# SNPdat: rapid annotation of novel SNPs for model and non-model organisms

Anthony G Doran<sup>1</sup> and Christopher J Creevey<sup>1\*</sup>

<sup>1</sup> Teagasc Animal and Bioscience Research Department, Animal & Grassland Research and Innovation Centre, Teagasc, Grange, Dunsany, Co. Meath, Ireland.Contact details and corresponding author

\*corresponding author: <a href="mailto:chris.creevey@teagasc.ie">chris.creevey@teagasc.ie</a> and anthony.doran@teagasc.ie

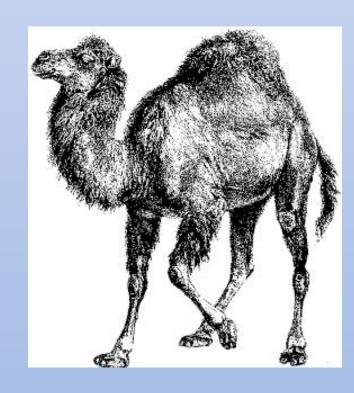
### What is SNPdat?

**SNPdat** (**SNP D**ata **A**nalysis **T**ool) is a command line tool for rapid annotation of both novel and known single nucleotide polymorphisms (SNPs) for any organism with a reference sequence and annotation (even those in draft status)

### **Before running SNPdat?**

SNPdat can be run on any machine that has a recent version of Perl installed.

**Perl** is a high-level, generalpurpose, interpreted, dynamic programming language. Current versions of Perl are available from <a href="https://www.perl.org">www.perl.org</a>



### Why use SNPdat?

### **SNPdat**

- •is an easy to use command-line tool for annotating the results of *de novo* SNP discovery projects.
- •is specifically designed for use with organisms which are either not supported by other tools or have a small number of annotated SNPs available
- •can also be used to analyse datasets from organisms which are densely sampled for SNPs.
- •is a high throughput analysis tool that can provide a comprehensive annotation of both novel and known SNPs for any organism with a draft sequence and annotation.

SNPdat will be of most use to researchers involved in de novo SNP discovery studies for organisms with poor (or non at all) coverage of SNPs in databases such as dbSNP

### Where to get SNPdat?

SNPdat is freely available from:

http://code.google.com/p/snpdat

You will also find sample files, further information on SNPdat and a manual here.



SNPdat - A Simple High Throughput Analysis Tool for Annotating SNPs

**Project Home** 

Downloads

Summary People

#### Project Information

₹ Recommend this on Google

Project feeds

Code license GNU GPL v2

#### Labels

SingleNucleotidePolymorphism, Annotation, SNP, SNPanalysis, gtf, gff, fasta, sequenceanalysis

🐸 Members

d1an...@gmail.com, anthony....@teagasc.ie, chris.cr...@gmail.com SNPdat (SNP Data Analysis Tool) is a high throughput analysis tool that can provide a comprehensive annotation of both novel and known single nucleotide polymorphisms (SNPs). It is specifically designed for use with organisms which are either not supported by other tools or have a small number of annotated SNPs available, however it can also be used to analyse datasets from organisms which are densely sampled for SNPs. It can be used for analysis of any organism with a draft sequence and annotation. SNPdat makes possible analyses involving non-model organisms that are not supported by the vast majority of SNP annotation tools currently available.

You can easily download the software, manual, sample dataset and short tutorial on SNPdat using the downloads tab on the left (<a href="http://code.google.com/p/snpdat/downloads/">http://code.google.com/p/snpdat/downloads/</a>). Additional scripts and a sample input file for SNPdat are available on the downloads page.

#### Table of Contents

- How to use SNPdat
  - Some Prerequisites
    - 1. Perl
  - Mandatory Input files:
    - 1. User input file
    - 2. Gene annotation file (GTF)
    - 3. FASTA sequence file
    - Additional script for retrieving FASTA and GTF files
  - · Optional Input File:
    - Additional script for retrieving FLAT files from dbSNP
    - Preparing the optional input file
  - · Output file:
    - Output file format
- Running SNPdat
- Further instructions
- Authors and Contact

### Input files

## **SNP**dat requires three mandatory input files:

- 1. User input file
- 2. Gene annotation file (GTF)
- 3. FASTA sequence file

### And one optional input file:

4. A processed FLAT file

### User input file

SNPdat accepts two file formats as user input.

This file must be specified on the command line using the -i switch when using SNPdat. This will be explained in greater detail later.

