              <vgc:IdType>EMBL</vgc:IdType>
              <vgc:Accession>DQ486039</vgc:Accession>

            ...
            <vgc:GenomicId>
              <vgc:IdType>EMBL</vgc:IdType>
              <vgc:Accession>M14694</vgc:Accession>

          </vgc:GenomicIds>
        </vgc:Gene>
      </vgc:GeneByVariationIdResult>
    </vgc:GeneByVariationIdResults>
  </ns:return>
</ns:byVariationIdThreePrimeOutputIdTypesResponse>
```

### 5.4.6 Searching in the gene or in the 5' flanking region

This web service returns genes for which the variations specified by the given variation identifiers are in the gene or in the 5' flanking region of the gene. The variation identifier is the “rs number” of the variation.

URL:

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/gene/byVariationIdInGeneFivePrime?id=<id\\_list>&fivePrimeSize=<five\\_prime\\_window\\_size>&firstResult=<first\\_result>&email=<email\\_address>](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/gene/byVariationIdInGeneFivePrime?id=<id_list>&fivePrimeSize=<five_prime_window_size>&firstResult=<first_result>&email=<email_address>)*

Where:

- <id\_list> is a list of variation identifiers. Identifiers are separated by “+” symbols. For example: rs858528+rs2856753
- <five\_prime\_window\_size> is an integer number of base pairs to search in the 5' direction from the end of the gene.
- <first\_result> indicates the index of the first row to be returned. The web service will only return a set number of rows and the client is responsible for requesting additional results. The first row number starts with 0.
- <email\_address> is the email address of a registered GeneCruiser user.

### Examples

Searching for a gene with the single variation identifier:

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/gene/byVariationIdInGeneFivePrime?id=rs858528&fivePrimeSize=1000&firstResult=0&email=joe@example.com](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/gene/byVariationIdInGeneFivePrime?id=rs858528&fivePrimeSize=1000&firstResult=0&email=joe@example.com)*

```
<ns:byVariationIdInGeneFivePrimeResponse>
  <ns:return>
    <vgc:GeneByVariationIdResults>
      <vgc:GeneByVariationIdResult>
        <vgc:QueryParameter>
          <vgc:VariationName>rs858528</vgc:VariationName>
        </vgc:QueryParameter>
        <vgc:Gene>
          <vgc:GeneId>67394</vgc:GeneId>
          <vgc:Description>
            Cellular tumor antigen p53 (Tumor suppressor
            p53) (Phosphoprotein p53) (Antigen NY-CO-13).
            [Source:Uniprot/SWISSPROT;Acc:P04637]
          </vgc:Description>
          <vgc:Source>ensembl</vgc:Source>
          <vgc:Biotype>protein_coding</vgc:Biotype>
          <vgc:Chromosome>17</vgc:Chromosome>
          <vgc:Start>7512464</vgc:Start>
          <vgc:End>7531642</vgc:End>
          <vgc:Strand>-1</vgc:Strand>
          <vgc:StableId>ENSG00000141510</vgc:StableId>
        </vgc:Gene>
      </vgc:GeneByVariationIdResult>
    </vgc:GeneByVariationIdResults>
  </ns:return>
</ns:byVariationIdInGeneFivePrimeResponse>
```

```

        </vgc:GeneByVariationIdResult>
    </vgc:GeneByVariationIdResults>
</ns:return>
</ns:byVariationIdInGeneFivePrimeResponse>

```

### 5.4.7 Searching in the gene or in the 5' flanking region OutputIdTypes

This web service returns genes for which the variations specified by the given variation identifiers are in the gene or in the 5' flanking region of the gene. The variation identifier is the "rs number" of the variation.

This service also allows the caller to specify which external database accessions should be displayed. Note that retrieving the external accessions is costly, so unneeded accessions should not be retrieved for best performance.

URL:

*http://genecruiser.broad.mit.edu/genecruiser3\_services/rest/gene/byVariationIdInGeneFivePrime?id=<id\_list>&fivePrimeSize=<five\_prime\_window\_size>&outputIdTypes=<output\_id\_types>&firstResult=<first\_result>&email=<email\_address>*

Where:

- <id\_list> is a list of variation identifiers. Identifiers are separated by "+" symbols. For example: rs858528+rs2856753
- <five\_prime\_window\_size> is an integer number of base pairs to search in the 5' direction from the end of the gene.
- <output\_id\_types> is a list of output identifier types. The possible values for output\_id\_type can be retrieved using General/outputIdTypes service.
- <first\_result> indicates the index of the first row to be returned. The web service will only return a set number of rows and the client is responsible for requesting additional results. The first row number starts with 0.
- <email\_address> is the email address of a registered GeneCruiser user.

### Examples

Searching for a gene with the single variation identifier:

*http://genecruiser.broad.mit.edu/genecruiser3\_services/rest/gene/byVariationIdInGeneFivePrimeOutputIdTypes?id=rs858528&fivePrimeSize=1000&outputIdTypes=EntrezGene+UniGene+EMBL&firstResult=0&email=joe@example.com*

```

<ns:byVariationIdInGeneFivePrimeOutputIdTypesResponse>
  <ns:return>
    <vgc:GeneByVariationIdResults>
      <vgc:GeneByVariationIdResult>
        <vgc:QueryParameter>
          <vgc:VariationName>rs858528</vgc:VariationName>
        </vgc:QueryParameter>
        <vgc:Gene>

```

```

    <vgc:GeneId>67394</vgc:GeneId>
    <vgc:Description>
    Cellular tumor antigen p53 (Tumor suppressor
    p53) (Phosphoprotein p53) (Antigen NY-CO-13).
    [Source:Uniprot/SWISSPROT;Acc:P04637]
    </vgc:Description>
    <vgc:Source>ensembl</vgc:Source>
    <vgc:Biotype>protein_coding</vgc:Biotype>
    <vgc:Chromosome>17</vgc:Chromosome>
    <vgc:Start>7512464</vgc:Start>
    <vgc:End>7531642</vgc:End>
    <vgc:Strand>-1</vgc:Strand>
    <vgc:StableId>ENSG00000141510</vgc:StableId>
    <vgc:GenomicIds>
      <vgc:GenomicId>
        <vgc:IdType>EntrezGene</vgc:IdType>
        <vgc:Accession>7157</vgc:Accession>
      </vgc:GenomicId>
    </vgc:GenomicIds>
    <vgc:DisplayName>TP53</vgc:DisplayName>
    </vgc:GenomicId>
    <vgc:GenomicId>
      <vgc:IdType>EMBL</vgc:IdType>
    </vgc:GenomicId>
    <vgc:Accession>X60012</vgc:Accession>
    <vgc:DisplayName>X60012</vgc:DisplayName>
    </vgc:GenomicId>
    ...
    <vgc:GenomicId>
      <vgc:IdType>EMBL</vgc:IdType>
    </vgc:GenomicId>
    <vgc:Accession>AF210310</vgc:Accession>
    <vgc:DisplayName>AF210310</vgc:DisplayName>
    </vgc:GenomicId>
    </vgc:GenomicIds>
  </vgc:Gene>
</vgc:GeneByVariationIdResult>
</vgc:GeneByVariationIdResults>
</ns:return>
</ns:byVariationIdInGeneFivePrimeOutputIdTypesResponse>

```

#### 5.4.8 Searching in the gene or in the 3' flanking region

This web service returns genes for which the variations specified by the given variation identifiers are in the gene or in the 3' flanking region of the gene. The variation identifier is the "rs number" of the variation.

URL:

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/gene/byVariationIdInGeneThreePrime?id=<id\\_list>&threePrimeSize=<three\\_prime\\_window\\_size>&firstResult=<first\\_result>&email=<email\\_address>](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/gene/byVariationIdInGeneThreePrime?id=<id_list>&threePrimeSize=<three_prime_window_size>&firstResult=<first_result>&email=<email_address>)*

Where:

- <id\_list> is a list of variation identifiers. Identifiers are separated by “+” symbols. For example: rs858528+rs2856753
- <three\_prime\_window\_size> is an integer number of base pairs to search in the 3’ direction from the end of the gene.
- <first\_result> indicates the index of the first row to be returned. The web service will only return a set number of rows and the client is responsible for requesting additional results. The first row number starts with 0.
- <email\_address> is the email address of a registered GeneCruiser user.

## Examples

Searching for a gene with the single variation identifier:

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/gene/byVariationIdInGeneThreePrime?id=rs17885780&threePrimeSize=1000&firstResult=0&email=joe@example.com](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/gene/byVariationIdInGeneThreePrime?id=rs17885780&threePrimeSize=1000&firstResult=0&email=joe@example.com)*

This returns two genes:

```
<ns:byVariationIdInGeneThreePrimeResponse>
  <ns:return>
    <vgc:GeneByVariationIdResults>
      <vgc:GeneByVariationIdResult>
        <vgc:QueryParameter>

