

# A toolkit for DNA sequence analysis and manipulation

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## Chapter 1

# Introduction

Recent advances in DNA sequencing have revolutionized the field of genomics, making it possible for research groups to generate large amounts of sequenced data, very rapidly and at substantially lower cost. Its storage have been made using specific file formats, such as FASTQ and FASTA. Therefore, its analysis and manipulation is crucial [?]. Several frameworks for analysis and manipulation emerged, namely GALAXY [?], GATK [?], HTSeq [?], MEGA [?], among others. In the majority, these frameworks require licenses and do not provide a low level access to the information, since they are commonly approached by scripting or interfaces.

We describe GOOSE, a (free) novel toolkit for analyzing and manipulating FASTA-FASTQ formats and sequences (DNA, amino acids, text), with many complementary tools. The toolkit is for Linux-based systems, built for fast processing. GOOSE supports pipes for easy integration. It includes tools for information display, randomizing, edition, conversion, extraction, searching, calculation and visualization. GOOSE is prepared to deal with very large datasets, typically in the scale Gigabytes or Terabytes.

The toolkit is a command line version, using the prefix "goose-" followed by the suffix with the respective name of the program. GOOSE is implemented in C language and it is available, under GPLv3, at:

```
https://pratas.github.io/goose
```

#### 1.1 Installation

For GOOSE installation, run:

```
git clone https://github.com/pratas/goose.git
cd goose/src/
make
```

#### 1.2 License

The license is **GPLv3**. In resume, everyone is permitted to copy and distribute verbatim copies of this license document, but changing it is not allowed. For details on the license, consult: http://www.gnu.org/

licenses/gpl-3.0.html.

## Chapter 2

# FASTQ tools

Current available tools for FASTQ format analysis and manipulation include:

- 1. goose-fastq2fasta: it converts a FASTQ file format to a pseudo FASTA file.
- 2. goose-fastq2mfasta: it converts a FASTQ file format to a pseudo Multi-FASTA file.
- 3. goose-FastqExcludeN: it discards the FASTQ reads with the minimum number of "N" symbols.
- 4. goose-FastqExtractQualityScores: it extracts all the quality-scores from FASTQ reads.
- 5. goose-FastqInfo: it analyses the basic informations of FASTQ file format.
- 6. goose-FastqMaximumReadSize: it filters the FASTQ reads with the length higher than the value defined.
- 7. goose-FastqMinimumQualityScore: it discards reads with average quality-score below of the defined.
- 8. goose-FastqMinimumReadSize: it filters the FASTQ reads with the length smaller than the value defined.
- 9. goose-randfastqextrachars: it substitues in the FASTQ files, the DNA sequence the outside ACGT chars by random ACGT symbols.
- 10. goose-seq2fastq: it converts a genomic sequence to pseudo FASTQ file format.
- 11. goose-mutatefastq: it creates a synthetic mutation of a FASTQ file given specific rates of mutations, deletions and additions.

## $2.1 \quad Program\ goose-fastq2 fasta$

The goose-fastq2fasta converts a FASTQ file format to a pseudo FASTA file. However, it does not align the sequence. Also, it extracts the sequence and adds a pseudo header. For help type:

```
./goose-fastq2fasta -h
```

In the following subsections, we explain the input and output paramters.

#### Input parameters

The goose-fastq2fasta program needs two streams for the computation, namely the input and output standard. The input stream is a FASTQ file.

The attribution is given according to:

An example on such an input file is:

#### Output

The output of the goose-fastq2fasta program a FASTA file.

An example, for the input, is:

```
GGGTGATGGCCGCTGCCGATGGCGTCAAATCCCACCAAGTTACCCTTAACAACTTAAGGGTTTTCAAATAGA
GTTCAGGGATACGACGTTTGTATTTTAAGAATCTGAAGCAGAAGTCGATGATAATACGCGTCGTTTTATCAT
```

## 2.2 Program goose-fastq2mfasta

The goose-fastq2mfasta onverts a FASTQ file format to a pseudo Multi-FASTA file. However, it does not align the sequence. Also, it extracts the sequence and adds a pseudo header.

For help type:

```
./goose-fastq2mfasta-h
```

In the following subsections, we explain the input and output paramters.

#### Input parameters

The goose-fastq2mfasta program needs two streams for the computation, namely the input and output standard. The input stream is a FASTQ file.

The attribution is given according to:

```
Usage: ./goose-fastq2mfasta [options] [[--] args]
    or: ./goose-fastq2mfasta [options]

It converts a FASTQ file format to a pseudo Multi-FASTA file.

It does NOT align the sequence.

It extracts the sequence and adds each header in a Multi-FASTA format.

-h, --help show this help message and exit

Basic options
    < input.fastq Input FASTQ file format (stdin)
    > output.mfasta Output Multi-FASTA file format (stdout)

Example: ./goose-fastq2mfasta < input.fastq > output.mfasta
```

An example on such an input file is:

#### Output

The output of the goose-fastq2mfasta program a Multi-FASTA file.

An example, for the input, is:

```
>SRROO1666.1 071112_SLXA-EAS1_s_7:5:1:817:345 length=72

GGGTGATGGCCGCTGCCGATGGCGTCAAATCCCACCAAGTTACCCTTAACAACTTAAGGGTTTTCAAATAGA

>SRROO1666.2 071112_SLXA-EAS1_s_7:5:1:801:338 length=72

GTTCAGGGATACGACGTTTGTATTTTAAGAATCTGAAGCAGAAGTCGATGATAATACGCGTCGTTTTATCAT
```

## 2.3 Program goose-FastqExcludeN

The goose-FastqExcludeN discards the FASTQ reads with the minimum number of "N" symbols. Also, if present, it will erase the second header (after +).