The first user input file format accepted by SNPdat is a simple tab-delimited text file.
 This file should not have any header information. Any line beginning with # will be skipped.
 This file should contain three columns:

```
chromosome_id position mutation e.g. tab-delmited text file
```

| 25 | 1234568 | Α |
|----|---------|---|
| X  | 1234568 | Т |
| 19 | 1234568 | G |
| 1  | 1234568 | C |

The chromosome IDs used in the input file must be the same format in ALL input files

### User input file

The second user input file format accepted is a Variant Calling Format (VCF) file.
 This file should be tab-delimited. This file must have a header line that begins with ##fileformat=vcf.
 Any line after the first that begins with # will be skipped. This file should have as its first five columns:

```
Chromosome_id position snp_id ref_base mutation
```

Any column after the first five will be ignored by SNPdat

e.g. VCF file

The chromosome IDs used in the input file must be the same format in ALL input files

### **FASTA** format sequence File

The second file is a DNA FASTA format sequence file for the species of interest. FASTA format is a text based format that is standard for storing sequence information.

FASTA files can be downloaded from Ensmbl at: <a href="http://www.ensembl.org/info/data/ftp/index.html">http://www.ensembl.org/info/data/ftp/index.html</a>

Or using GTF\_FASTA\_finder.pl

This file must be specified on the command line using the -f switch when using SNPdat. This will be explained in greater detail later.

The chromosome IDs used in the FASTA file must be the same format in ALL input files

>25
TGATCGATAAGATGTCCCAGCAGGGATTCTCTCTCTGGGCAGGTGAGGTCTTCTCTTAAA
GAAACCCCATATATAAGGGATTTTTAGGCCTTTTTTGGATCAAAATTATCCCAGTTTTTCA
AGATATTATTCAAGGGGCGTGGTTCTGGACTGTTAGGCCCCATCTGTAAGAGAGAAAAA
GCAGTGCCCAGTGCCATGGCTTTTTTCTACCAGAGGTGTCTGCCCCTGCTCTAGACGGGG
TTGGAGACTGATTCTCCAACTAAGCTTCCTTCCCTGGCCAGAGCTAACCTGTGTCTTACA
GGTGAAGGTGTCCTACTCCCAACCCCCCTCGTTCTACCACCCCAGGTATGGTGGAGATGCA
CCAGGGTTAGACCTGGTGGCATCCCAGATTGATGTCCAGGTCCTCACCATTAAAACCTTG
GTTTGATTTCCCTTTTTATTCCGGCACCACCTGGGGTGGGACAGAGTAGTTTTCCGGATGC
TTGCTGAGCCAAGACCACTAGGGGAAGCACCCCAACTTCTGTGTACCTGTGCACATTCCTC

### **Gene Transfer File**

The final mandatory input file is a gene annotation file. This file should be in gene transfer format (GTF). GTF files are a standard format for storing information on gene structure. Further information can be found here: <a href="http://genome.ucsc.edu/FAQ/FAQformat.html#format4">http://genome.ucsc.edu/FAQ/FAQformat.html#format4</a>.

GTF files can be downloaded from Ensembl at: <a href="http://www.ensembl.org/info/data/ftp/index.html">http://www.ensembl.org/info/data/ftp/index.html</a>

Or using GTF\_FASTA\_finder.pl

This file must be specified on the command line using the -g switch when using SNPdat. This will be explained in greater detail later.

The chromosome IDs used in the GTF file must be the same format in ALL input files

### **Ensembl Downloads Page**

http://www.ensembl.org/info/data/ftp/index.html



If you want to filter or customise your download, please try

Custom data sets

Biomart, a web-based querying tool.

Login · Registe

A > Help & Documentation

FTP Download

#### **FTP Download**

#### **API Code**

If you do not have access to CVS, you can obtain our latest API code as a gzipped tarball:

Download complete API for this release

Note: the API version needs to be the same as the databases you are accessing, so please use CVS to obtain a previous version if querying older databases.

#### Database dumps

Entire databases can be downloaded from our FTP site in a variety of formats. Please be aware that some of these files can run to many gigabytes of data.

Looking for MySQL dumps to install databases locally? See our web installation instructions for full details.

Each directory on ftp.ensembl.org contains a README file, explaining the directory structure.

#### Multi-species data

| Database             |       |            |            |     |                   |
|----------------------|-------|------------|------------|-----|-------------------|
| Comparative genomics | MySQL | <u>EMF</u> | <u>BED</u> | XML | Ancestral Alleles |
| BioMart              | MySQL | -          | -          | -   | -                 |