        <vgc:VariationName>rs17885780</vgc:VariationName>
        </vgc:QueryParameter>
        <vgc:Gene>
          <vgc:GeneId>67394</vgc:GeneId>
          <vgc:Description>
            Cellular tumor antigen p53 (Tumor suppressor
            p53) (Phosphoprotein p53) (Antigen NY-CO-13).
            [Source:Uniprot/SWISSPROT;Acc:P04637]
          </vgc:Description>
          <vgc:Source>ensembl</vgc:Source>
          <vgc:Biotype>protein_coding</vgc:Biotype>
          <vgc:Chromosome>17</vgc:Chromosome>
          <vgc:Start>7512464</vgc:Start>
          <vgc:End>7531642</vgc:End>
          <vgc:Strand>-1</vgc:Strand>
          <vgc:StableId>ENSG00000141510</vgc:StableId>
        </vgc:Gene>
      </vgc:GeneByVariationIdResult>
      <vgc:GeneByVariationIdResult>
        <vgc:QueryParameter>

        <vgc:VariationName>rs17885780</vgc:VariationName>
        </vgc:QueryParameter>
        <vgc:Gene>
          <vgc:GeneId>71418</vgc:GeneId>
          <vgc:Description>
            WD repeat protein 79. [Source:Uniprot/SWISSPROT;Acc:Q9BUR4]
```

```

        </vgc:Description>
        <vgc:Source>ensembl</vgc:Source>
        <vgc:Biotype>protein_coding</vgc:Biotype>
        <vgc:Chromosome>17</vgc:Chromosome>
        <vgc:Start>7530723</vgc:Start>
        <vgc:End>7547542</vgc:End>
        <vgc:Strand>1</vgc:Strand>
        <vgc:StableId>ENSG00000141499</vgc:StableId>
    </vgc:Gene>
  </vgc:GeneByVariationIdResult>
</vgc:GeneByVariationIdResults>
</ns:return>
</ns:byVariationIdInGeneThreePrimeResponse>

```

### 5.4.9 Searching in the gene or in the 3' flanking region OutputIdTypes

This web service returns genes for which the variations specified by the given variation identifiers are in the gene or in the 3' flanking region of the gene. The variation identifier is the "rs number" of the variation.

This service also allows the caller to specify which external database accessions should be displayed. Note that retrieving the external accessions is costly, so unneeded accessions should not be retrieved for best performance.

URL:

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/gene/byVariationIdInGeneThreePrime?id=<id\\_list>&threePrimeSize=<three\\_prime\\_window\\_size>&outputIdTypes=<output\\_id\\_types>&firstResult=<first\\_result>&email=<email\\_address>](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/gene/byVariationIdInGeneThreePrime?id=<id_list>&threePrimeSize=<three_prime_window_size>&outputIdTypes=<output_id_types>&firstResult=<first_result>&email=<email_address>)*

Where:

- <id\_list> is a list of variation identifiers. Identifiers are separated by "+" symbols. For example: rs858528+rs2856753
- <three\_prime\_window\_size> is an integer number of base pairs to search in the 3' direction from the end of the gene.
- <output\_id\_types> is a list of output identifier types. The possible values for output\_id\_type can be retrieved using General/outputIdTypes service.
- <first\_result> indicates the index of the first row to be returned. The web service will only return a set number of rows and the client is responsible for requesting additional results. The first row number starts with 0.
- <email\_address> is the email address of a registered GeneCruiser user.

### Examples

Searching for a gene with the single variation identifier:

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/gene/byVariationIdInGeneThreePrimeOutputIdTypes?id=rs17885780&threePrimeSize=1000&outputIdTypes=EntrezGene+UniGene+EMBL&firstResult=0&email=joe@example.com](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/gene/byVariationIdInGeneThreePrimeOutputIdTypes?id=rs17885780&threePrimeSize=1000&outputIdTypes=EntrezGene+UniGene+EMBL&firstResult=0&email=joe@example.com)*

This returns two genes:

```
<ns:byVariationIdInGeneThreePrimeOutputIdTypesResponse>
  <ns:return>
    <vgc:GeneByVariationIdResults>
      <vgc:GeneByVariationIdResult>
        <vgc:QueryParameter>

        <vgc:VariationName>rs17885780</vgc:VariationName>
        </vgc:QueryParameter>
        <vgc:Gene>
          <vgc:GeneId>67394</vgc:GeneId>
          <vgc:Description>
            Cellular tumor antigen p53 (Tumor suppressor
            p53) (Phosphoprotein p53) (Antigen NY-CO-13).
            [Source:Uniprot/SWISSPROT;Acc:P04637]
          </vgc:Description>
          <vgc:Source>ensembl</vgc:Source>
          <vgc:Biotype>protein_coding</vgc:Biotype>
          <vgc:Chromosome>17</vgc:Chromosome>
          <vgc:Start>7512464</vgc:Start>
          <vgc:End>7531642</vgc:End>
          <vgc:Strand>-1</vgc:Strand>
          <vgc:StableId>ENSG00000141510</vgc:StableId>
          <vgc:GenomicIds>
            <vgc:GenomicId>
              <vgc:IdType>EntrezGene</vgc:IdType>
              <vgc:Accession>7157</vgc:Accession>

            <vgc:GenomicId>
              <vgc:IdType>EMBL</vgc:IdType>
              <vgc:Accession>BT019622</vgc:Accession>

            <vgc:GenomicId>
              <vgc:IdType>EMBL</vgc:IdType>
              <vgc:Accession>X60018</vgc:Accession>

          </vgc:GenomicIds>
        </vgc:Gene>
      </vgc:GeneByVariationIdResult>
    </vgc:GeneByVariationIdResults>
  </ns:return>
</ns:byVariationIdInGeneThreePrimeOutputIdTypesResponse>
```

```

    <vgc:Gene>
      <vgc:GeneId>71418</vgc:GeneId>
      <vgc:Description>
        WD          repeat          protein          79.
[Source:Uniprot/SWISSPROT;Acc:Q9BUR4]
      </vgc:Description>
      <vgc:Source>ensembl</vgc:Source>
      <vgc:Biotype>protein_coding</vgc:Biotype>
      <vgc:Chromosome>17</vgc:Chromosome>
      <vgc:Start>7530723</vgc:Start>
      <vgc:End>7547542</vgc:End>
      <vgc:Strand>1</vgc:Strand>
      <vgc:StableId>ENSG00000141499</vgc:StableId>
      <vgc:GenomicIds>
        <vgc:GenomicId>
          <vgc:IdType>UniGene</vgc:IdType>

<vgc:Accession>Hs.437460</vgc:Accession>

<vgc:DisplayName>Hs.437460</vgc:DisplayName>
  </vgc:GenomicId>
  <vgc:GenomicId>
    <vgc:IdType>EntrezGene</vgc:IdType>

<vgc:Accession>55135</vgc:Accession>

<vgc:DisplayName>WDR79</vgc:DisplayName>
  </vgc:GenomicId>
  <vgc:GenomicId>
    <vgc:IdType>EMBL</vgc:IdType>

<vgc:Accession>AY766322</vgc:Accession>

<vgc:DisplayName>AY766322</vgc:DisplayName>
  </vgc:GenomicId>
  ...
  <vgc:GenomicId>
    <vgc:IdType>EMBL</vgc:IdType>

<vgc:Accession>BC002336</vgc:Accession>

<vgc:DisplayName>BC002336</vgc:DisplayName>
  </vgc:GenomicId>
  </vgc:GenomicIds>
</vgc:Gene>
  </vgc:GeneByVariationIdResult>
</vgc:GeneByVariationIdResults>
</ns:return>
</ns:byVariationIdInGeneThreePrimeOutputIdTypesResponse>

```

#### 5.4.10 Searching in the 5' or 3' flanking region

This web service returns genes for which the variations specified by the given variation identifiers are in 5' or 3' flanking regions of the gene. The variation identifier is the “rs number” of the variation.

URL:

*http://genecruiser.broad.mit.edu/genecruiser3\_services/rest/gene/byVariationIdFivePrimeThreePrime?id=<id\_list>&fivePrimeSize=<five\_prime\_window\_size>&threePrimeSize=<three\_prime\_window\_size>&firstResult=<first\_result>&email=<email\_address>*

Where:

- <id\_list> is a list of variation identifiers. Identifiers are separated by “+” symbols. For example: rs858528+rs2856753
- <three\_prime\_window\_size> is an integer number of base pairs to search in the 5’ direction from the end of the gene.
- <three\_prime\_window\_size> is an integer number of base pairs to search in the 3’ direction from the end of the gene.
- <first\_result> indicates the index of the first row to be returned. The web service will only return a set number of rows and the client is responsible for requesting additional results. The first row number starts with 0.
- <email\_address> is the email address of a registered GeneCruiser user.

### **Examples**

Searching for a gene with the single variation identifier:

*http://genecruiser.broad.mit.edu/genecruiser3\_services/rest/gene/byVariationIdFivePrimeThreePrime?id=rs12949853&fivePrimeSize=1000&threePrimeSize=1000&firstResult=0&email=joe@example.com*

This returns a single gene:

```
<ns:byVariationIdFivePrimeThreePrimeResponse>
  <ns:return>
    <vgc:GeneByVariationIdResults>
      <vgc:GeneByVariationIdResult>
        <vgc:QueryParameter>
          <vgc:VariationName>rs12949853</vgc:VariationName>
        </vgc:QueryParameter>
        <vgc:Gene>
          <vgc:GeneId>67394</vgc:GeneId>
          <vgc:Description>
            Cellular tumor antigen p53 (Tumor suppressor
            p53) (Phosphoprotein p53) (Antigen NY-CO-13).
            [Source:Uniprot/SWISSPROT;Acc:P04637]
          </vgc:Description>
          <vgc:Source>ensembl</vgc:Source>
          <vgc:Biotype>protein_coding</vgc:Biotype>
          <vgc:Chromosome>17</vgc:Chromosome>
          <vgc:Start>7512464</vgc:Start>
          <vgc:End>7531642</vgc:End>
          <vgc:Strand>-1</vgc:Strand>
```

```

        <vgc:StableId>ENSG00000141510</vgc:StableId>
      </vgc:Gene>
    </vgc:GeneByVariationIdResult>
  </vgc:GeneByVariationIdResults>
</ns:return>
</ns:byVariationIdFivePrimeThreePrimeResponse>

```

#### 5.4.11 Searching in the 5' or 3' flanking region OutputIdTypes

This web service returns genes for which the variations specified by the given variation identifiers are in 5' or 3' flanking regions of the gene. The variation identifier is the “rs number” of the variation.

This service also allows the caller to specify which external database accessions should be displayed. Note that retrieving the external accessions is costly, so unneeded accessions should not be retrieved for best performance.

URL:

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/gene/byVariationIdFivePrimeThreePrime?id=<id\\_list>&fivePrimeSize=<five\\_prime\\_window\\_size>&threePrimeSize=<three\\_prime\\_window\\_size>&outputIdTypes=<output\\_id\\_types>&firstResult=<first\\_result>&email=<email\\_address>](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/gene/byVariationIdFivePrimeThreePrime?id=<id_list>&fivePrimeSize=<five_prime_window_size>&threePrimeSize=<three_prime_window_size>&outputIdTypes=<output_id_types>&firstResult=<first_result>&email=<email_address>)*

Where:

- <id\_list> is a list of variation identifiers. Identifiers are separated by “+” symbols. For example: rs858528+rs2856753
- <three\_prime\_window\_size> is an integer number of base pairs to search in the 5' direction from the end of the gene.
- <three\_prime\_window\_size> is an integer number of base pairs to search in the 3' direction from the end of the gene.
- <output\_id\_types> is a list of output identifier types. The possible values for output\_id\_type can be retrieved using General/outputIdTypes service.
- <first\_result> indicates the index of the first row to be returned. The web service will only return a set number of rows and the client is responsible for requesting additional results. The first row number starts with 0.
- <email\_address> is the email address of a registered GeneCruiser user.

#### Examples

Searching for a gene with the single variation identifier:

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/gene/byVariationIdFivePrimeThreePrimeOutputIdTypes?id=rs12949853&fivePrimeSize=1000&threePrimeSize=1000&outputIdTypes=EntrezGene+UniGene+EMBL&firstResult=0&email=joe@example.com](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/gene/byVariationIdFivePrimeThreePrimeOutputIdTypes?id=rs12949853&fivePrimeSize=1000&threePrimeSize=1000&outputIdTypes=EntrezGene+UniGene+EMBL&firstResult=0&email=joe@example.com)*

This returns a single gene:

```

<ns:byVariationIdFivePrimeThreePrimeOutputIdTypesResponse>
  <ns:return>
    <vgc:GeneByVariationIdResults>
      <vgc:GeneByVariationIdResult>
        <vgc:QueryParameter>

        <vgc:VariationName>rs12949853</vgc:VariationName>
        </vgc:QueryParameter>
        <vgc:Gene>
          <vgc:GeneId>67394</vgc:GeneId>
          <vgc:Description>
            Cellular tumor antigen p53 (Tumor suppressor
            p53) (Phosphoprotein p53) (Antigen NY-CO-13).
            [Source:Uniprot/SWISSPROT;Acc:P04637]
          </vgc:Description>
          <vgc:Source>ensembl</vgc:Source>
          <vgc:Biotype>protein_coding</vgc:Biotype>
          <vgc:Chromosome>17</vgc:Chromosome>
          <vgc:Start>7512464</vgc:Start>
          <vgc:End>7531642</vgc:End>
          <vgc:Strand>-1</vgc:Strand>
          <vgc:StableId>ENSG00000141510</vgc:StableId>
          <vgc:GenomicIds>
            <vgc:GenomicId>
              <vgc:IdType>EntrezGene</vgc:IdType>
              <vgc:Accession>7157</vgc:Accession>

            <vgc:GenomicId>
              <vgc:IdType>EMBL</vgc:IdType>
              <vgc:Accession>BC003596</vgc:Accession>

            <vgc:GenomicId>
              <vgc:IdType>EMBL</vgc:IdType>
              <vgc:Accession>DQ075362</vgc:Accession>

            ...
          </vgc:GenomicIds>
        </vgc:Gene>
      </vgc:GeneByVariationIdResult>
    </vgc:GeneByVariationIdResults>
  </ns:return>
</ns:byVariationIdFivePrimeThreePrimeOutputIdTypesResponse>

```

### 5.4.12 Searching in the gene or in the 5' or 3' flanking region

This web service returns genes for which the variations specified by the given variation identifiers are in the gene or in 5' or 3' flanking regions of the gene. The variation identifier is the "rs number" of the variation.

URL:

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/gene/byVariationIdInGeneFivePrimeThreePrime?id=<id\\_list>&fivePrimeSize=<five\\_prime\\_window\\_size>&threePrimeSize=<three\\_prime\\_window\\_size>&firstResult=<first\\_result>&email=<email\\_address>](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/gene/byVariationIdInGeneFivePrimeThreePrime?id=<id_list>&fivePrimeSize=<five_prime_window_size>&threePrimeSize=<three_prime_window_size>&firstResult=<first_result>&email=<email_address>)*

Where:

- <id\_list> is a list of variation identifiers. Identifiers are separated by "+" symbols. For example: rs858528+rs2856753
- <five\_prime\_window\_size> is an integer number of base pairs to search in the 5' direction from the end of the gene.
- <three\_prime\_window\_size> is an integer number of base pairs to search in the 3' direction from the end of the gene.
- <first\_result> indicates the index of the first row to be returned. The web service will only return a set number of rows and the client is responsible for requesting additional results. The first row number starts with 0.
- <email\_address> is the email address of a registered GeneCruiser user.

#### Examples

Searching for a gene with the single variation identifier:

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/gene/byVariationIdInGeneFivePrimeThreePrime?id=rs12949853&fivePrimeSize=1000&threePrimeSize=1000&firstResult=0&email=joe@example.com](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/gene/byVariationIdInGeneFivePrimeThreePrime?id=rs12949853&fivePrimeSize=1000&threePrimeSize=1000&firstResult=0&email=joe@example.com)*

This returns a single gene.

```
<ns:byVariationIdInGeneFivePrimeThreePrimeResponse>
  <ns:return>
    <vgc:GeneByVariationIdResults>
      <vgc:GeneByVariationIdResult>
        <vgc:QueryParameter>

        <vgc:VariationName>rs12949853</vgc:VariationName>
        </vgc:QueryParameter>
        <vgc:Gene>
          <vgc:GeneId>67394</vgc:GeneId>
          <vgc:Description>
            Cellular tumor antigen p53 (Tumor suppressor
            p53) (Phosphoprotein p53) (Antigen NY-CO-13).
            [Source:Uniprot/SWISSPROT;Acc:P04637]
          </vgc:Description>
```

```

        <vgc:Source>ensembl</vgc:Source>
        <vgc:Biotype>protein_coding</vgc:Biotype>
        <vgc:Chromosome>17</vgc:Chromosome>
        <vgc:Start>7512464</vgc:Start>
        <vgc:End>7531642</vgc:End>
        <vgc:Strand>-1</vgc:Strand>
        <vgc:StableId>ENSG00000141510</vgc:StableId>
      </vgc:Gene>
    </vgc:GeneByVariationIdResult>
  </vgc:GeneByVariationIdResults>
</ns:return>
</ns:byVariationIdInGeneFivePrimeThreePrimeResponse>

```

### 5.4.13 Searching in the gene or in the 5' or 3' flanking region OutputIdTypes

This web service returns genes for which the variations specified by the given variation identifiers are in the gene or in 5' or 3' flanking regions of the gene. The variation identifier is the “rs number” of the variation.

This service also allows the caller to specify which external database accessions should be displayed. Note that retrieving the external accessions is costly, so unneeded accessions should not be retrieved for best performance.

Url:

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/gene/byVariationIdInGeneFivePrimeThreePrime?id=<id\\_list>&fivePrimeSize=<five\\_prime\\_window\\_size>&threePrimeSize=<three\\_prime\\_window\\_size>&outputIdTypes=<output\\_id\\_types>&firstResult=<first\\_result>&email=<email\\_address>](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/gene/byVariationIdInGeneFivePrimeThreePrime?id=<id_list>&fivePrimeSize=<five_prime_window_size>&threePrimeSize=<three_prime_window_size>&outputIdTypes=<output_id_types>&firstResult=<first_result>&email=<email_address>)*

Where:

- <id\_list> is a list of variation identifiers. Identifiers are separated by “+” symbols. For example: rs858528+rs2856753
- <five\_prime\_window\_size> is an integer number of base pairs to search in the 5' direction from the end of the gene.
- <three\_prime\_window\_size> is an integer number of base pairs to search in the 3' direction from the end of the gene.
- <output\_id\_types> is a list of output identifier types. The possible values for output\_id\_type can be retrieved using General/outputIdTypes service.
- <first\_result> indicates the index of the first row to be returned. The web service will only return a set number of rows and the client is responsible for requesting additional results. The first row number starts with 0.
- <email\_address> is the email address of a registered GeneCruiser user.

### Examples

Searching for a gene with the single variation identifier:

*http://genecruiser.broad.mit.edu/genecruiser3\_services/rest/gene/byVariationIdInGeneFivePrimeThreePrimeOutputIdTypes?id=rs12949853&fivePrimeSize=1000&threePrimeSize=1000&outputIdTypes=EntrezGene+UniGene+EMBL&firstResult=0&email=joe@example.com*

This returns a single gene.

