

# The Genomics Toolkit

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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 [?]. The storage of genomic data is being addressed using specific file formats, such as FASTQ and FASTA. Therefore, its analysis and manipulation is crucial [?]. Many frameworks for analysis and manipulation emerged, namely GALAXY [?], GATK [?], HTSeq [?], MEGA [?], among others. Several of these frameworks require licenses, while others do not provide a low level access to the information, since they are commonly approached by scripting or programming laguages not efficient for the purpose. Moreover, several lack on variety, namely the ability to perform multiple tasks using only one toolkit.

We describe GTO, a complete toolkit for genomics, namely for FASTA-FASTQ formats and sequences (DNA, amino acids, text), with many complementary tools. The toolkit is for Linux- and Unix-based systems, built for ultra-fast computations. GTO supports pipes for easy integration with the sub-programs belonging to GTO as well as external tools. GTO works as the *LEGOs*, since it allows the construction of multiple pipelines with many combinations.

GTO includes tools for information display, randomization, edition, conversion, extraction, search, calculation, and visualization. GTO is prepared to deal with very large datasets, typically in the scale Gigabytes or Terabytes (but not limited).

The complete toolkit is an optimized command line version, using the prefix "gto\_" followed by the suffix with the respective name of the program. GTO is implemented in C language and it is available, under the MIT license, at:

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

#### 1.1 Installation

For GTO installation, run:

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

## 1.2 License

The license is MIT. In resume, it is a short and simple permissive license with conditions only requiring preservation of copyright and license notices. Licensed works, modifications, and larger works may be distributed under different terms and without source code.

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# Chapter 2

# Amino acid sequence tools

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

- 1. gto\_amino\_acid\_to\_group: it converts an amino acid sequence to a group sequence.
- 2. gto\_amino\_acid\_to\_pseudo\_dna: it converts an amino acid (protein) sequence to a pseudo DNA sequence.

# 2.1 Program gto amino acid to group

The gto\_amino\_acid\_to\_group converts an amino acid sequence to a group sequence.

For help type:

```
./gto_amino_acid_to_group -h
```

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

## Input parameters

The gto\_amino\_acid\_to\_group 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:

```
Р
           Amino acids with electric charged side chains: POSITIVE
        Р
        N
           Amino acids with electric charged side chains: NEGATIVE
S
        IJ
        П
          Amino acids with electric UNCHARGED side chains
U
        S
G
        S Special cases
Р
        S
        Н
Ι
        н
        Н
        H Amino acids with hydrophobic side chains
        Н
Y
        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 gto\_amino\_acid\_to\_group program is a group sequence.

An example, for the input, is:

HSHHHPPUHHHHNPHHHUPHPUHHSSPHPHHSSSSHPHNSHHSHHHPHHSHUHPHSHSHUNUUHUHUSHPNHUHUSUUHS UHHSPHNHPHSNUUNHHHPSSHHHPSHHPPSNNUHUHHUNNSHHPUSNHSNHNNUSUHHHUNPHPNHHPUUUSPHHHSUH HNUPHSPNPHHNUHHHHHNHPPHHUHHHHHSSHNUHNNHHPUHUHPHPNPHNHHPUUNHHPHHN

# 2.2 Program gto amino acid to pseudo dna

The gto\_amino\_acid\_to\_pseudo\_dna converts an amino acid (protein) sequence to a pseudo DNA sequence.

For help type:

```
./gto_amino_acid_to_pseudo_dna -h
```

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

### Input parameters

The gto\_amino\_acid\_to\_pseudo\_dna 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: ./gto_amino_acid_to_pseudo_dna [options] [[--] args]
   or: ./gto_amino_acid_to_pseudo_dna [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: ./gto_amino_acid_to_pseudo_dna < input.prot > output.dna
Prot
        DNA
         GCA
С
         TGC
         GAC
D
Ε
         GAG
F
         T\,T\,T
G
         \tt G\,G\,C
Н
         CAT
Ι
         ATC
K
         A\ A\ A
L
         CTG
         ATG
N
         AAC
P
         CCG
Q
         CAG
        CGT
R
S
         TCT
T
         A \; C \; G
V
         GTA
W
         TGG
Y
         TAC
         TAG
Х
         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 gto\_amino\_acid\_to\_pseudo\_dna program is a DNA sequence. An example, for the input, is:

# Chapter 3

# FASTQ tools

Current available tools for FASTQ format analysis and manipulation include:

- 1. gto\_fastq\_to\_fasta: it converts a FASTQ file format to a pseudo FASTA file.
- 2. gto\_fastq\_to\_mfasta: it converts a FASTQ file format to a pseudo Multi-FASTA file.
- 3. gto\_fastq\_exclude\_n: it discards the FASTQ reads with the minimum number of "N" symbols.
- 4. gto\_fastq\_extract\_quality\_scores: it extracts all the quality-scores from FASTQ reads.
- 5. gto\_fastq\_info: it analyses the basic information of FASTQ file format.
- 6. gto\_fastq\_maximum\_read\_size: it filters the FASTQ reads with the length higher than the value defined.
- 7. gto\_fastq\_minimum\_quality\_score: it discards reads with average quality-score below of the defined.
- 8. gto\_fastq\_minimum\_read\_size: it filters the FASTQ reads with the length smaller than the value defined.
- 9. gto\_fastq\_rand\_extra\_chars: it substitues in the FASTQ files, the DNA sequence the outside ACGT chars by random ACGT symbols.
- 10. gto\_fastq\_from\_seq: it converts a genomic sequence to pseudo FASTQ file format.
- 11. gto\_fastq\_mutate: it creates a synthetic mutation of a FASTQ file given specific rates of mutations, deletions and additions.
- 12. gto\_fastq\_split: it splits Paired End files according to the direction of the strand ('/1' or '/2').
- 13. gto\_fastq\_pack: it packages each FASTQ read in a single line.
- 14. gto\_fastq\_unpack: it unpacks the FASTQ reads packaged using the gto\_fastq\_pack tool.

- 15. gto\_fastq\_quality\_score\_info: it analyses the quality-scores of a FASTQ file.
- 16. gto\_fastq\_quality\_score\_min: it analyses the minimal quality-scores of a FASTQ file.
- 17. gto\_fastq\_quality\_score\_max: it analyses the maximal quality-scores of a FASTQ file.
- 18. gto\_fastq\_cut: it cuts read sequences in a FASTQ file.
- 19. gto\_fastq\_minimum\_local\_quality\_score\_forward: it filters the reads considering the quality score average of a defined window size of bases.
- 20. gto\_fastq\_minimum\_local\_quality\_score\_reverse: it filters the reverse reads, considering the average window size score defined by the bases.

# 3.1 Program gto fastq to fasta

The gto\_fastq\_to\_fasta 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:

```
./gto_fastq_to_fasta -h
```

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

#### Input parameters

The gto\_fastq\_to\_fasta 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: ./gto_fastq_to_fasta [options] [[--] args]
    or: ./gto_fastq_to_fasta [options]

It converts a FASTQ file format to a pseudo FASTA file.
It does NOT align the sequence.
It extracts the sequence and adds a pseudo header.

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

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

Example: ./gto_fastq_to_fasta < input.fastq > output.fasta
```

An example on such an input file is:

## Output

The output of the gto\_fastq\_to\_fasta program a FASTA file.

An example, for the input, is:

```
GGGTGATGGCCGCTGCCGATGGCGTCAAATCCCACCAAGTTACCCTTAACAACTTAAGGGTTTTCAAATAGA
GTTCAGGGATACGACGTTTGTATTTTAAGAATCTGAAGCAGAAGTCGATGATAATACGCGTCGTTTTATCAT
```

# 3.2 Program gto\_fastq\_to\_mfasta

The gto\_fastq\_to\_mfasta 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:

```
./gto_fastq_to_mfasta -h
```

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

#### Input parameters

The gto\_fastq\_to\_mfasta program needs two streams for the computation, namely the input and output standard. The input stream is a FASTQ file.

```
Usage: ./gto_fastq_to_mfasta [options] [[--] args]
    or: ./gto_fastq_to_mfasta [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: ./gto_fastq_to_mfasta < input.fastq > output.mfasta
```

### Output

The output of the gto\_fastq\_to\_mfasta program a Multi-FASTA file.