#### Single species data

novemcinctus

|  | $\sim$         |                 |                  |                                |                                 |                                    |            |                    |                    |                    |                    |                     |               |            |
|--|----------------|-----------------|------------------|--------------------------------|---------------------------------|------------------------------------|------------|--------------------|--------------------|--------------------|--------------------|---------------------|---------------|------------|
| Show All 🕶 entri   | es /           |                 |                  |                                |                                 | Show/hi                            | de/column  | s                  |                    |                    |                    | Filter              |               |            |
| Species  | DNA<br>(FASTA) | cDNA<br>(FASTA) | ncRNA<br>(FASTA) | Protein<br>sequence<br>(FASTA) | Annotated<br>sequence<br>(EMBL) | Annotated<br>sequence<br>(GenBank) | Gene sets  | Whole<br>databases | Variation<br>(EMF) | Variation<br>(GVF) | Variation<br>(VEP) | Regulation<br>(GFF) | Data<br>files | BAM        |
| Ailuropoda<br>melanoleuca<br>(Panda)                     | FASTA          | <u>FASTA</u>    | <u>FASTA</u>     | FASTA                          | EMBL                            | <u>GenBank</u>                     | <u>GTF</u> | <u>MySQL</u>       | -                  | -                  | -                  | -                   | -             | -          |
| Anolis<br>carolinensis<br>(Anole lizard)                 | <u>FASTA</u>   | <u>FASTA</u>    | <u>FASTA</u>     | <u>FASTA</u>                   | EMBL                            | <u>GenBank</u>                     | <u>GTF</u> | MySQL              | -                  | -                  | -                  | -                   | -             | -          |
| Bos taurus (Cow)   | <u>FASTA</u>   | <u>FASTA</u>    | <u>FASTA</u>     | <u>FASTA</u>                   | EMBL                            | <u>GenBank</u>                     | <u>GTF</u> | MySQL              | -                  | <u>GVF</u>         | <u>VEP</u>         | -                   | -             | -          |
| Caenorhabditis<br>elegans<br>(Caenorhabditis<br>elegans) | <u>FASTA</u>   | FASTA           | FASTA            | <u>FASTA</u>                   | EMBL                            | <u>GenBank</u>                     | GTF        | MySQL              | -                  | -                  | -                  | -                   | -             | -          |
| Callithrix<br>jacchus<br>(Marmoset)                      | <u>FASTA</u>   | FASTA           | <u>FASTA</u>     | FASTA                          | EMBL                            | <u>GenBank</u>                     | <u>GTF</u> | MySQL              | -                  | -                  | -                  | -                   | -             | -          |
| Canis familiaris<br>(Dog)                                | <u>FASTA</u>   | <u>FASTA</u>    | <u>FASTA</u>     | <u>FASTA</u>                   | EMBL                            | <u>GenBank</u>                     | <u>GTF</u> | MySQL              | -                  | <u>GVF</u>         | -                  | -                   | -             | <u>BAM</u> |
| Cavia porcellus<br>(Guinea Pig)                          | <u>FASTA</u>   | <u>FASTA</u>    | <u>FASTA</u>     | <u>FASTA</u>                   | EMBL                            | <u>GenBank</u>                     | <u>GTF</u> | MySQL              | -                  | -                  | -                  | -                   | -             | -          |
| Choloepus<br>hoffmanni (Sloth)                           | <u>FASTA</u>   | <u>FASTA</u>    | <u>FASTA</u>     | <u>FASTA</u>                   | EMBL                            | <u>GenBank</u>                     | <u>GTF</u> | <u>MySQL</u>       | -                  | -                  | -                  | -                   | -             | -          |
| Ciona intestinalis<br>(C.intestinalis)                   | <u>FASTA</u>   | <u>FASTA</u>    | <u>FASTA</u>     | <u>FASTA</u>                   | EMBL                            | <u>GenBank</u>                     | <u>GTF</u> | <u>MySQL</u>       | -                  | -                  | -                  | -                   | -             | -          |
| Ciona savignyi<br>(C.savignyi)                           | <u>FASTA</u>   | <u>FASTA</u>    | <u>FASTA</u>     | <u>FASTA</u>                   | EMBL                            | <u>GenBank</u>                     | <u>GTF</u> | <u>MySQL</u>       | -                  | -                  | -                  | -                   | -             | -          |
| <b>Danio rerio</b><br>(Zebrafish)                        | FASTA          | <u>FASTA</u>    | <u>FASTA</u>     | <u>FASTA</u>                   | EMBL                            | <u>GenBank</u>                     | GTE        | MySQL              | -                  | <u>GVF</u>         | VEP                | -                   | -             | -          |
| Dasypus  | FASTA          | <u>FASTA</u>    | <u>FASTA</u>     | <u>FASTA</u>                   | EMBL                            | <u>GenBank</u>                     | GTF /      | MySQL              | -                  | -                  | -                  | -                   | -             | -          |

### GTF\_FASTA\_finder.pl

This is an interactive script designed to retrieve FASTA (dna) and GTF files from Ensembl. You will need to be connected to the internet to
use this script. This is written in Perl but uses the system call cURL to retrieve the information from Ensembl. cURL is a part of most Linux
distributions and Mac OS X and can also be provided for windows through cygwin, which is a collection of tools that provide a Linux-like
environment for windows. To run this script simply type 'perl GTF FASTA finder.pl' into your terminal and follow the prompts.