For help type:

```
./goose-FastqExcludeN-h
```

In the following subsections, we explain the input and output paramters.

#### Input parameters

The goose-FastqExcludeN program needs two streams for the computation, namely the input and output standard. The input stream is a FASTQ file.

The attribution is given according to:

```
Usage: ./goose-FastqExcludeN [options] [[--] args]
   or: ./goose-FastqExcludeN [options]
It discards the FASTQ reads with the minimum number of ''N'' symbols. If present,
it will erase the second header (after +).
    -h, --help
                          show this help message and exit
Basic options
   -m, --max = <int>
                          The maximum of of "N" symbols in the read
   < input.fastq</pre>
                        Input FASTQ file format (stdin)
   > output
                          Output read information (stdout)
Example: ./goose-FastqExcludeN < input.fastq > output
Output example :
<FASTQ non-filtered reads>
Total reads : value
Filtered reads : value
```

An example on such an input file is:

#### Output

The output of the goose-FastqExcludeN program is a set of all the filtered FASTQ reads, followed by the execution report.

Using the max value as 5, an example for this input, is:

### 2.4 Program goose-FastqExtractQualityScores

The goose-FastqExtractQualityScores extracts all the quality-scores from FASTQ reads. For help type:

```
./goose-FastqExtractQualityScores -h
```

In the following subsections, we explain the input and output paramters.

#### Input parameters

The goose-FastqExtractQualityScores program needs two streams for the computation, namely the input and output standard. The input stream is a FASTQ file.

The attribution is given according to:

An example on such an input file is:

#### Output

The output of the goose-FastqExtractQualityScores program is a set of all the quality scores from the FASTQ reads, followed by the execution report.

An example, for the input, is:

## 2.5 Program goose-FastqInfo

The goose-FastqInfo analyses the basic informations of FASTQ file format.

For help type:

```
./goose-FastqInfo-h
```

In the following subsections, we explain the input and output paramters.

#### Input parameters

The goose-FastqInfo program needs two streams for the computation, namely the input and output standard. The input stream is a FASTQ file.

```
Usage: ./goose-FastqInfo [options] [[--] args]
  or: ./goose-FastqInfo [options]
It analyses the basic informations of FASTQ file format.
   -h, --help
                       show this help message and exit
Basic options
                     Input FASTQ file format (stdin)
   < input.fastq</pre>
   > output
                       Output read information (stdout)
Example: ./goose-FastqInfo < input.fastq > output
Output example :
Total reads : value
Max read length : value
Min read length : value
Min QS value : value
Max QS value : value
QS range : value
```

#### Output

The output of the goose-FastqInfo program is a set of informations related with the file readed. An example, for the input, is:

```
Total reads : 2

Max read length : 72

Min read length : 72

Min QS value : 41

Max QS value : 73

QS range : 33
```

## 2.6 Program goose-FastqMaximumReadSize

The goose-FastqMaximumReadSize filters the FASTQ reads with the length higher than the value defined. For help type:

```
./goose-FastqMaximumReadSize -h
```

In the following subsections, we explain the input and output paramters.

#### Input parameters

The goose-FastqMaximumReadSize program needs two streams for the computation, namely the input and output standard. The input stream is a FASTQ file.

```
Usage: ./goose-FastqMaximumReadSize [options] [[--] args]
or: ./goose-FastqMaximumReadSize [options]

It filters the FASTQ reads with the length higher than the value defined.

If present, it will erase the second header (after +).

-h, --help show this help message and exit

Basic options
```

```
-s, --size=<int> The maximum read length
< input.fastq Input FASTQ file format (stdin)
> output Output read information (stdout)

Example: ./goose-FastqMaximumReadSize < input.fastq > output

Output example:
<FASTQ non-filtered reads>
Total reads : value
Filtered reads : value
```

#### Output

The output of the goose-FastqMaximumReadSize program is a set of all the filtered FASTQ reads, followed by the execution report.

Using the size value as 60, an example for this input, is:

## 2.7 Program goose-FastqMinimumQualityScore

The goose-FastqMinimumQualityScore discards reads with average quality-score below of the defined. For help type:

```
./goose-FastqMinimumQualityScore -h
```

In the following subsections, we explain the input and output paramters.

#### Input parameters

The goose-FastqMinimumQualityScore program needs two streams for the computation, namely the input and output standard. The input stream is a FASTQ file.

```
Usage: ./goose-FastqMinimumQualityScore [options] [[--] args]
   or: ./goose-FastqMinimumQualityScore [options]
It discards reads with average quality-score below value.
   -h, --help
                         show this help message and exit
Basic options
   -m, --min=<int>
                         The minimum average quality-score (Value 25 or 30 is commonly used)
   < input.fastq
                        Input FASTQ file format (stdin)
   > output
                         Output read information (stdout)
Example: ./goose-FastqMinimumQualityScore < input.fastq > output
Output example :
<FASTQ non-filtered reads>
Total reads : value
Filtered reads : value
```

#### Output

The output of the goose-FastqMinimumQualityScore program is a set of all the filtered FASTQ reads, followed by the execution report.

Using the minimum average value as 30, an example for this input, is:

## $2.8\quad {\bf Program\ goose\text{-}FastqMinimumReadSize}$

The goose-FastqMinimumReadSize filters the FASTQ reads with the length smaller than the value defined. For help type:

```
./goose-FastqMinimumReadSize -h
```

In the following subsections, we explain the input and output paramters.

#### Input parameters

The goose-FastqMinimumReadSize program needs two streams for the computation, namely the input and output standard. The input stream is a FASTQ file.