```
<ns:byVariationIdInGeneFivePrimeThreePrimeOutputIdTypesResponse>
  <ns:return>
    <vgc:GeneByVariationIdResults>
      <vgc:GeneByVariationIdResult>
        <vgc:QueryParameter>

        <vgc:VariationName>rs12949853</vgc:VariationName>
        </vgc:QueryParameter>
        <vgc:Gene>
          <vgc:GeneId>67394</vgc:GeneId>
          <vgc:Description>
            Cellular tumor antigen p53 (Tumor suppressor
            p53) (Phosphoprotein p53) (Antigen NY-CO-13).
            [Source:Uniprot/SWISSPROT;Acc:P04637]
          </vgc:Description>
          <vgc:Source>ensembl</vgc:Source>
          <vgc:Biotype>protein_coding</vgc:Biotype>
          <vgc:Chromosome>17</vgc:Chromosome>
          <vgc:Start>7512464</vgc:Start>
          <vgc:End>7531642</vgc:End>
          <vgc:Strand>-1</vgc:Strand>
          <vgc:StableId>ENSG00000141510</vgc:StableId>
          <vgc:GenomicIds>
            <vgc:GenomicId>
              <vgc:IdType>EntrezGene</vgc:IdType>
              <vgc:Accession>7157</vgc:Accession>

            <vgc:GenomicId>
              <vgc:IdType>EntrezGene</vgc:IdType>
              <vgc:Accession>7157</vgc:Accession>

          <vgc:DisplayName>TP53</vgc:DisplayName>
          </vgc:GenomicId>
          <vgc:GenomicId>
            <vgc:IdType>EMBL</vgc:IdType>

          <vgc:Accession>AF209136</vgc:Accession>

          <vgc:DisplayName>AF209136</vgc:DisplayName>
          </vgc:GenomicId>
          ...
          <vgc:GenomicId>
            <vgc:IdType>EMBL</vgc:IdType>

          <vgc:Accession>AF209132</vgc:Accession>

          <vgc:DisplayName>AF209132</vgc:DisplayName>
          </vgc:GenomicId>
          </vgc:GenomicIds>
        </vgc:Gene>
      </vgc:GeneByVariationIdResult>
    </vgc:GeneByVariationIdResults>
```

```
</ns:return>
</ns:byVariationIdInGeneFivePrimeThreePrimeOutputIdTypesResponse>
```

## 5.5 Genes byVariationIdRegulatory

There are two services for retrieving genes byVariationIdRegulatory – a simple version (byVariationIdRegulatory) and an extended version that also retrieves accession numbers from external databases (byVariationIdRegulatoryOutputIdTypes).

These two services retrieve genes for which the variations identified by the input variation ids are in the regulatory region of the gene.

These services are described below in more detail.

### 5.5.1 Genes byVariationIdRegulatory

This web service retrieve genes for which the variations identified by the input variation ids are in the regulatory region of the gene. The variation identifier is the “rs number” of the variation.

URL:

```
http://genecruiser.broad.mit.edu/genecruiser3_services/rest/gene/byVariationIdRegulatory?id=
<id_list>&firstResult=<first_result>&email=<email_address>
```

Where:

- <id\_list> is a list of variation identifiers. Identifiers are separated by “+” symbols. For example: rs858528+rs2856753
- <first\_result> indicates the index of the first row to be returned. The web service will only return a set number of rows and the client is responsible for requesting additional results. The first row number starts with 0.
- <email\_address> is the email address of a registered GeneCruiser user.

#### 5.5.1.1 Examples

Searching for a gene with the single variation identifier:

```
http://genecruiser.broad.mit.edu/genecruiser3_services/rest/gene/byVariationIdRegulatory?id=r
s5004340&firstResult=0&email=joe@example.com
```

This returns a single gene:

```
<ns:byVariationIdRegulatoryResponse>
  <ns:return>
    <vgc:GeneByVariationIdResults>
      <vgc:GeneByVariationIdResult>
        <vgc:QueryParameter>

        <vgc:VariationName>rs5004340</vgc:VariationName>
```

```

</vgc:QueryParameter>
<vgc:Gene>
  <vgc:GeneId>52239</vgc:GeneId>
  <vgc:Description>
    Eukaryotic translation initiation factor 1b
    (eIF1b) (Protein translation factor SUI1
    homolog GC20).
    [Source:Uniprot/SWISSPROT;Acc:O60739]
  </vgc:Description>
  <vgc:Source>ensembl</vgc:Source>
  <vgc:Biotype>protein_coding</vgc:Biotype>
  <vgc:Chromosome>3</vgc:Chromosome>
  <vgc:Start>40326177</vgc:Start>
  <vgc:End>40328919</vgc:End>
  <vgc:Strand>1</vgc:Strand>
  <vgc:StableId>ENSG00000114784</vgc:StableId>
</vgc:Gene>
</vgc:GeneByVariationIdResult>
</vgc:GeneByVariationIdResults>
</ns:return>
</ns:byVariationIdRegulatoryResponse>

```

## 5.5.2 GenesByVariationIdRegulatoryOutputIdTypes

This web service retrieve genes for which the variations identified by the input variation ids are in the regulatory region of the gene. The identifier is the variation “rs number.”

This service also allows the caller to specify which external database accessions should be displayed. Note that retrieving the external accessions is costly, so unneeded accessions should not be retrieved for best performance.

URL:

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/gene/byVariationIdRegulatoryOutputIdTypes?id=<id\\_list>&outputIdTypes=<output\\_id\\_types>&firstResult=<first\\_result>&email=<email\\_address>](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/gene/byVariationIdRegulatoryOutputIdTypes?id=<id_list>&outputIdTypes=<output_id_types>&firstResult=<first_result>&email=<email_address>)*

Where:

- <id\_list> is a list of identifiers. Identifiers are separated by “+” symbols. For example: rs858528+rs2856753
- <output\_id\_types> is a list of output identifier types. The possible values for output\_id\_type can be retrieved using General/outputIdTypes service.
- <first\_result> indicates the index of the first row to be returned. The web service will only return a set number of rows and the client is responsible for requesting additional results. The first row number starts with 0.
- <email\_address> is the email address of a registered GeneCruiser user.

### 5.5.2.1 Examples

Searching for a gene with the single variation identifier:

[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/gene/byVariationIdRegulatoryOutputIdTypes?id=rs5004340&outputIdTypes=EntrezGene+UniGene+EMBL&firstResult=0&email=joe@example.com](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/gene/byVariationIdRegulatoryOutputIdTypes?id=rs5004340&outputIdTypes=EntrezGene+UniGene+EMBL&firstResult=0&email=joe@example.com)

This returns a single gene. Some of the EMBL accessions have been removed for brevity.

```
<ns:byVariationIdRegulatoryOutputIdTypesResponse>
  <ns:return>
    <vgc:GeneByVariationIdResults>
      <vgc:GeneByVariationIdResult>
        <vgc:QueryParameter>

        <vgc:VariationName>rs5004340</vgc:VariationName>
        </vgc:QueryParameter>
        <vgc:Gene>
          <vgc:GeneId>52239</vgc:GeneId>
          <vgc:Description>
            Eukaryotic translation initiation factor 1b
            (eIF1b) (Protein translation factor SUI1
            homolog GC20).
            [Source:Uniprot/SWISSPROT;Acc:O60739]
          </vgc:Description>
          <vgc:Source>ensembl</vgc:Source>
          <vgc:Biotype>protein_coding</vgc:Biotype>
          <vgc:Chromosome>3</vgc:Chromosome>
          <vgc:Start>40326177</vgc:Start>
          <vgc:End>40328919</vgc:End>
          <vgc:Strand>1</vgc:Strand>
          <vgc:StableId>ENSG00000114784</vgc:StableId>
          <vgc:GenomicIds>
            <vgc:GenomicId>
              <vgc:IdType>EntrezGene</vgc:IdType>

          <vgc:Accession>10289</vgc:Accession>

          <vgc:DisplayName>EIF1B</vgc:DisplayName>
          </vgc:GenomicId>
          <vgc:GenomicId>
            <vgc:IdType>EMBL</vgc:IdType>

          <vgc:Accession>BC006996</vgc:Accession>

          <vgc:DisplayName>BC006996</vgc:DisplayName>
          </vgc:GenomicId>
          ...
          <vgc:GenomicId>
            <vgc:IdType>UniGene</vgc:IdType>