#### e.g.

```
user@server $HOME/directory/with_all/SNPdat/files/
$ perl GTF_FASTA_finder.pl
36 /pub/release-56
                      |37| /pub/release-57
                                             |38| /pub/release-58
39| /pub/release-59
                      |40| /pub/release-60
                                             |41| /pub/release-61
42 /pub/release-62
                      |43| /pub/release-63
                                             |44| /pub/release-64
45 /pub/release-65
                                             |47| /pub/release-67
                      |46| /pub/release-66
Please select a release to choose from by typing its number:
You chose release-67
0 ailuropoda_melanoleuca
   ancestral_alleles
   anolis_carolinensis
3 bos_taurus
|56| tursiops_truncatus
|57| vicugna_pacos
|58| xenopus_tropicalis
Please select an organism (enter the corresponding number) to retrieve the FASTA file for:
You chose organism Saccharomyces_cerevisiae
Now retrieving the relevant FASTA file for this organism
|O| ailuropoda_melanoleuca
|1| anolis_carolinensis
|2| bos_taurus
|55| tursiops_truncatus
|56| vicugna_pacos
|57| xenopus_tropicalis
Please select an organism (enter the corresponding number) to retrieve the GTF file for:
Now retrieving the selected GTF file
Retrieving GTF: Saccharomyces_cerevisiae.EF4.67.gtf.gz
FASTA information has been retrieved for saccharomyces_cerevisiae from release release-67 of ensembl
GTF information has been retrieved for saccharomyces_cerevisiae from release release-67 of ensembl
.gtf.gz and .fa.gz files need be unzipped using the command 'gzip -d filename'
If you have any queries regarding SNPdat or the additional scripts please consult the website:
http://code.google.com/p/snpdat/
```

### **Optional Input file**

SNPdat has one optional input file. This file is a processed dbSNP FLAT file. This file can be used to identify SNPs that already have annotations in dbSNP. ASN\_FLAT files can be downloaded from <a href="ftp://ftp.ncbi.nih.gov/snp/organisms/">ftp://ftp.ncbi.nih.gov/snp/organisms/</a>.

Or using dbSNP\_finder.pl

This FLAT file must first be processed using the SNPdat\_parse\_dbsnp.pl script. This will create a file in the input format required for SNPdat. Scripts for processing FLAT files from other databases are available from the authors on request.

Once this file has been processed it can supplied as an input file for SNPdat using the –d switch. This will be explained in greater detail later.

| 10<br>10<br>10<br>10<br>10<br>10<br>4<br>4<br>4<br>4<br>25<br>25<br>25 | 86180529<br>86180332<br>37342771<br>83618673<br>83618951<br>83619862<br>83619857<br>71663812<br>71663942<br>71664184<br>21649618<br>21649613<br>14097644<br>21649549 | rs17870235<br>rs17870236<br>rs17870237<br>rs17870252<br>rs17870253<br>rs17870254<br>rs17870255<br>rs17870258<br>rs17870259<br>rs17870260<br>rs42767860<br>rs42767861<br>rs43615187<br>rs43639289 |
|--|--|--|
| 23   | 21049349   | 1 34 30 39 2 6 9   |

### dbSNP\_finder.pl

#### Additional script for retrieving FLAT files from dbSNP

you can use 'dbSNP\_finder.pl' to get dbSNP flat files for organisms contained in ftp://ftp.ncbi.nih.gov/snp/organisms/

#### e.g.

```
user@server $HOME/directory/with_all/SNPdat/files/
$ perl dbSNP_finder.pl
 Alectoris_9077/
 Bos_29061/
 almond_3755/
|109|
  _zebrafish_7955/
  _zebu_9915/
|111| zostera_29655/
Please select and organism by typing its corresponding number
You have chosen: arabidopsis_3702/
Retrieving file: 'ds_flat_ch2.flat.gz'
Retrieving file: 'ds_flat_ch4.flat.gz'
Now you will need to unzip these files.
This can be done using gzip -d
e.g.
gzip -d ds_*.gz
If you wish to use these (unzipped) files for SNPdat please run 'SNPdat_parse_dbSNP.pl' to process them first
You may want to join these files before using SNPdat_parse_dbSNP.pl
To join these files you can use the 'join' command
join ds*.flat > ds_arabidopsis_3702.all.flat
```

### **Output file**

SNPdat creates a single output file with upto 25 columns of information about each queried SNP. This output file is tab-delimited and can easily be opened with any spreadsheet software such as Microsoft Excel and OpenOffice.