The attribution is given according to:

```
Usage: ./goose-FastqMinimumReadSize [options] [[--] args]
   or: ./goose-FastqMinimumReadSize [options]
It filters the FASTQ reads with the length smaller than the value defined.
If present, it will erase the second header (after +).
    -h, --help
                          show this help message and exit
Basic options
    -s, --size=<int>
                         The minimum read length
   < input.fastq</pre>
                          Input FASTQ file format (stdin)
   > output
                          Output read information (stdout)
Example: ./goose-FastqMinimumReadSize < input.fastq > output
Output example :
<FASTQ non-filtered reads>
Total reads : value
Filtered reads : value
```

An example on such an input file is:

#### Output

The output of the goose-FastqMinimumReadSize program is a set of all the filtered FASTQ reads, followed by the execution report.

Using the size value as 65, an example for this input, is:

## 2.9 Program goose-mutatefastq

The goose-mutatefastq creates a synthetic mutation of a FASTQ file given specific rates of mutations, deletions and additions. All these parameters are defined by the user, and their are optional. For help type:

```
./goose-mutatefastq-h
```

In the following subsections, we explain the input and output paramters.

#### Input parameters

The goose-mutatefastq program needs two streams for the computation, namely the input and output standard. However, optional settings can be supplied too, such as the starting point to the random generator, and the edition, deletion and insertion rates. Also, the user can choose to use the ACGTN alphabet in the synthetic mutation. The input stream is a FASTQ File.

The attribution is given according to:

```
Usage: ./goose-mutatefastq [options] [[--] args]
   or: ./goose-mutatefastq [options]
Creates a synthetic mutation of a FASTQ file given specific rates of mutations,
deletions and additions
    -h, --help
                                  show this help message and exit
Basic options
    < input.fasta</pre>
                                 Input FASTQ file format (stdin)
    > output.fasta
                                  Output FASTQ file format (stdout)
Optional
    -s, --seed=<int>
                                  Starting point to the random generator
   -m, --mutation-rate=<dbl>
                                Defines the mutation rate (default 0.0)
    -d, --deletion-rate=<dbl>
                                Defines the deletion rate (default 0.0)
    -i, --insertion-rate=<dbl>
                                Defines the insertion rate (default 0.0)
    -a, --ACGTN-alphabet
                                  When active, the application uses the ACGTN alphabet
Example: ./goose-mutatefastq -s <seed> -m <mutation rate> -d <deletion rate> -i
<insertion rate> -a < input.fastq > output.fastq
```

An example on such an input file is:

#### Output

The output of the goose-mutatefastq program is a FASTQ file whith the synthetic mutation of input file. Using the seed value as 1 and the mutation rate as 0.5, an example for this input, is:

## 2.10 Program goose-randfastqextrachars

The goose-randfastqextrachars substitues in the FASTQ files, the DNA sequence the outside ACGT chars by random ACGT symbols.

For help type:

```
./goose-randfastqextrachars -h
```

In the following subsections, we explain the input and output paramters.

#### Input parameters

The goose-randfastqextrachars program needs two streams for the computation, namely the input and output standard. The input stream is a FASTQ file.

```
Usage: ./goose-randfastqextrachars [options] [[--] args]
or: ./goose-randfastqextrachars [options]

It substitues in the FASTQ files, the DNA sequence the outside ACGT chars by random ACGT symbols.

-h, --help show this help message and exit

Basic options
```

#### Output

The output of the goose-randfastqextrachars program is a FASTQ file.

An example, for the input, is:

## 2.11 Program goose-seq2fastq

The goose-seq2fastq converts a genomic sequence to pseudo FASTQ file format.

For help type:

```
./goose-seq2fastq -h
```

In the following subsections, we explain the input and output paramters.

#### Input parameters

The goose-seq2fastq program needs two streams for the computation, namely the input and output standard. The input stream is a sequence group file.

```
Usage: ./goose-seq2fastq [options] [[--] args]
  or: ./goose-seq2fastq [options]

It converts a genomic sequence to pseudo FASTQ file format.
```

#### Output

The output of the goose-seq2fastq program is a pseudo FASTQ file.

An example, using the size line as 80 and the read's header as "Seq2Fastq", for the input, is:

+
FFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFF
@Seq2Fastq7
GG A A GC GG C A GG A A T A A GG A A A G C A G C C T C C T G A C T T T C C T C G C T T G G T T T G A G T G G A C C T C C C A G G C C A G T G C C G
+
FFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFF
@Seq2Fastq8
GGCCCCTC AT AGGAGGAAGCTCGGGAGGTGGCC AGGCGGCAGGCACCCCCCCAGCAATCCGCGCGCCGGGAC
+
FFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFF
@Seq2Fastq9
A G A A T G C C C T G C A G G A A C T T C T T C T G G A A G A C C T T C T C C T C C T G C A A A T A A A A C C T C A C C C A T G A A T G C T C A C G C A A G T T
+
FFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFF
@Seq2Fastq10
TAATTACAGACCTGAA
+
FFFFFFFFFFFFF

## Chapter 3

## FASTA tools

Current available FASTA tools, for analysis and manipulation, are:

- 1. goose-fasta2seq: it converts a FASTA or Multi-FASTA file format to a seq.
- 2. goose-fastaextract: it extracts sequences from a FASTA file, which the range is defined by the user in the parameters.
- 3. goose-fastaextractbyread: it extracts sequences from each read in a Multi-FASTA file (splited by \n), which the range is defined by the user in the parameters.
- 4. goose-fastainfo: it shows the readed information of a FASTA or Multi-FASTA file format.
- 5. goose-mutatefasta: it reates a synthetic mutation of a fasta file given specific rates of editions, deletions and additions.
- 6. goose-randfastaextrachars: it substitues in the DNA sequence the outside ACGT chars by random ACGT symbols.
- 7. goose-extractreadbypattern: it extracts reads from a Multi-FASTA file format given a pattern in the header.
- 8. goose-findnpos: it reports the "N" regions in a sequence or FASTA (seq) file.
- 9. goose-seq2fasta: it converts a genomic sequence to pseudo FASTA file format.
- 10. goose-splitreads: it splits a Multi-FASTA file to multiple FASTA files.
- 11. goose-renamehumanheaders: it changes the headers of FASTA or Multi-FASTA file to simple chrX by order, where X is the number.