          <vgc:Accession>Hs.601063</vgc:Accession>

          <vgc:DisplayName>Hs.601063</vgc:DisplayName>
          </vgc:GenomicId>
          </vgc:GenomicIds>
```

```

        </vgc:Gene>
      </vgc:GeneByVariationIdResult>
    </vgc:GeneByVariationIdResults>
  </ns:return>
</ns:byVariationIdRegulatoryOutputIdTypesResponse>

```

## 5.6 Genes byVariationIdRegulatoryTranscript

There are two services for retrieving genes byVariationIdRegulatoryTranscript – a simple version (byVariationIdRegulatoryTranscript) and an extended version that also retrieves accession numbers from external databases (byVariationIdRegulatoryTranscriptOutputIdTypes).

These two services retrieve genes where the variations reside in post-transcriptional regulatory regions of the gene.

These services are described below in more detail.

### 5.6.1 Genes byVariationIdRegulatory

This web service retrieves genes where the variations reside in post-transcriptional regulatory regions of the gene. The variation identifier is the “rs number” of the variation.

URL:

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/gene/byVariationIdRegulatoryTranscript?id=<id\\_list>&firstResult=<first\\_result>&email=<email\\_address>](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/gene/byVariationIdRegulatoryTranscript?id=<id_list>&firstResult=<first_result>&email=<email_address>)*

Where:

- <id\_list> is a list of variation identifiers. Identifiers are separated by “+” symbols. For example: rs858528+rs2856753
- <first\_result> indicates the index of the first row to be returned. The web service will only return a set number of rows and the client is responsible for requesting additional results. The first row number starts with 0.
- <email\_address> is the email address of a registered GeneCruiser user.

#### 5.6.1.1 Examples

Searching for a gene with the single variation identifier:

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/gene/byVariationIdRegulatoryTranscript?id=rs6007619&firstResult=0&email=joe@example.com](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/gene/byVariationIdRegulatoryTranscript?id=rs6007619&firstResult=0&email=joe@example.com)*

This returns a single gene:

```

<ns:byVariationIdRegulatoryTranscriptResponse>
  <ns:return>
    <vgc:GeneByVariationIdResults>

```

```

    <vgc:GeneByVariationIdResult>
      <vgc:QueryParameter>

<vgc:VariationName>rs6007619</vgc:VariationName>
    </vgc:QueryParameter>
    <vgc:Gene>
      <vgc:GeneId>51955</vgc:GeneId>
      <vgc:Description>
        TBC1      domain      family      member      22A.
        [Source:Uniprot/SWISSPROT;Acc:Q8WUA7]
      </vgc:Description>
      <vgc:Source>ensembl</vgc:Source>
      <vgc:Biotype>protein_coding</vgc:Biotype>
      <vgc:Chromosome>22</vgc:Chromosome>
      <vgc:Start>45537193</vgc:Start>
      <vgc:End>45948393</vgc:End>
      <vgc:Strand>1</vgc:Strand>
      <vgc:StableId>ENSG00000054611</vgc:StableId>
    </vgc:Gene>
  </vgc:GeneByVariationIdResult>
</vgc:GeneByVariationIdResults>
</ns:return>
</ns:byVariationIdRegulatoryTranscriptResponse>

```

## 5.6.2 GenesByVariationIdRegulatoryOutputIdTypes

This web service retrieves genes where the variations reside in post-transcriptional regulatory regions of the gene. The identifier is the variation “rs number.”

This service also allows the caller to specify which external database accessions should be displayed. Note that retrieving the external accessions is costly, so unneeded accessions should not be retrieved for best performance.

URL:

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/gene/byVariationIdRegulatoryTranscriptOutputIdTypes?id=<id\\_list>&outputIdTypes=<output\\_id\\_types>&firstResult=<first\\_result>&email=<email\\_address>](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/gene/byVariationIdRegulatoryTranscriptOutputIdTypes?id=<id_list>&outputIdTypes=<output_id_types>&firstResult=<first_result>&email=<email_address>)*

Where:

- <id\_list> is a list of identifiers. Identifiers are separated by “+” symbols. For example: rs858528+rs2856753
- <output\_id\_types> is a list of output identifier types. The possible values for output\_id\_type can be retrieved using General/outputIdTypes service.
- <first\_result> indicates the index of the first row to be returned. The web service will only return a set number of rows and the client is responsible for requesting additional results. The first row number starts with 0.
- <email\_address> is the email address of a registered GeneCruiser user.

### 5.6.2.1 Examples

Searching for a gene with the single variation identifier:

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/gene/byVariationIdRegulatoryTranscriptOutputIdTypes?id=rs6007619&outputIdTypes=EntrezGene+UniGene+EMBL&firstResult=0&email=joe@example.com](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/gene/byVariationIdRegulatoryTranscriptOutputIdTypes?id=rs6007619&outputIdTypes=EntrezGene+UniGene+EMBL&firstResult=0&email=joe@example.com)*

This returns a single gene. Some of the EMBL accessions have been removed for brevity.

```
<ns:byVariationIdRegulatoryTranscriptOutputIdTypesResponse>
  <ns:return>
    <vgc:GeneByVariationIdResults>
      <vgc:GeneByVariationIdResult>
        <vgc:QueryParameter>

          <vgc:VariationName>rs6007619</vgc:VariationName>
          </vgc:QueryParameter>
          <vgc:Gene>
            <vgc:GeneId>51955</vgc:GeneId>
            <vgc:Description>
              TBC1      domain      family      member      22A.
              [Source:Uniprot/SWISSPROT;Acc:Q8WUA7]
            </vgc:Description>
            <vgc:Source>ensembl</vgc:Source>
            <vgc:Biotype>protein_coding</vgc:Biotype>
            <vgc:Chromosome>22</vgc:Chromosome>
            <vgc:Start>45537193</vgc:Start>
            <vgc:End>45948393</vgc:End>
            <vgc:Strand>1</vgc:Strand>
            <vgc:StableId>ENSG00000054611</vgc:StableId>
            <vgc:GenomicIds>
              <vgc:GenomicId>
                <vgc:IdType>EMBL</vgc:IdType>

                <vgc:Accession>BC001292</vgc:Accession>

                <vgc:DisplayName>BC001292</vgc:DisplayName>
                </vgc:GenomicId>
                <vgc:GenomicId>
                  <vgc:IdType>EMBL</vgc:IdType>

                <vgc:Accession>Z82187</vgc:Accession>

                <vgc:DisplayName>Z82187</vgc:DisplayName>
                </vgc:GenomicId>
                ...
                <vgc:GenomicId>
                  <vgc:IdType>EntrezGene</vgc:IdType>

                <vgc:Accession>25771</vgc:Accession>

                <vgc:DisplayName>TBC1D22A</vgc:DisplayName>
```

```
                </vgc:GenomicId>
            </vgc:GenomicIds>
        </vgc:Gene>
    </vgc:GeneByVariationIdResult>
</vgc:GeneByVariationIdResults>
</ns:return>
</ns:byVariationIdRegulatoryTranscriptOutputIdTypesResponse>
```

## 6 Variations

There are three ways to retrieve Variations (aka SNPs): by variation name, by genomic identifier or by genomic location. These services are described below in more detail.

### 6.1 Variations byName

This web service returns variations given the name (aka “rs number”) of the variation as input.

URL:

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/variation/byName?name=<name\\_list>&firstResult=0&email=joe@example.com](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/variation/byName?name=<name_list>&firstResult=0&email=joe@example.com)*

Where:

- <name\_list> is the type of genomic identifier. The possible values for id\_type can be retrieved using General/idTypes service.
- <first\_result> indicates the index of the first row to be returned. The web service will only return a set number of rows and the client is responsible for requesting additional results. The first row number starts with 0.
- <email\_address> is the email address of a registered GeneCruiser user.

#### 6.1.1 Examples

##### 6.1.1.1 Single Name

This example shows how to retrieve variations by name.

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/variation/byName?name=rs2856753&firstResult=0&email=joe@example.com](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/variation/byName?name=rs2856753&firstResult=0&email=joe@example.com)*

The output is shown below:

```
<ns:byNameResponse>
  <ns:return>
    <vgc:Variations>
      <vgc:Variation>
        <vgc:VariationName>rs2856753</vgc:VariationName>
        <vgc:Source>dbSNP</vgc:Source>
        <vgc:Allele>C/T</vgc:Allele>
        <vgc:ConsequenceType>INTRONIC</vgc:ConsequenceType>
        <vgc:Chromosome>17</vgc:Chromosome>
        <vgc:Start>7515730</vgc:Start>
        <vgc:End>7515730</vgc:End>
        <vgc:Strand>1</vgc:Strand>
      </vgc:Variation>
    </vgc:Variations>
```

```
</ns:return>
</ns:byNameResponse>
```

### 6.1.1.2 Multiple Names

This example shows how to retrieve variations by name, using multiple names as input.

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/variation/byName?name=rs2856753+rs858528&firstResult=0&email=joe@example.com](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/variation/byName?name=rs2856753+rs858528&firstResult=0&email=joe@example.com)*

The output is shown below:

```
<ns:byNameResponse>
  <ns:return>
    <vgc:Variations>
      <vgc:Variation>
        <vgc:VariationName>rs2856753</vgc:VariationName>
        <vgc:Source>dbSNP</vgc:Source>
        <vgc:Allele>C/T</vgc:Allele>
        <vgc:ConsequenceType>INTRONIC</vgc:ConsequenceType>
        <vgc:Chromosome>17</vgc:Chromosome>
        <vgc:Start>7515730</vgc:Start>
        <vgc:End>7515730</vgc:End>
        <vgc:Strand>1</vgc:Strand>
      </vgc:Variation>
      <vgc:Variation>
        <vgc:VariationName>rs858528</vgc:VariationName>
        <vgc:Source>dbSNP</vgc:Source>
        <vgc:Allele>C/T</vgc:Allele>
        <vgc:ConsequenceType>INTRONIC</vgc:ConsequenceType>
        <vgc:Chromosome>17</vgc:Chromosome>
        <vgc:Start>7515661</vgc:Start>
        <vgc:End>7515661</vgc:End>
        <vgc:Strand>1</vgc:Strand>
      </vgc:Variation>
    </vgc:Variations>
  </ns:return>
</ns:byNameResponse>
```

## 6.2 Variations byLocation

This web service returns the variations with a range of genomic locations. The genomic location range can be specified in a number of formats.

URL:

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/variation/byLocation?locations=<genomic\\_location\\_list>&<first\\_result>=0&email=<email\\_address>](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/variation/byLocation?locations=<genomic_location_list>&<first_result>=0&email=<email_address>)*

Where:

- <genomic\_location\_list> is a list of genomic location ranges. Ranges are separated by “+” symbols. For example: 6:1234-5678+7:1234-5678. See Appendix for more details.

- <first\_result> indicates the index of the first row to be returned. The web service will only return a set number of rows and the client is responsible for requesting additional results. The first row number starts with 0.
- <email\_address> is the email address of a registered GeneCruiser user.

## 6.2.1 Examples

Searching for Variations in the ranges 6:1234-5678 and 7:1234-5678.

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/variation/byLocation?locations=6:1234-5678+7:1234-5678&firstResult=0&email=joe@example.com](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/variation/byLocation?locations=6:1234-5678+7:1234-5678&firstResult=0&email=joe@example.com)*

```
<ns:byLocationResponse>
  <ns:return>
    <vgc:Variations>
      <vgc:Variation>
        <vgc:VariationName>rs1419824</vgc:VariationName>
        <vgc:Source>dbSNP</vgc:Source>
        <vgc:Allele>C/T</vgc:Allele>
        <vgc:ConsequenceType>INTERGENIC</vgc:ConsequenceType>
        <vgc:Chromosome>6</vgc:Chromosome>
        <vgc:Start>5658</vgc:Start>
        <vgc:End>5658</vgc:End>
        <vgc:Strand>1</vgc:Strand>
      </vgc:Variation>
      <vgc:Variation>
        <vgc:VariationName>rs2854679</vgc:VariationName>
        <vgc:Source>dbSNP</vgc:Source>
        <vgc:Allele>T/C</vgc:Allele>
        <vgc:ConsequenceType>INTERGENIC</vgc:ConsequenceType>
        <vgc:Chromosome>6</vgc:Chromosome>
        <vgc:Start>5597</vgc:Start>
        <vgc:End>5597</vgc:End>
        <vgc:Strand>-1</vgc:Strand>
      </vgc:Variation>
    </vgc:Variations>
  </ns:return>
</ns:byLocationResponse>
```

## 6.3 Variations byld

This web service returns the variations in a gene, given an id as input. This web service supports using gene, transcript, and translation ids. The id can be specified by accession number or by display name.

This web service returns variations in a gene, given an accession number as input. There are several forms of this web service that allow for searching in the gene itself, in the 3' flanking region, in the 5' flanking region, and combinations of the three.

### 6.3.1 Searching in the Gene

This web service searches for variations in the identified gene(s).

URL:

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/variation/byId?idType=<id\\_type>&id=<id\\_list>&firstResult=<first\\_result>&email=<email\\_address>](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/variation/byId?idType=<id_type>&id=<id_list>&firstResult=<first_result>&email=<email_address>)*

Where:

- <id\_type> is the type of genomic identifier. The possible values for id\_type can be retrieved using General/idTypes service.
- <id\_list> is a list of identifiers. Identifiers are separated by “+” symbols. For example: ENSG00000141510+ENSG00000067646
- <first\_result> indicates the index of the first row to be returned. The web service will only return a set number of rows and the client is responsible for requesting additional results. The first row number starts with 0.
- <email\_address> is the email address of a registered GeneCruiser user.

### 6.3.2 Searching in the 5' flanking region

This web service searches for variations flanking the gene(s) in the 5' direction.

URL:

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/variation/byIdFivePrime?idType=<id\\_type>&id=<id\\_list>&fivePrimeSize=<five\\_prime\\_window\\_size>&firstResult=<first\\_result>&email=<email\\_address>](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/variation/byIdFivePrime?idType=<id_type>&id=<id_list>&fivePrimeSize=<five_prime_window_size>&firstResult=<first_result>&email=<email_address>)*

Where:

- <id\_type> is the type of genomic identifier. The possible values for id\_type can be retrieved using General/idTypes service.
- <id\_list> is a list of identifiers. Identifiers are separated by “+” symbols. For example: ENSG00000141510+ENSG00000067646
- <five\_prime\_window\_size> is an integer number of base pairs to search in the 5' direction from the end of the gene.
- <first\_result> indicates the index of the first row to be returned. The web service will only return a set number of rows and the client is responsible for requesting additional results. The first row number starts with 0.
- <email\_address> is the email address of a registered GeneCruiser user.

### 6.3.3 Searching in the 3' flanking region

This web service searches for variations flanking the gene(s) in the 3' direction.

URL:

*http://genecruiser.broad.mit.edu/genecruiser3\_services/rest/variation/byIdThreePrime?idType=<id\_type>&id=<id\_list>&threePrimeSize=<three\_prime\_window\_size>&firstResult=<first\_result>&email=<email\_address>*

Where:

- <id\_type> is the type of genomic identifier. The possible values for id\_type can be retrieved using General/idTypes service.
- <id\_list> is a list of identifiers. Identifiers are separated by “+” symbols. For example: ENSG00000141510+ENSG00000067646
- <three\_prime\_window\_size> is an integer number of base pairs to search in the 3’ direction from the end of the gene.
- <first\_result> indicates the index of the first row to be returned. The web service will only return a set number of rows and the client is responsible for requesting additional results. The first row number starts with 0.
- <email\_address> is the email address of a registered GeneCruiser user.

#### **6.3.4 Searching in gene or in the 5’ flanking region**

This web service searches for variations in the gene or flanking the gene(s) in the 5’ direction.

URL:

*http://genecruiser.broad.mit.edu/genecruiser3\_services/rest/variation/byIdInGeneFivePrime?idType=<id\_type>&id=<id\_list>&fivePrimeSize=<five\_prime\_window\_size>&firstResult=<first\_result>&email=<email\_address>*

Where:

- <id\_type> is the type of genomic identifier. The possible values for id\_type can be retrieved using General/idTypes service.
- <id\_list> is a list of identifiers. Identifiers are separated by “+” symbols. For example: ENSG00000141510+ENSG00000067646
- <five\_prime\_window\_size> is an integer number of base pairs to search in the 5’ direction from the end of the gene.
- <first\_result> indicates the index of the first row to be returned. The web service will only return a set number of rows and the client is responsible for requesting additional results. The first row number starts with 0.
- <email\_address> is the email address of a registered GeneCruiser user.

### 6.3.5 Searching in gene or in the 3' flanking region

This web service searches for variations in the gene or flanking the gene(s) in the 3' direction.

URL:

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/variation/byIdThreePrime?idType=<id\\_type>&id=<id\\_list>&threePrimeSize=<three\\_prime\\_window\\_size>&firstResult=<first\\_result>&email=<email\\_address>](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/variation/byIdThreePrime?idType=<id_type>&id=<id_list>&threePrimeSize=<three_prime_window_size>&firstResult=<first_result>&email=<email_address>)*

Where:

- <id\_type> is the type of genomic identifier. The possible values for id\_type can be retrieved using General/idTypes service.
- <id\_list> is a list of identifiers. Identifiers are separated by “+” symbols. For example: ENSG00000141510+ENSG00000067646
- <three\_prime\_window\_size> is an integer number of base pairs to search in the 3' direction from the end of the gene.
- <first\_result> indicates the index of the first row to be returned. The web service will only return a set number of rows and the client is responsible for requesting additional results. The first row number starts with 0.
- <email\_address> is the email address of a registered GeneCruiser user.

### 6.3.6 Searching in gene or in the 5' or 3' flanking region

This web service searches for variations in the gene or flanking the gene(s) in the 5' or 3' direction.

URL:

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/variation/byIdFivePrimeThreePrime?idType=<id\\_type>&id=<id\\_list>&fivePrimeSize=<five\\_prime\\_window\\_size>&threePrimeSize=<three\\_prime\\_window\\_size>&firstResult=<first\\_result>&email=<email\\_address>](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/variation/byIdFivePrimeThreePrime?idType=<id_type>&id=<id_list>&fivePrimeSize=<five_prime_window_size>&threePrimeSize=<three_prime_window_size>&firstResult=<first_result>&email=<email_address>)*

Where:

- <id\_type> is the type of genomic identifier. The possible values for id\_type can be retrieved using General/idTypes service.
- <id\_list> is a list of identifiers. Identifiers are separated by “+” symbols. For example: ENSG00000141510+ENSG00000067646
- <five\_prime\_window\_size> is an integer number of base pairs to search in the 5' direction from the end of the gene.
- <three\_prime\_window\_size> is an integer number of base pairs to search in the 3' direction from the end of the gene.

- <first\_result> indicates the index of the first row to be returned. The web service will only return a set number of rows and the client is responsible for requesting additional results. The first row number starts with 0.
- <email\_address> is the email address of a registered GeneCruiser user.

## 6.3.7 Examples

### 6.3.7.1 Single Id

This example shows how to retrieve the SNPs in a gene, using the Ensemble Gene Stable Id to identify the gene.

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/variation/byGenomicId?idType=ensembl\\_gene\\_stable\\_id&id=ENSG00000141510&firstResult=0&email=joe@example.com](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/variation/byGenomicId?idType=ensembl_gene_stable_id&id=ENSG00000141510&firstResult=0&email=joe@example.com)*

A portion of the output is show below. Some of the VariationQueryResult objects are not shown for brevity.