An output file can be specified using the -o switch. This will be explained in greater detail later. If no output file is specified SNPdat will create one with the same name as the input file but with the suffix .result

|    | А        | В         | С        | D         | Е        | F       | G        | Н        | 1           | J        | K         | L        | М          | N         | 0        | Р         | Q       | R          | S        | T       | U        | V        | W          | χ         | Υ        |
|----|----------|-----------|----------|-----------|----------|---------|----------|----------|-------------|----------|-----------|----------|------------|-----------|----------|-----------|---------|------------|----------|---------|----------|----------|------------|-----------|----------|
| 1  | Chromsor | SNP Posit | Within a | Region    | Distance | Feature | Number o | Number o | Start of cu | nd of cu | gene ID o | gene nar | transcript | transcrip | Annotate | Strand se | Annotat | e Estimate | Estimate | Codon   | Amino Ad | synonymo | Protein IC | RSidentif | Addition |
| 2  | 25       | 117035    | Υ        | Exonic    | NA       | exon    | 2        | [1/1]    | 117033      | 117108   | ENSBTAG   | RHDF1_B  | ENSBTATO   | RHDF1_B   | [15/18]  | -         | NA      | <b>*-2</b> | 0        | [C/T]AG | [Q/-]    | N        | NA         | NA        |          |
| 3  | 25       | 117035    | Υ        | Exonic    | NA       | CDS     | 2        | [1/1]    | 117033      | 117108   | ENSBTAG   | RHDF1_B  | ENSBTATO   | RHDF1_B   | [15/18]  | -         | -:      | NA NA      | 0        | [C/T]AG | [Q/-]    | N        | ENSBTAPO   | NA        |          |
| 4  | 25       | 639       | N        | Intergeni | 216      | exon    | NA       | NA       | 304         | 423      | ENSBTAG   | (NA      | ENSBTATO   | NA        | [1/1]    | +         | NA      | NA         | NA       | NA      | NA       | NA       | NA         | NA        |          |
| 5  | 25       | 646       | N        | Intergeni | 223      | exon    | NA       | NA       | 304         | 423      | ENSBTAG   | (NA      | ENSBTATO   | NA        | [1/1]    | +         | NA      | NA         | NA       | NA      | NA       | NA       | NA         | rs1328994 | 15       |
| 6  | 25       | 769       | N        | Intergeni | 346      | exon    | NA       | NA       | 304         | 423      | ENSBTAG   | (NA      | ENSBTATO   | NA        | [1/1]    | +         | NA      | NA         | NA       | NA      | NA       | NA       | NA         | NA        |          |
| 7  | 25       | 782       | N        | Intergeni | 359      | exon    | NA       | NA       | 304         | 423      | ENSBTAG   | (NA      | ENSBTATO   | NA        | [1/1]    | +         | NA      | NA         | NA       | NA      | NA       | NA       | NA         | NA        |          |
| 8  | 25       | 1211      | N        | Intergeni | 788      | exon    | NA       | NA       | 304         | 423      | ENSBTAG   | (NA      | ENSBTATO   | NA        | [1/1]    | +         | NA      | NA         | NA       | NA      | NA       | NA       | NA         | NA        |          |
| 9  | 25       | 1254      | N        | Intergeni | 831      | exon    | NA       | NA       | 304         | 423      | ENSBTAG   | (NA      | ENSBTATO   | NA        | [1/1]    | +         | NA      | NA         | NA       | NA      | NA       | NA       | NA         | NA        |          |
| 10 | 25       | 1288      | N        | Intergeni | 865      | exon    | NA       | NA       | 304         | 423      | ENSBTAG   | (NA      | ENSBTATO   | NA        | [1/1]    | +         | NA      | NA         | NA       | NA      | NA       | NA       | NA         | NA        |          |
| 11 | 25       | 2125      | N        | Intergeni | 1702     | exon    | NA       | NA       | 304         | 423      | ENSBTAG   | NA       | ENSBTATO   | NA        | [1/1]    | +         | NA      | NA         | NA       | NA      | NA       | NA       | NA         | rs1365772 | .65      |