## 3.1 Program goose-fasta2seq

The goose-fasta2seq converts a FASTA or Multi-FASTA file format to a sequence. For help type:

```
./goose-fasta2seq -h
```

In the following subsections, we explain the input and output paramters.

#### Input parameters

The goose-fasta2seq program needs two streams for the computation, namely the input and output standard. The input stream is a FASTA or Multi-FASTA file.

The attribution is given according to:

An example on such an input file is:

#### Output

The output of the goose-fasta2seq program is a group sequence.

An example, for the input, is:

 

## 3.2 Program goose-fastaextract

The goose-fastaextract extracts sequences from a FASTA file, which the range is defined by the user in the parameters.

For help type:

```
./goose-fastaextract -h
```

In the following subsections, we explain the input and output paramters.

#### Input parameters

The goose-fastaextract program needs two parameters, which defines the begin and the end of the extraction, and two streams for the computation, namely the input and output standard. The input stream is a FASTA file.

The attribution is given according to:

```
Usage: ./goose-fastaextract [options] [[--] args]
or: ./goose-fastaextract [options]

It extracts sequences from a FASTA file.

-h, --help show this help message and exit

Basic options
-i, --init=<int> The first position to start the extraction (default 0)
-e, --end=<int> The last extract position (default 100)
< input.fasta Input FASTA or Multi-FASTA file format (stdin)
> output.seq Output sequence file (stdout)

Example: ./goose-fastaextract -i <init> -e <end> < input.fasta > output.seq
```

An example on such an input file is:

```
> ABOOO 264 | acc = ABOOO 264 | descr = Homo sapiens mRNA
ACAAGACGGCCTCCTGCTGCTGCTGCTCCCGGGGCCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGCCCCTCATAGGAGAGGCTCGGGAGCTGGCCAGGCGGCAGGAAGCAGGCCAGTGCC
GCGAATCCGCGCGCCCGGGACAGATCTCCTGCAAAGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCACCCCCCCAGC
TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
```

#### Output

The output of the goose-fastaextract program is a group sequence.

An example, using the value 0 as extraction starting point and the 50 as the end, for the provided input,

## 3.3 Program goose-fastaextractbyread

The goose-fastaextractbyread extracts sequences from a FASTA or Multi-FASTA file, which the range is defined by the user in the parameters.

For help type:

```
./goose-fastaextractbyread-h
```

In the following subsections, we explain the input and output paramters.

#### Input parameters

The goose-fastaextractbyread program needs two parameters, which defines the begin and the end of the extraction, and two streams for the computation, namely the input and output standard. The input stream is a FASTA or Multi-FASTA file.

The attribution is given according to:

```
Usage: ./goose-fastaextractbyread [options] [[--] args]
or: ./goose-fastaextractbyread [options]

It extracts sequences from each read in a Multi-FASTA file (splited by \n)

-h, --help show this help message and exit

Basic options
-i, --init=<int> The first position to start the extraction (default 0)
-e, --end=<int> The last extract position (default 100)
< input.fasta Input FASTA or Multi-FASTA file format (stdin)
> output.fasta Output FASTA or Multi-FASTA file format (stdout)

Example: ./goose-fastaextractbyread -i <init> -e <end> < input.fasta > output.fasta
```

An example on such an input file is:

#### Output

The output of the goose-fastaextractbyread program is FASTA or Multi-FASTA file wiht the extracted sequences.

An example, using the value 0 as extraction starting point and the 50 as the end, for the provided input, is:

```
>AB000264 |acc=AB000264|descr=Homo sapiens mRNA
ACAAGACGGCCTCCTGCTGCTGCTCCTCCGGGGCCACGGCCCTGGAGG
>AB000263 |acc=AB000263|descr=Homo sapiens mRNA
ACAAGATGCCATTGTCCCCCGGCCTCCTGCTGCTGCTCCCGGGGCC
```

### 3.4 Program goose-fastainfo

The goose-fastainfo shows the readed information of a FASTA or Multi-FASTA file format. For help type:

```
./goose-fastainfo-h
```

In the following subsections, we explain the input and output paramters.

#### Input parameters

The goose-fastainfo program needs two streams for the computation, namely the input and output standard. The input stream is a FASTA or Multi-FASTA file.

```
Usage: ./goose-fastainfo [options] [[--] args]
   or: ./goose-fastainfo [options]
It shows read information of a FASTA or Multi-FASTA file format.
    -h, --help
                          show this help message and exit
Basic options
   < input.fasta</pre>
                          Input FASTA or Multi-FASTA file format (stdin)
   > output
                          Output read information (stdout)
Example: ./goose-fastainfo < input.fasta > output
Output example :
Number of reads
                    : value
Number of bases
                    : value
MIN of bases in read : value
```

```
MAX of bases in read : value
AVG of bases in read : value
```

#### Output

The output of the goose-fastainfo program is a set of informations related with the file readed. An example, for the input, is:

```
Number of reads : 2
Number of bases : 736
MIN of bases in read : 368
MAX of bases in read : 368
AVG of bases in read : 368.0000
```

## 3.5 Program goose-mutatefasta

The goose-mutatefasta creates a synthetic mutation of a FASTA file given specific rates of editions, deletions and additions. All these parameters are defined by the user, and their are optional.