```
<ns:byGenomicIdResponse>
  <ns:return>
    <vgc:VariationQueryResults>
      <vgc:VariationQueryResult>
        <vgc:QueryParameter>
          <vgc:IdType>Ensembl Gene Stable Id</vgc:IdType>
          <vgc:Id>ENSG00000141510</vgc:Id>

        <vgc:IdDisplayName>ENSG00000141510</vgc:IdDisplayName>
        </vgc:QueryParameter>
        <vgc:Variation>

          <vgc:VariationName>rs1794293</vgc:VariationName>
          <vgc:Source>dbSNP</vgc:Source>
          <vgc:Allele>G/C</vgc:Allele>

          <vgc:ConsequenceType>3PRIME_UTR</vgc:ConsequenceType>
          <vgc:Chromosome>17</vgc:Chromosome>
          <vgc:Start>7512595</vgc:Start>
          <vgc:End>7512595</vgc:End>
          <vgc:Strand>1</vgc:Strand>
        </vgc:Variation>
      </vgc:VariationQueryResult>
      <vgc:VariationQueryResult>
        <vgc:QueryParameter>
          <vgc:IdType>Ensembl Gene Stable Id</vgc:IdType>
          <vgc:Id>ENSG00000141510</vgc:Id>

        <vgc:IdDisplayName>ENSG00000141510</vgc:IdDisplayName>
        </vgc:QueryParameter>
        <vgc:Variation>
```

```

<vgc:VariationName>rs1794292</vgc:VariationName>
      <vgc:Source>dbSNP</vgc:Source>
      <vgc:Allele>G/C</vgc:Allele>

<vgc:ConsequenceType>3PRIME_UTR</vgc:ConsequenceType>
      <vgc:Chromosome>17</vgc:Chromosome>
      <vgc:Start>7512596</vgc:Start>
      <vgc:End>7512596</vgc:End>
      <vgc:Strand>1</vgc:Strand>
    </vgc:Variation>
  </vgc:VariationQueryResult>
  ...
</vgc:VariationQueryResults>
</ns:return>
</ns:byGenomicIdResponse>

```

### 6.3.7.2 Multiple ids

This example shows how to retrieve the SNPs in two genes, using the Ensemble Gene Stable Id to identify the genes.

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/variation/byGenomicId?idType=embl\\_gene\\_stable\\_id&id=ENSG00000141510+ENSG00000067646&firstResult=0&email=joe@example.com](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/variation/byGenomicId?idType=embl_gene_stable_id&id=ENSG00000141510+ENSG00000067646&firstResult=0&email=joe@example.com)*

A portion of the output is show below; some of the VariationQueryResult objects are not shown for brevity.

```

<ns:byGenomicIdResponse xmlns:ns="http://service.genecruiser.org/xsd">
  <ns:return>
    <vgc:VariationQueryResults xmlns:vgc="service.genecruiser.org">
      <vgc:VariationQueryResult>
        <vgc:QueryParameter>
          <vgc:IdType>Ensembl Gene Stable Id</vgc:IdType>
          <vgc:Id>ENSG00000067646</vgc:Id>
          <vgc:IdDisplayName>ENSG00000067646</vgc:IdDisplayName>
        </vgc:QueryParameter>
        <vgc:Variation>
          <vgc:VariationName>rs1270119</vgc:VariationName>
          <vgc:Source>dbSNP</vgc:Source>
          <vgc:Allele>T/C</vgc:Allele>
          <vgc:ConsequenceType>5PRIME_UTR</vgc:ConsequenceType>
          <vgc:Chromosome>Y</vgc:Chromosome>
          <vgc:Start>2863717</vgc:Start>
          <vgc:End>2863717</vgc:End>
          <vgc:Strand>1</vgc:Strand>
        </vgc:Variation>
      </vgc:VariationQueryResult>
      <vgc:VariationQueryResult>
        <vgc:QueryParameter>
          <vgc:IdType>Ensembl Gene Stable Id</vgc:IdType>
          <vgc:Id>ENSG00000067646</vgc:Id>

```

```

        <vgc:IdDisplayName>ENSG00000067646</vgc:IdDisplayName>
    </vgc:QueryParameter>
    <vgc:Variation>
        <vgc:VariationName>rs13304839</vgc:VariationName>
        <vgc:Source>dbSNP</vgc:Source>
        <vgc:Allele>C/T</vgc:Allele>
        <vgc:ConsequenceType>INTRONIC</vgc:ConsequenceType>
        <vgc:Chromosome>Y</vgc:Chromosome>
        <vgc:Start>2866136</vgc:Start>
        <vgc:End>2866136</vgc:End>
        <vgc:Strand>1</vgc:Strand>
    </vgc:Variation>
</vgc:VariationQueryResult>

</vgc:VariationQueryResults>
</ns:return>
</ns:byidResponse>

```

### 6.3.7.3 Empty Result

A query that does not find any matching variations will return an empty `VariationQueryResult` tag:

```

<ns:byGenomicIdResponse xmlns:ns="http://service.genecruiser.org/xsd">
    <ns:return>
        <vgc:VariationQueryResults xmlns:vgc="service.genecruiser.org"/>
    </ns:return>
</ns:byGenomicIdResponse>

```

### 6.3.7.4 Accession Numbers versus Display Names.

The service allows you to use either accession numbers as input or display names. Here's an example using a display name.

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/variation/byGenomicId?idType=HGNC\\_GENE&id=brca1&firstResult=0&email=joe@example.com](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/variation/byGenomicId?idType=HGNC_GENE&id=brca1&firstResult=0&email=joe@example.com)*

A portion of the result file is shown below.

```

<ns:byGenomicIdResponse xmlns:ns="http://service.genecruiser.org/xsd">
    <ns:return>
        <vgc:VariationQueryResults xmlns:vgc="service.genecruiser.org">
            <vgc:VariationQueryResult>
                <vgc:QueryParameter>
                    <vgc:IdType>HGNC Symbol (HUGO)</vgc:IdType>
                    <vgc:Id>1100</vgc:Id>
                    <vgc:IdDisplayName>BRCA1</vgc:IdDisplayName>
                </vgc:QueryParameter>
                <vgc:Variation>
                    <vgc:VariationName>rs8176320</vgc:VariationName>
                    <vgc:Source>dbSNP</vgc:Source>
                </vgc:Variation>
            </vgc:VariationQueryResult>
        </vgc:VariationQueryResults>
    </ns:return>
</ns:byGenomicIdResponse>

```

```

        <vgc:Allele>C/T</vgc:Allele>
        <vgc:ConsequenceType>3PRIME_UTR</vgc:ConsequenceType>
        <vgc:Chromosome>17</vgc:Chromosome>
        <vgc:Start>38449889</vgc:Start>
        <vgc:End>38449889</vgc:End>
        <vgc:Strand>1</vgc:Strand>
      </vgc:Variation>
    </vgc:VariationQueryResult>
  </vgc:VariationQueryResults>
</ns:return>
</ns:byGenomicIdResponse>

```

The same query can be performed using the HGNC Gene Symbol:

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/variation/byGenomicId?idType=HGNC\\_GENE&id=1100&firstResult=0&email=joe@example.com](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/variation/byGenomicId?idType=HGNC_GENE&id=1100&firstResult=0&email=joe@example.com)*

### 6.3.8 Errors

If the web service encounters an exception, it will wrap the exception in XML and return it. Here are some example errors:

In this URL, the `first_result` parameter is invalid:

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/variation/byGenomicId?idType=HGNC\\_GENE&id=1100&firstResult=foo&email=joe@example.com](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/variation/byGenomicId?idType=HGNC_GENE&id=1100&firstResult=foo&email=joe@example.com)*

The result would be:

```

<ns:byGenomicIdResponse xmlns:ns="http://service.genecruiser.org/xsd">
  <ns:return>
    <vgc:Exception xmlns:vgc="service.genecruiser.org">
      <vgc:ExceptionType>java.lang.NumberFormatException</vgc:ExceptionType>
      <vgc:Message>For input string: "foo"</vgc:Message>
      <vgc:ServiceClass>org.genecruiser.service.VariationService</vgc:ServiceClass>
      <vgc:MethodName> byGenomicIdImpl</vgc:MethodName>
    </vgc:Exception>
  </ns:return>
</ns:byGenomicIdResponse>

```

In this URL, the `id_type` is invalid:

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/variation/byGenomicId?idType=foo&id=1100&firstResult=0&email=joe@example.com](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/variation/byGenomicId?idType=foo&id=1100&firstResult=0&email=joe@example.com)*

The result would be:

```

<ns:byGenomicIdResponse>
  <ns:return>

```

```

        <vgc:Exception>
            <vgc:ExceptionType>

org.genecruiser.persistence.InvalidGenomicIdentifierException
            </vgc:ExceptionType>
            <vgc:Message>InvalidGenomicIdentifier: foo</vgc:Message>

        <vgc:ServiceClass>org.genecruiser.service.VariationService</vgc:Service
Class>
            <vgc:MethodName> byGenomicIdImpl</vgc:MethodName>
        </vgc:Exception>
    </ns:return>
</ns:byGenomicIdResponse>

```

In this URL, the email address is unknown:

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/variation/byGenomicId?idType=ensembl\\_gene\\_stable\\_id&id=ENSG00000141510&firstResult=0&email=foo](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/variation/byGenomicId?idType=ensembl_gene_stable_id&id=ENSG00000141510&firstResult=0&email=foo)*

```

<ns:byGenomicIdResponse>
    <ns:return>
        <vgc:Exception>

            <vgc:ExceptionType>java.lang.IllegalArgumentException</vgc:ExceptionType>
e>
                <vgc:Message>Unknown email address</vgc:Message>

            <vgc:ServiceClass>org.genecruiser.service.VariationService</vgc:Service
Class>
                <vgc:MethodName>byGenomicIdHelper</vgc:MethodName>
            </vgc:Exception>
        </ns:return>
    </ns:byGenomicIdResponse>

```

## 6.4 Variations byIdRegulatory

This web service returns the variations in the regulatory region of a gene, given an id as input. This web service supports using gene, transcript, and translation ids. The id can be specified by accession number or by display name.

### 6.4.1 Syntax

This web service searches for variations in the identified gene(s).

URL:

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/variation/byIdRegulatory?idType=<id\\_type>&id=<id\\_list>&firstResult=<first\\_result>&email=<email\\_address>](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/variation/byIdRegulatory?idType=<id_type>&id=<id_list>&firstResult=<first_result>&email=<email_address>)*

Where:

- <id\_type> is the type of genomic identifier. The possible values for id\_type can be retrieved using General/idTypes service.

- <id\_list> is a list of identifiers. Identifiers are separated by “+” symbols. For example: ENSG00000141510+ENSG00000067646
- <first\_result> indicates the index of the first row to be returned. The web service will only return a set number of rows and the client is responsible for requesting additional results. The first row number starts with 0.
- <email\_address> is the email address of a registered GeneCruiser user.

## 6.4.2 Example

This example shows how to retrieve variations in the regulatory region of a gene.

*http://genecruiser.broad.mit.edu/genecruiser3\_services/rest/variation/byGenomicIdRegulatory?idType=ensembl\_gene\_stable\_id&id=ENSG00000114784&firstResult=0&email=joe@example.com*

The output is shown below:

```
<ns:byGenomicIdRegulatoryResponse>
  <ns:return>
    <vgc:VariationQueryResults>
      <vgc:VariationQueryResult>
        <vgc:QueryParameter>
          <vgc:IdType>Ensembl Gene Stable Id</vgc:IdType>
          <vgc:Id>ENSG00000114784</vgc:Id>

        <vgc:IdDisplayName>ENSG00000114784</vgc:IdDisplayName>
        </vgc:QueryParameter>
        <vgc:Variation>

          <vgc:VariationName>rs5004340</vgc:VariationName>
          <vgc:Source>dbSNP</vgc:Source>
          <vgc:Allele>C/G</vgc:Allele>

          <vgc:ConsequenceType>REGULATORY_REGION,UPSTREAM</vgc:ConsequenceType>
          <vgc:Chromosome>3</vgc:Chromosome>
          <vgc:Start>40326144</vgc:Start>
          <vgc:End>40326144</vgc:End>
          <vgc:Strand>1</vgc:Strand>
        </vgc:Variation>
      </vgc:VariationQueryResult>
    </vgc:VariationQueryResults>
  </ns:return>
</ns:byGenomicIdRegulatoryResponse>
```

## 6.5 Variations byIdRegulatoryTranscript

This web service retrieves variations that reside in the post-transcriptional regulatory regions of the gene.

This web service supports using gene, transcript, and translation ids. The id can be specified by accession number or by display name.

### 6.5.1 Syntax

This web service searches for variations in the identified gene(s).

URL:

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/variation/byIdRegulatoryTrnascript?idType=<id\\_type>&id=<id\\_list>&firstResult=<first\\_result>&email=<email\\_address>](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/variation/byIdRegulatoryTrnascript?idType=<id_type>&id=<id_list>&firstResult=<first_result>&email=<email_address>)*

Where:

- <id\_type> is the type of genomic identifier. The possible values for id\_type can be retrieved using General/idTypes service.
- <id\_list> is a list of identifiers. Identifiers are separated by “+” symbols. For example: ENSG00000141510+ENSG00000067646
- <first\_result> indicates the index of the first row to be returned. The web service will only return a set number of rows and the client is responsible for requesting additional results. The first row number starts with 0.
- <email\_address> is the email address of a registered GeneCruiser user.

### 6.5.2 Example

This example shows how to retrieve variations in the post-transcriptional regulatory regions of a gene.

*[http://genecruiser.broad.mit.edu/genecruiser3\\_services/rest/variation/byGenomicIdRegulatoryTrnascript?idType=ensembl\\_gene\\_stable\\_id&id=ENSG00000054611&firstResult=0&email=joe@example.com](http://genecruiser.broad.mit.edu/genecruiser3_services/rest/variation/byGenomicIdRegulatoryTrnascript?idType=ensembl_gene_stable_id&id=ENSG00000054611&firstResult=0&email=joe@example.com)*

The output is shown below:

```
<ns:byGenomicIdRegulatoryTranscriptResponse>
  <ns:return>
    <vgc:VariationQueryResults>
      <vgc:VariationQueryResult>
        <vgc:QueryParameter>
          <vgc:IdType>Ensembl Gene Stable Id</vgc:IdType>
          <vgc:Id>ENSG00000054611</vgc:Id>

          <vgc:IdDisplayName>ENSG00000054611</vgc:IdDisplayName>
        </vgc:QueryParameter>
        <vgc:Variation>

          <vgc:VariationName>rs6007619</vgc:VariationName>
          <vgc:Source>dbSNP</vgc:Source>
          <vgc:Allele>G/C</vgc:Allele>
```

```
<vgc:ConsequenceType>
  REGULATORY_REGION, 3PRIME_UTR
</vgc:ConsequenceType>
<vgc:Chromosome>22</vgc:Chromosome>
<vgc:Start>45948235</vgc:Start>
<vgc:End>45948235</vgc:End>
<vgc:Strand>1</vgc:Strand>
</vgc:Variation>
</vgc:VariationQueryResult>
</vgc:VariationQueryResults>
</ns:return>
</ns:byGenomicIdRegulatoryTranscriptResponse>
```

## 7 Appendix Parameter Details

This appendix contains detailed descriptions for the complex web service parameters.

### 7.1 id\_types and output\_id\_types

We do not anymore provide a list of available id\_types and output\_id\_types. List of available identifiers can be obtained dynamically using general/idTypes and general/outputIdTypes services.

### 7.2 genomic\_location\_list

The Variation byLocation web service takes a list of genomic location ranges and searches for variations within those ranges. These ranges can be specified in several ways, as described below. The ranges are delimited by “+” on the URL.

#### 7.2.1 Chromosome, Start and End

You can specify a range using a chromosome, start and end. For example:

```
chr6:1234-5678
```

This specifies a range on chromosome 6, starting at position 1234 and ending at 5678. The “chr” prefix is optional. The previous range can also be specified as:

```
6:1234-5678
```

### 7.3 Chromosome, Start, End and Strand

You can specify a range using a chromosome, start, end, and strand. For example:

```
chr6:1234-5678:-1
```

or:

```
chr6:1234-5678:1
```

Note that the following is not acceptable to the web service (although the web application user interface allows it):

```
chr6:1234-5678:+
```

### 7.4 Chromosome, Start and Strand

You can specify a range using a chromosome, start and a strand. For example:

```
chr6:1234:1
```

Note that the following is not acceptable to the web service (although the web application user interface allows it):

```
chr6:1234:+
```

## **7.5 Chromosome**

You can specify a range using only a chromosome. For example:

```
chr6
```

## **7.6 Chromosome and Strand.**

You can specify a range using only a chromosome and a strand. For example:

```
chr6::1
```

Note that the following is not acceptable to the web service (although the web application user interface allows it):

```
chr6::+
```