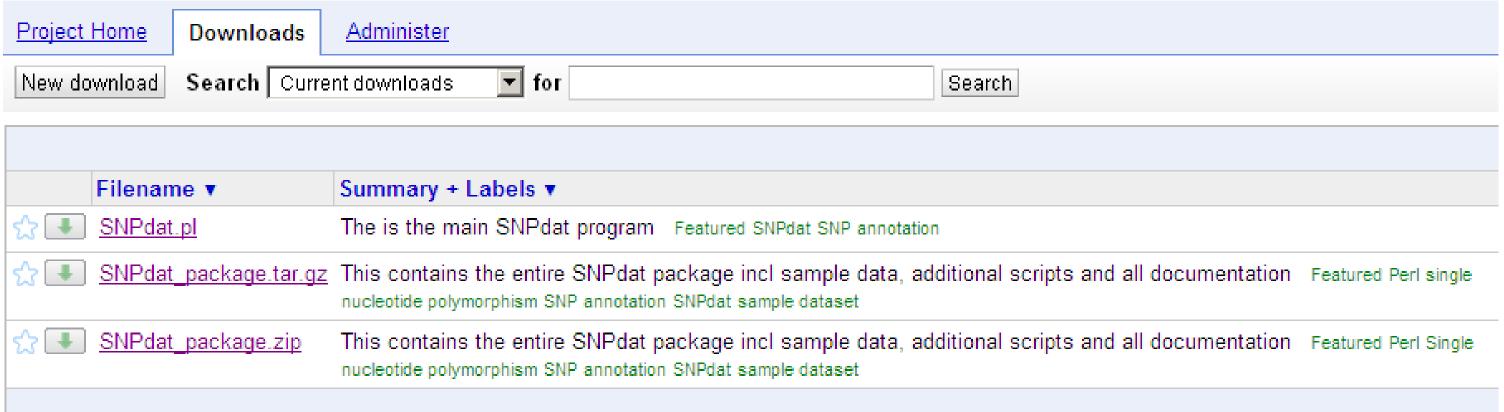
| Column Number | Description  |
|---------------|--|
| 1             | The queried SNPs chromosome ID   |
| 2             | The queried SNPs genomic location  |
| 3             | Whether or not the SNP was within a feature  |
| 4             | Region containing the SNP; either exonic, intronic, or intergenic  |
| 5             | Distance to nearest feature  |
| 6             | Either the closest feature to the SNP or the feature containing the SNP  |
| 7             | The number of different features that the SNP is annotated to  |
| 8             | The number of annotations of the current feature   |
| 9             | Start of feature (bp)  |
| 10            | End of feature (bp)  |
| 11            | The gene ID for the current feature  |
| 12            | The gene name for the current feature  |
| 13            | The transcript ID for the current feature  |
| 14            | The transcript name for the current feature  |
| 15            | The exon that contains the current feature and the total number of annotated exons for the gene containing the feature |
| 16            | The strand sense of the feature  |
| 17            | The annotated reading frame (when contained in GTF)  |
| 18            | The reading frame estimated by SNPdat  |
| 19            | The estimated number of stop codons in the estimated reading frame   |
| 20            | The codon containing the SNP, position in the codon and reference base and mutation                                    |
| 21            | The amino acid for the reference codon and new amino acid with mutation in place                                       |
| 22            | Whether or not the mutation is synonymous  |
| 23            | The protein ID for the current feature   |
| 24            | The RS identifier for queries that map to known SNPs   |
| 25            | Error messages, warnings etc   |

### An example

Sample input files can be downloaded from <a href="http://www.code.google./p/snpdat/downloads">http://www.code.google./p/snpdat/downloads</a>

From here you can download SNPdat. You can also, download the SNPdat package, which includes all additional scripts, the manual, a tutorial and sample data.





## There should be 4 folders and the SNPdat script within this download

- •Tutorial two files
  - •SNPdat\_tutorial.ppt
  - •SNPdat\_tutorial.pdf

- additional\_scripts three files
  - •dbSNP\_finder.pl
  - •GTF\_FASTA\_finder.pl
  - •SNPdat\_parse\_dbsnp.pl

- •Sample dataset 6 files
  - •SNPdat\_sample\_input.txt
  - •SNPdat\_sample\_input.txt.output
  - •SNPdat\_sample\_UMD3.1.gtf
  - •SNPdat\_sample\_UMD3.1.fa
  - •SNPdat\_dbSNP\_example.flat
  - •dbSNP\_processed.txt

- •Manual two files
  - •SNPdat\_manual.doc
  - •SNPdat\_manual.pdf

SNPdat.pl

### An example

Once these files have been downloaded, extract them to a location you can navigate to using the terminal. Ensure all files are in the same directory.

Once this has been done, the first thing you will want to do is process the dbSNP FLAT file using **SNPdat\_parse\_dbsnp.pl**This is only a sample of the full FLAT file available from <a href="mailto:ttp://ftp.ncbi.nih.gov/snp/organisms/">ttp://ftp.ncbi.nih.gov/snp/organisms/</a>. dbSNP files may contain information on multiple genome builds for the same organism, so you will also need to know what build you are interested in.
In the below example we are interested in Bos\_taurus\_UMD\_3.1 e.g.

```
adoran@server ~/directory/with_all/SNPdat_files/
$ perl SNPdat_parse_dbsnp.pl SNPdat_dbSNP_example.flat dbSNP_processed.txt

This is a perl script to process a dbSNP FLAT file of SNP data and produce an output file that can be supplied as input for the software SNPdat

Please enter the assembly you want to map rs ids to :Bos_taurus_UMD_3.1

The resulting output file is dbnsp_processed.txt
This file can be used as input for SNPdat using the -d switch
```

### SNPdat\_parse\_dbsnp.pl

SNPdat\_parse\_dbsnp.pl is and additional script provided with the SNPdat package to parse dbSNP FLAT files and create a file in a format suitable for use with SNPdat

```
adoran@server ~/directory/with_all/SNPdat_files/
$ perl SNPdat_parse_dbsnp.pl SNPdat_dbSNP_example.flat dbSNP_processed.txt

This is a perl script to process a dbSNP FLAT file of SNP data and produce an output file that can be supplied as input for the software SNPdat

Please enter the assembly you want to map rs ids to :Bos_taurus_UMD_3.1

The resulting output file is dbnsp_processed.txt

This file can be used as input for SNPdat using the -d switch
```

The parsed output file (dbSNP\_processed.txt in the above example), is a simple tab-delimited file with 3 columns
The first column is the chromosome name
The second column is the coordinate on the chromosome of the SNP
The third column is the mutation

### **Running SNPdat**

Now you have created the optional input file and can run SNPdat.