For help type:

```
./goose-mutatefasta-h
```

In the following subsections, we explain the input and output paramters.

#### Input parameters

The goose-mutatefasta program needs two streams for the computation, namely the input and output standard. However, optional settings can be supplied too, such as the starting point to the random generator, and the edition, deletion and insertion rates. Also, the user can choose to use the ACGTN alphabet in the synthetic mutation. The input stream is a FASTA or Multi-FASTA File.

```
Usage: ./goose-mutatefasta [options] [[--] args]
   or: ./goose-mutatefasta [options]
Creates a synthetic mutation of a fasta file given specific rates of editions,
deletions and additions
    -h, --help
                                  show this help message and exit
Basic options
   < input.fasta</pre>
                                 Input FASTA or Multi-FASTA file format (stdin)
    > output.fasta
                                  Output FASTA or Multi-FASTA file format (stdout)
Optional
    -s, --seed=<int>
                                  Starting point to the random generator
   -e, --edit-rate=<dbl>
                                Defines the edition rate (default 0.0)
    -d, --deletion-rate=<dbl>
                                Defines the deletion rate (default 0.0)
    -i, --insertion-rate=<dbl> Defines the insertion rate (default 0.0)
    -a, --ACGTN-alphabet
                                 When active, the application uses the ACGTN alphabet
Example: ./goose-mutatefasta -s <seed> -e <edit rate> -d <deletion rate> -i
<insertion rate> -a < input.fasta > output.fasta
```

#### Output

The output of the goose-mutatefasta program is a FASTA or Multi-FASTA file whith the synthetic mutation of input file.

Using the seed value as 1 and the edition rate as 0.5, an example for this input, is:

```
> ABOOO 264 | acc = ABOOO 264 | descr = Homo sapiens mRNA
ACGCAACGNATTCCTGCTGATCATANTGTNCCGCNCCCCNGCGACGGGGNCTCNCNNGCACACATNGTACCATTGTCCAC
NCTTNCANGTNANCGCTAGCAGGCTACNGTTTNTCCTCNCCTANNCCAANCNGGCGTNNNTACACTGGCACGTGCAGGCA
TNGGTCGGCNGGNNCCTCCGGNAACGGCACCGGAGACGTCGGNGGNTATACAGGTGTCANGAAACATCCCCGCGNC
GNGTGNCCNNGAANCCANAGAGTATCTCACTCACAACCCTGCGTGCACNTCTAGAGNANGACCTTACNCACCNTCCCNTT
NNGTACCACACCAATGAACGCTGCAGAAAGTCTGTTTNNAGGNGNGCA
> ABOOO 263 | acc = ABOOO 263 | descr = Homo sapiens mRNA
ATTTGAAGGCAANCGGNCCAGNAATNCGGNGGGTGCNGCTCNTGTNGGCTACGGNCATCGCGGCCCTGCTNTANTAAGCN
```

TGAACCACCGNTCGNNGCACTTAGCAATNGCGNAANCCGTCGGCACGGCGGAGACNAANCCGCTANTNNTTTCCCGCTNA
ATGGNTGTACAAGACCNACTANACCANCCTCCGTCACCACACTGGAGCGCANGATGGNNCGCTGNCTAGNAGNCNNTGAG
GCGCTCCNTCCTANAAANCCGTGGNCGAGCNCCCTATGGNAGNGTGGGGGTTTTACCGGAAGACCNTCGNGCCCTATGGG
AGCAATCANAANCTAGAAAGCTTACNGATGGTGANGAANTAGACTANG

## 3.6 Program goose-randfastaextrachars

The goose-randfastaextrachars substitues in the DNA sequence the outside ACGT chars by random ACGT symbols. It works both in FASTA and Multi-FASTA file formats.

For help type:

```
./goose-randfastaextrachars -h
```

In the following subsections, we explain the input and output paramters.

#### Input parameters

The goose-randfastaextrachars program needs two streams for the computation, namely the input and output standard. The input stream is a FASTA or Multi-FASTA file.

The attribution is given according to:

```
Usage: ./goose-randfastaextrachars [options] [[--] args]
or: ./goose-randfastaextrachars [options]

It substitues in the DNA sequence the outside ACGT chars by random ACGT symbols.

It works both in FASTA and Multi-FASTA file formats

-h, --help show this help message and exit

Basic options
<input.fasta Input FASTA or Multi-FASTA file format (stdin)
> output.fasta Output FASTA or Multi-FASTA file format (stdout)

Example: ./goose-randfastaextrachars < input.fasta > output.fasta
```

An example on such an input file is:

#### Output

The output of the goose-randfastaextrachars program is a FASTA or Multi-FASTA file. An example, for the input, is:

## 3.7 Program goose-extractreadbypattern

The goose-extractreadbypattern extracts reads from a Multi-FASTA file format given a pattern in the header. Also, this pattern is case insensitive.

For help type:

```
./goose-extractreadbypattern -h
```

In the following subsections, we explain the input and output paramters.

#### Input parameters

The goose-extractreadbypattern program needs two streams for the computation, namely the input and output standard. The input stream is a Multi-FASTA file.

```
Example: ./goose-extractreadbypattern-p <pattern> < input.fasta > output.fasta
```

#### Output

The output of the goose-extractreadbypattern program is a Multi-FASTA file.