When using SNPdat you must specify each file type on the command line.

NOTE – Chromosome Identifiers must be the same format across all input files

To specify file types for SNPdat a unique switch is used

- -i input file
- -g GTF file
- -f FASTA file
- -o output file
- -d processed dbSNP file
- -s summary report file

|adoran@server ~/directory/with\_all/SNPdat\_files/

💲 perl SNPdat.pl -i SNPdat\_sample\_input.txt -g SNPdat\_sample\_UMD3.1.gtf -f SNPdat\_sample\_UMD3.1.fa -d dbSNP\_processed.txt -o SNPdat\_sample\_input.txt.output

### **SNPdat output**

|    | А        | В          | С           | D        | Е           | F       | G         | Н         |              | J           | K          | L         | M                |
|----|----------|------------|-------------|----------|-------------|---------|-----------|-----------|--------------|-------------|------------|-----------|------------------|
| 1  | Chromson | SNP Positi | Within a Fe | Region   | Distance to | Feature | Number of | Number of | Start of cur | End of curr | gene ID co | gene name | transcript II tr |
| 2  | CHR25    | 1818721    | N           | Intronic | 470         | exon    | NA        | NA        | 1818078      | 1818251     | ENSBTAG    | E1BCR1_   | ENSBTATIE        |
| 3  | CHR25    | 1818721    | N           | Intronic | 470         | CDS     | NA        | NA        | 1818078      | 1818251     | ENSBTAG    | E1BCR1_   | ENSBTAT(E        |
| 4  | CHR25    | 1818938    | N           | Intronic | 687         | exon    | NA        | NA        | 1818078      | 1818251     | ENSBTAG    | E1BCR1_   | ENSBTATIE        |
| 5  | CHR25    | 1818938    | N           | Intronic | 687         | CDS     | NA        | NA        | 1818078      | 1818251     | ENSBTAG    | E1BCR1_   | ENSBTATIE        |
| 6  | CHR25    | 1819363    | N           | Intronic | 547         | exon    | NA        | NA        | 1819910      | 1820030     | ENSBTAG    | E1BCR1_   | ENSBTAT(E        |
| 7  | CHR25    | 1819363    | N           | Intronic | 547         | CDS     | NA        | NA        | 1819910      | 1820030     | ENSBTAG    | E1BCR1_   | ENSBTAT(E        |
| 8  | CHR25    | 1819436    | N           | Intronic | 474         | exon    | NA        | NA        | 1819910      | 1820030     | ENSBTAG    | E1BCR1_   | ENSBTATIE        |
| 9  | CHR25    | 1819436    | N           | Intronic | 474         | CDS     | NA        | NA        | 1819910      | 1820030     | ENSBTAG    | E1BCR1_   | ENSBTATIE        |
| 10 | CHR25    | 1819646    | N           | Intronic | 264         | exon    | NA        | NA        | 1819910      | 1820030     | ENSBTAG    | E1BCR1_   | ENSBTATIE        |
| 11 | CHR25    | 1819646    | N           | Intronic | 264         | CDS     | NA        | NA        | 1819910      | 1820030     | ENSBTAG    | E1BCR1_   | ENSBTAT(E        |
| 12 | CHR25    | 1819682    | N           | Intronic | 228         | exon    | NA        | NA        | 1819910      | 1820030     | ENSBTAG    | E1BCR1_   | ENSBTAT(E        |
| 13 | CHR25    | 1819682    | N           | Intronic | 228         | CDS     | NA        | NA        | 1819910      | 1820030     | ENSBTAG    | E1BCR1_   | ENSBTAT(E        |
| 14 | CHR25    | 1819940    | Υ           | Exonic   | NA          | exon    | 2         | [1/1]     | 1819910      | 1820030     | ENSBTAG    | E1BCR1_   | ENSBTAT(E        |
| 15 | CHR25    | 1819940    | Υ           | Exonic   | NA          | CDS     | 2         | [1/1]     | 1819910      | 1820030     | ENSBTAG    | E1BCR1_   | ENSBTAT(E        |