An example, using the pattern "264", for the provided input, is:

## 3.8 Program goose-findnpos

The goose-findness reports the "N" regions in a sequence or FASTA (seq) file.

For help type:

```
./goose-findnpos-h
```

In the following subsections, we explain the input and output paramters.

#### Input parameters

The goose-findness program needs two streams for the computation, namely the input and output standard. The input stream is a FASTA file or a sequence.

```
Usage: ./goose-findnpos [options] [[--] args]
  or: ./goose-findnpos [options]

It reports the 'N' regions in a sequence or FASTA (seq) file.
```

```
> ABOOO 264 | acc = ABOOO 264 | descr = Homo sapiens mRNA
NCNNNACGGCCTCCTGCTGCTGCTCCTCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCCATTGTCCCC
GNCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTNGTTTGAGTGGACCTCCGGGCCCCTCATAGGAGAGGCAAGCTCGGGAGGTGCCAGGCGAGAGCAGGCCAGTGCC
GCGAATCCGCGCGCGCGGGACAGATCTCCTGCAAAGCCCTGCAGGAACNTCTTCTGGAAGACCTTCTCCACCCCCCAGC
TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAN
```

#### Output

The output of the goose-findness program is a structured report of "N" appearances in the sequence or FASTA file. The first column is the first position of the "N" appearance, the second is the position of the last "N" in the interval found, and the last column is the count of "N" in this interval.

An example, for the input, is:

```
1 1 1
3 5 3
82 82 1
163 163 1
289 289 1
```

## 3.9 Program goose-seq2fasta

The goose-seq2fasta converts a genomic sequence to pseudo FASTA file format.

For help type:

```
./goose-seq2fasta-h
```

In the following subsections, we explain the input and output paramters.

#### Input parameters

The goose-seq2fasta program needs two streams for the computation, namely the input and output standard. The input stream is a sequence group file.

#### Output

The output of the goose-seq2fasta program is a pseudo FASTA file.

An example, using the size line as 80 and the read's header as "Seq2Fasta", for the input, is:

## 3.10 Program goose-splitreads

The goose-splitreads splits a Multi-FASTA file to multiple FASTA files.

For help type:

```
./goose-splitreads -h
```

In the following subsections, we explain the input and output paramters.

#### Input parameters

The goose-splitreads program needs one stream for the computation, namely the input standard. This input stream is a Multi-FASTA file.

The attribution is given according to:

An example on such an input file is:

#### Output

The output of the goose-splitreads program is a report summary of the execution, and the files created in the defined location.

An example, for the input, is:

```
1 : Splitting to file:./out1.fasta
2 : Splitting to file:./out2.fasta
```

## 3.11 Program goose-renamehumanheaders

The goose-renamehumanheaders changes the headers of FASTA or Multi-FASTA file to simple chrX by order, where X is the number.

For help type:

```
./goose-renamehumanheaders -h
```

In the following subsections, we explain the input and output paramters.

#### Input parameters

The goose-renamehumanheaders program needs two streams for the computation, namely the input and output standard. The input stream is a FASTA or Multi-FASTA file.

The attribution is given according to:

```
Usage: ./goose-renamehumanheaders [options] [[--] args]
or: ./goose-renamehumanheaders [options]

It changes the headers of FASTA or Multi-FASTA file to simple chr$1 by order.

-h, --help show this help message and exit

Basic options
< input.fasta Input FASTA or Multi-FASTA file format (stdin)
> output.fasta Output FASTA or Multi-FASTA file format (stdout)

Example: ./goose-renamehumanheaders < input.fasta > output.fasta
```

An example on such an input file is:

```
> ABOOO264 | acc = ABOOO264 | descr = Homo sapiens mRNA
ACAAGACGGCCTCCTGCTGCTGCTGCTCCCGGGGCCCACGGCCCTGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGCCCCTCATAGGAAGGCAGGAAGCTCGGGAGGTGGCCAGGCGAGGAAGCAGGCCAGTGCC
GCGAATCCGCGCGCCCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCACCCCCCCAGC
TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA

> ABOOO263 | acc = ABOOO263 | descr = Homo sapiens mRNA
ACAAGATGCCATTGTCCCCCGGGCCTCCTGCTGCTGCTCTCCCGGGGCCACCGCCACCGCTGCCCTTGCCCCTGGAGGGT
GGCCCCACCGGCCGAGACAGCATATGCAGGAAGCGGCAGGAATAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTG
GTGGTTTGAGTGGACCTCCCAGGCCAGTGCCGGGCCCCTCATAGGAGAGCTCCGGGAAGACCTTCTCCTCCTCCTGCAAA
TAAAACCTCACCCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
```

### Output

The output of the goose-renamehumanheaders program is a FASTA or Multi-FASTA file. An example, for the input, is:

#### >chr1

ACAAGACGCCTCCTGCTGCTGCTGCTGCTCCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGCCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGAGAACCTTCTCCACCCCCAGC
GCGAATCCGCGCGCGCGCGGACAGATCTCCTGCAAAGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCACCCCCCAGC
TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA

#### > ch r2

ACAAGATGCCATTGTCCCCCGGCCTCCTGCTGCTGCTGCTCCTCCCGGGGCCACGGCCACCGCTGCCCTGCCCTGGAGGGT
GGCCCCACCGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAATAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTG
GTGGTTTGAGTGGACCTCCCAGGCCAGTGCCGGGCCCCTCATAGGAGAGCTCGGGAAGCTCCTGGCAGGCGGCAGGAAG
GCGCACCCCCCAGCAATCCGCGCGCCGGGACAGATGCCCTGCAGGAACTTCTTCTTCTGGAAGACCTTCTCCTCCTGCAAA
TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA

# Chapter 4

# Genomic sequence tools

Current available genomic sequence tools, for analysis and manipulation, are:

- 1. goose-genrandomdna: it generates a synthetic DNA.
- goose-randseqextrachars: it substitues in the DNA sequence the outside ACGT chars by random ACGT symbols.