### **SNPdat output**

|    | M            | N             | 0         | Р          | Q           | R         | S         | Т       | U          | ٧         | W            | Χ           | Υ              |
|----|--------------|---------------|-----------|------------|-------------|-----------|-----------|---------|------------|-----------|--------------|-------------|----------------|
| 1  | transcript l | (transcript r | Annotated | Strand sen | Annotated ( | Estimated | Estimated | Codon   | Amino Acid | synonymol | Protein ID ( | RSidentifie | Additional Not |
| 2  | ENSBTAT      | (E1BCR1_8     | [8/30]    | -          | NA          | NA        | NA        | NA      | NA         | NA        | NA           | NA          |                |
| 3  | ENSBTAT      | (E1BCR1_8     | [8/30]    | -          | -2          | NA        | NA        | NA      | NA         | NA        | ENSBTAP      | NA          |                |
| 4  | ENSBTAT      | (E1BCR1_8     | [8/30]    | -          | NA          | NA        | NA        | NA      | NA         | NA        | NA           | NA          |                |
| 5  | ENSBTAT      | (E1BCR1_8     | [8/30]    | -          | -2          | NA        | NA        | NA      | NA         | NA        | ENSBTAP      | NA          |                |
| 6  | ENSBTAT      | (E1BCR1_6     | [7/30]    | -          | NA          | NA        | NA        | NA      | NA         | NA        | NA           | NA          |                |
| 7  | ENSBTAT      | (E1BCR1_6     | [7/30]    | -          | 0           | NA        | NA        | NA      | NA         | NA        | ENSBTAP      | NA          |                |
| 8  | ENSBTAT      | (E1BCR1_6     | [7/30]    | -          | NA          | NA        | NA        | NA      | NA         | NA        | NA           | NA          |                |
| 9  | ENSBTAT      | (E1BCR1_6     | [7/30]    | -          | 0           | NA        | NA        | NA      | NA         | NA        | ENSBTAP      | NA          |                |
| 10 | ENSBTAT      | (E1BCR1_6     | [7/30]    | -          | NA          | NA        | NA        | NA      | NA         | NA        | NA           | NA          |                |
| 11 | ENSBTAT      | (E1BCR1_6     | [7/30]    | -          | 0           | NA        | NA        | NA      | NA         | NA        | ENSBTAP      | NA          |                |
| 12 | ENSBTAT      | (E1BCR1_6     | [7/30]    | -          | NA          | NA        | NA        | NA      | NA         | NA        | NA           | NA          |                |
| 13 | ENSBTAT      | (E1BCR1_6     | [7/30]    | -          | 0           | NA        | NA        | NA      | NA         | NA        | ENSBTAP      | NA          |                |
| 14 | ENSBTAT      | (E1BCR1_6     | [7/30]    | -          | NA          | *-1       | 0         | C[A/T]G | [Q/L]      | N         | NA           | NA          |                |
| 15 | ENSBTAT      | E1BCR1_E      | [7/30]    | -          | 0           | NA        | 0         | C[A/T]G | [Q/L]      | N         | ENSBTAP      | NA          |                |

### Licence

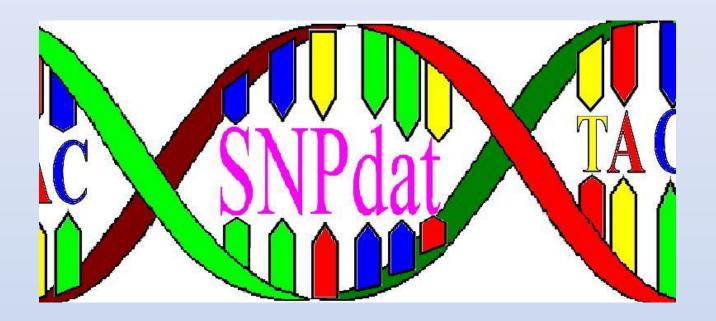
SNPdat is freely available under a GNU Public License (Version 2 <a href="http://www.gnu.org/licenses/gpl-2.0.html">http://www.gnu.org/licenses/gpl-2.0.html</a>) at: <a href="http://code.google.com/p/snpdat">http://code.google.com/p/snpdat</a>

SNPdat: rapid annotation of novel SNPs for model and non-model organisms Copyright (C) 2012 Anthony Doran

This program is free software; you can redistribute it and/or modify it under the terms of the GNU General Public License as published by the Free Software Foundation; either version 2 of the License any later version.

This program is distributed in the hope that it will be useful, but WITHOUT ANY WARRANTY; without even the implied warranty of MERCHANTABILITY or FITNESS FOR A PARTICULAR PURPOSE. See the GNU General Public License for more details.

You should have received a copy of the GNU General Public License along with this program; if not, write to the Free Software Foundation, Inc., 51 Franklin Street, Fifth Floor, Boston, MA 02110-1301, USA.



This tutorial is only a brief summary and introduction to SNPdat and how to use SNPdat. For more information on using SNPdat or any of the available options (not discussed here) please read the manual

The manual is available from the website (<a href="http://code.google.com/p/snpdat/">http://code.google.com/p/snpdat/</a>).

The published manuscript for SNPdat is available from BMC Bioinformatics (<a href="http://www.biomedcentral.com/1471-2105/14/45#">http://www.biomedcentral.com/1471-2105/14/45#</a>).