## 4.1 Program goose-genrandomdna

The goose-genrandomdna generates a synthetic DNA.

For help type:

```
./goose-genrandomdna-h
```

In the following subsections, we explain the input and output paramters.

#### Input parameters

The goose-genrandomdna program needs one stream for the computation, namely the output standard. The attribution is given according to:

```
Usage: ./goose-genrandomdna [options] [[--] args]
or: ./goose-genrandomdna [options]

It generates a synthetic DNA.

-h, --help show this help message and exit

Basic options
> output.seq Output synthetic DNA sequence (stdout)
-s, --seed=<int> Starting point to the random generator (Default 0)
-n, --nSymbols=<int> Number of symbols generated (Default 100)
-f, --frequency=<str> The frequency of each base. It should be represented in the following format: <fa,fc,fg,ft>.
```

```
Example: ./goose-genrandomdna > output.seq
```

#### Output

The output of the goose-genrandomdna program is a sequence group file whith the synthetic DNA. Using the seed value as 1 and the number of symbols as 400, an example of an execution, is:

## 4.2 Program goose-randseqextrachars

The goose-randseqextrachars substitues in the DNA sequence the outside ACGT chars by random ACGT symbols. It works in sequence file formats.

For help type:

```
./goose-randseqextrachars-h
```

In the following subsections, we explain the input and output paramters.

#### Input parameters

The goose-randseqextrachars program needs two streams for the computation, namely the input and output standard. The input stream is a sequence file.

The attribution is given according to:

```
Usage: ./goose-randseqextrachars [options] [[--] args]
or: ./goose-randseqextrachars [options]

It substitues in the DNA sequence the outside ACGT chars by random ACGT symbols.

It works in sequence file formats

-h, --help show this help message and exit

Basic options
<input.seq Input sequence file (stdin)
> output.seq Output sequence file (stdout)

Example: ./goose-randseqextrachars < input.seq > output.seq
```

An example on such an input file is:

#### Output

The output of the goose-randseqextrachars program is a sequence file. An example, for the input, is:

# Chapter 5

# Amino acid sequence tools

Current available amino acid sequence tools, for analysis and manipulation, are:

- 1. goose-AminoAcidToGroup: it converts an amino acid sequence to a group sequence.
- 2. goose-ProteinToPseudoDNA: it converts an amino acid (protein) sequence to a pseudo DNA sequence.

## 5.1 Program goose-AminoAcidToGroup

The goose-AminoAcidToGroup converts an amino acid sequence to a group sequence. For help type:

```
./goose-AminoAcidToGroup-h
```

In the following subsections, we explain the input and output paramters.

#### Input parameters

The goose-AminoAcidToGroup program needs two streams for the computation, namely the input and output standard. The input stream is an amino acid sequence. The attribution is given according to:

```
Usage: ./goose-AminoAcidToGroup [options] [[--] args]
or: ./goose-AminoAcidToGroup [options]

It converts a amino acid sequence to a group sequence.

-h, --help show this help message and exit

Basic options
<input.prot Input amino acid sequence file (stdin)
> output.group Output group sequence file (stdout)

Example: ./goose-AminoAcidToGroup < input.prot > output.group

Table:
Prot Group
R P
```

```
Amino acids with electric charged side chains: POSITIVE
Н
K
D
    N
Ε
   N Amino acids with electric charged side chains: NEGATIVE
S
Т
N
   U Amino acids with electric UNCHARGED side chains
Q
    U
C
   S
U
G
   S Special cases
Р
    Н
   Н
V
Ι
L
М
   H Amino acids with hydrophobic side chains
F
   Н
Y
   Н
   Н
    * Others
   X Unknown
```

It can be used to group amino acids by properties, such as electric charge (positive and negative), uncharged side chains, hydrophobic side chains and special cases. An example on such an input file is:

```
IPFLLKKQFALADKLVLSKLRQLLGGRIKMMPCGGAKLEPAIGLFFHAIGINIKLGYGMTETTATVSCWHDFQFNPNSIG
TLMPKAEVKIGENNEILVRGGMVMKGYYKKPEETAQAFTEDGFLKTGDAGEFDEQGNLFITDRIKELMKTSNGKYIAPQY
IESKIGKDKFIEQIAIIADAKKYVSALIVPCFDSLEEYAKQLNIKYHDRLELLKNSDILKMFE
```

#### Output

The output of the goose-AminoAcidToGroup program is a group sequence.

An example, for the input, is:

HSHHHPPUHHHHNPHHHUPHPUHHSSPHPHHSSSSHPHNSHHSHHPHHSHUHPHSHSHUNUUHUHUSHPNHUHUSUUHS UHHSPHNHPHSNUUNHHHPSSHHHPSHHPPSNNUHUHHUNNSHHPUSNHSNHNNUSUHHHUNPHPNHHPUUUSPHHHSUH HNUPHSPNPHHNUHHHHHNHPPHHUHHHHSSHNUHNNHHPUHUHPHPNPHNHHPUUNHHPHHN

## 5.2 Program goose-ProteinToPseudoDNA

The goose-ProteinToPseudoDNA converts an amino acid (protein) sequence to a pseudo DNA sequence. For help type:

```
./goose-ProteinToPseudoDNA -h
```

In the following subsections, we explain the input and output paramters.

#### Input parameters

The goose-ProteinToPseudoDNA program needs two streams for the computation, namely the input and output standard. The input stream is an amino acid sequence. The attribution is given according to:

```
Usage: ./goose-ProteinToPseudoDNA [options] [[--] args]
   or: ./goose-ProteinToPseudoDNA [options]
It converts a protein sequence to a pseudo DNA sequence.
   -h, --help show this help message and exit
Basic options
   < input.prot</pre>
                    Input amino acid sequence file (stdin)
   > output.dna
                    Output DNA sequence file (stdout)
Example: ./goose-ProteinToPseudoDNA < input.prot > output.dna
Table:
Prot
       DNA
Α
   GCA
С
   TGC
D
   GAC
Ε
   GAG
   TTT
F
G
   GGC
   CAT
Н
Ι
   ATC
K
   AAA
   CTG
L
М
   ATG
N
   AAC
P
   CCG
Q
   CAG
R
   CGT
   TCT
T
   ACG
V
   GTA
W
   TGG
Y
   TAC
   TAG
   GGG
```

It can be used to generate pseudo-DNA with characteristics passed by amino acid (protein) sequences. An example on such an input file is:

```
IPFLLKKQFALADKLVLSKLRQLLGGRIKMMPCGGAKLEPAIGLFFHAIGINIKLGYGMTETTATVSCWHDFQFNPNSIG
TLMPKAEVKIGENNEILVRGGMVMKGYYKKPEETAQAFTEDGFLKTGDAGEFDEQGNLFITDRIKELMKTSNGKYIAPQY
IESKIGKDKFIEQIAIIADAKKYVSALIVPCFDSLEEYAKQLNIKYHDRLELLKNSDILKMFE
```

## Output

The output of the goose-ProteinToPseudoDNA program is a DNA sequence. An example, for the input, is:

## Chapter 6

# General purpose tools

- 1. goose-char2line: it splits a sequence into lines, creating an output sequence which has a char for each line.
- 2. goose-reverse: it reverses the order of a sequence.
- 3. goose-newlineonnews: it splits different rows with a new empty row.

## 6.1 Program goose-char2line

The goose-char2line splits a sequence into lines, creating an output sequence which has a char for each line.

For help type:

```
./goose-char2line -h
```

In the following subsections, we explain the input and output paramters.

#### Input parameters

The goose-char2line program needs two streams for the computation, namely the input and output standard. The input stream is a sequence file.

```
Example: ./goose-char2line < input.seq > output.seq
```

#### Output

The output of the goose-char2line program is a group sequence splited by \n foreach character. An example, for the input, is:

```
A
С
С
G
G
С
С
Т
С
С
T
G
С
Т
G
С
T
```

## 6.2 Program goose-reverse

The goose-reverse reverses the order of a sequence file.

For help type:

```
./goose-reverse -h
```

In the following subsections, we explain the input and output paramters.

#### Input parameters

The goose-reverse program needs two streams for the computation, namely the input and output standard. The input stream is a sequence file.

The attribution is given according to:

An example on such an input file is:

ACAAGACGGCCTCCTGCTGCTGCTGCTCCCGGGGCCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGCCCCTCATAGGAGAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGTGCC
GCGAATCCGCGCCCCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCACCCCCCCAGC
TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAAACAAGATGCCATTGTCCCCCGGCCTCCTGCTG
CTGCTGCTCTCCGGGGCCACGGCCACCGCTGCCCCTGCCCTTGGTGGTTTGAGTGGACACCTCCCAGGCCAGTGCCG
GGAAGCGGCAGGAATAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGTGGTTTTGAGTGGACCTCCCAGGCCAGTGCCG
GGCCCCTCATAGGAAGAGCTCGGGAGGTGGCCAGGCGAGAATCCGCCCCCCCAGGAC
AGAATGCCCTGCAGGAACTTCTTCTTGGAAGACCTTCCTCCTCCTCCTCCAAATAAAACCTCACCCATGAATCCACCCAAGTT
TAATTACAGACCTGAA

#### Output

The output of the goose-reverse program is a group sequence.

An example, for the input, is:

## 6.3 Program goose-newlineonnewx

The goose-newlineonnewx splits different rows with a new empty row.

For help type:

```
./goose-newlineonnewx -h
```

In the following subsections, we explain the input and output paramters.

#### Input parameters

The goose-newlineonnews program needs two streams for the computation, namely the input and output standard. The input stream is a matrix file format with 3 columns.

The attribution is given according to:

```
Usage: ./goose-newlineonnewx [options] [[--] args]
or: ./goose-newlineonnewx [options]

It splits different rows with a new empty row.

-h, --help show this help message and exit

Basic options
<input Input file with 3 column matrix format (stdin)
> output Output file with 3 column matrix format (stdout)

Example: ./goose-newlineonnewx < input > output
```

An example on such an input file is:

```
      1
      2
      2

      1
      2
      2

      4
      4
      1

      10
      12
      2

      15
      15
      1

      45
      47
      3

      45
      47
      3

      45
      47
      3

      45
      47
      3

      55
      55
      1
```

#### Output

The output of the goose-newlineonnews program is a 3 column matrix, with an empty line between different rows.

An example, for the input, is:

1.000000	2.000000	2.000000
4.000000	4.000000	1.000000
10.000000	12.000000	2.000000
15.000000	15.000000	1.000000
45.000000	47.000000	3.000000
45.000000 45.000000	47.000000 47.000000	3.000000
45.000000	47.000000	3.000000
55.000000	55.000000	1.000000