An example, for the input, is:

```
>SRR001666.1 071112_SLXA-EAS1_s_7:5:1:817:345 length=72

GGGTGATGGCCGCTGCCGATGGCGTCAAATCCCACCAAGTTACCCTTAACAACTTAAGGGTTTTCAAATAGA
>SRR001666.2 071112_SLXA-EAS1_s_7:5:1:801:338 length=72

GTTCAGGGGATACGACGTTTGTATTTTAAGAATCTGAAGCAGAAGTCGATGATAATACGCGTCGTTTTATCAT
```

# 3.3 Program gto fastq exclude n

The gto\_fastq\_exclude\_n discards the FASTQ reads with the minimum number of "N" symbols. Also, if present, it will erase the second header (after +).

For help type:

```
./gto_fastq_exclude_n -h
```

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

## Input parameters

The gto\_fastq\_exclude\_n program needs two streams for the computation, namely the input and output standard. The input stream is a FASTQ file.

```
Usage: ./gto_fastq_exclude_n [options] [[--] args]
or: ./gto_fastq_exclude_n [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 Input FASTQ file format (stdin)
> output.fastq Output FASTQ file format (stdout)

Example: ./gto_fastq_exclude_n < input.fastq > output.fastq

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

## Output

The output of the gto\_fastq\_exclude\_n program is a set of all the filtered FASTQ reads, followed by the execution report. The execution report only appears in the console.

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

# $3.4 \quad Program\ gto\_fastq\_extract\ quality\ scores$

The  ${\tt gto\_fastq\_extract\_quality\_scores}$  extracts all the quality-scores from FASTQ reads.

For help type:

```
./gto_fastq_extract_quality_scores -h
```

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

#### Input parameters

The gto\_fastq\_extract\_quality\_scores program needs two streams for the computation, namely the input and output standard. The input stream is a FASTQ file.

```
Usage: ./gto_fastq_extract_quality_scores [options] [[--] args]
   or: ./gto_fastq_extract_quality_scores [options]
It extracts all the quality-scores from FASTQ reads.
    -h, --help
                         show this help message and exit
Basic options
   < input.fastq</pre>
                          Input FASTQ file format (stdin)
    > output.fastq
                          Output FASTQ file format (stdout)
Example: ./gto_fastq_extract_quality_scores < input.fastq > output.fastq
Console output example:
<FASTQ quality scores>
Total reads
                    : value
Total Quality-Scores : value
```

#### Output

The output of the gto\_fastq\_extract\_quality\_scores program is a set of all the quality scores from the FASTQ reads, followed by the execution report. The execution report only appears in the console.

An example, for the input, is:

# 3.5 Program gto\_fastq\_info

The gto\_fastq\_info analyses the basic information of FASTQ file format. For help type:

```
./gto_fastq_info -h
```

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

## Input parameters

The gto\_fastq\_info 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: ./gto_fastq_info [options] [[--] args]
  or: ./gto_fastq_info [options]
It analyses the basic information of FASTQ file format.
    -h, --help
                         show this help message and exit
Basic options
   < input.fastq</pre>
                        Input FASTQ file format (stdin)
   > output
                          Output read information (stdout)
Example: ./gto_fastq_info < 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
```

An example on such an input file is:

## Output

The output of the gto\_fastq\_info program is a set of information related to the file read. 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
```

# 3.6 Program gto fastq maximum read size

The gto\_fastq\_maximum\_read\_size filters the FASTQ reads with the length higher than the value defined. For help type:

```
./gto_fastq_maximum_read_size -h
```

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

### Input parameters

The gto\_fastq\_maximum\_read\_size 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: ./gto_fastq_maximum_read_size [options] [[--] args]
   or: ./gto_fastq_maximum_read_size [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</pre>
                        Input FASTQ file format (stdin)
   > output.fastq
                         Output FASTQ file format (stdout)
Example: ./gto_fastq_maximum_read_size -s <size> < input.fastq > output.fastq
Console 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 gto\_fastq\_maximum\_read\_size program is a set of all the filtered FASTQ reads, followed by the execution report. The execution report only appears in the console.

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

# 3.7 Program gto fastq minimum quality score

The gto\_fastq\_minimum\_quality\_score discards reads with average quality-score below of the defined. For help type:

```
./gto_fastq_minimum_quality_score -h
```

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

### Input parameters

The gto\_fastq\_minimum\_quality\_score 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: ./gto_fastq_minimum_quality_score [options] [[--] args]
   or: ./gto_fastq_minimum_quality_score [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</pre>
                          Input FASTQ file format (stdin)
                          Output FASTQ file format (stdout)
    > output.fastq
Example: ./gto_fastq_minimum_quality_score -m <min> < input.fastq > output.fastq
Console output example:
<FASTQ non-filtered reads>
Total reads : value
Filtered reads : value
```

An example on such an input file is:

```
+SRR001666.2 071112_SLXA-EAS1_s_7:5:1:801:338 length=72
54599<>77977==6=?I6IBI::33344235521677999>>><<<@@A@BBCDGGBFFH>IIIII-I)8I
```

### Output

The output of the gto\_fastq\_minimum\_quality\_score 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:

## 3.8 Program gto fastq minimum read size

The gto\_fastq\_minimum\_read\_size filters the FASTQ reads with the length smaller than the value defined. For help type:

```
./gto_fastq_minimum_read_size -h
```

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

#### Input parameters

The gto\_fastq\_minimum\_read\_size program needs two streams for the computation, namely the input and output standard. The input stream is a FASTQ file.

```
Usage: ./gto_fastq_minimum_read_size [options] [[--] args]
   or: ./gto_fastq_minimum_read_size [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.fastq
                          Output FASTQ file format (stdout)
Example: ./gto_fastq_minimum_read_size -s <size> < input.fastq > output.fastq
Console output example:
<FASTQ non-filtered reads>
Total reads : value
```

```
Filtered reads : value
```

### Output

The output of the gto\_fastq\_minimum\_read\_size program is a set of all the filtered FASTQ reads, followed by the execution report. The execution report only appears in the console.

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

# 3.9 Program gto\_fastq\_rand\_extra\_chars

The gto\_fastq\_rand\_extra\_chars substitues in the FASTQ files, the DNA sequence the outside ACGT chars by random ACGT symbols.

For help type:

```
./gto_fastq_rand_extra_chars -h
```

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

## Input parameters

The gto\_fastq\_rand\_extra\_chars program needs two streams for the computation, namely the input and output standard. The input stream is a FASTQ file.

```
Usage: ./gto_fastq_rand_extra_chars [options] [[--] args]
or: ./gto_fastq_rand_extra_chars [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
```

## Output

The output of the gto\_fastq\_rand\_extra\_chars program is a FASTQ file. An example, for the input, is:

# 3.10 Program gto fastq from seq

The gto\_fastq\_from\_seq converts a genomic sequence to pseudo FASTQ file format.

```
For help type:
```

```
./gto_fastq_from_seq -h
```

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

### Input parameters

The gto\_fastq\_from\_seq 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 gto\_fastq\_from\_seq program is a pseudo FASTQ file.

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

```
TA 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 T A A T T A C A G A C C T G A A A C A A G A T G C C A T T G T C C C C G G C C T C C T G C T G
@SeqToFastq6
GG A A GC GG C A GG A A T A A GG A A A A GC A G C C T C C T G A C T T T C C T C GC T T G G T G T T T G A G T G G A C C T C C C A G T G C C G
@SeqToFastq8
@SeqToFastq9
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 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
@SeqToFastq10
TAATTACAGACCTGAA
FFFFFFFFFFFFFF
```

# 3.11 Program gto fastq mutate

The gto\_fastq\_mutate 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:

```
./gto_fastq_mutate -h
```

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

#### Input parameters

The gto\_fastq\_mutate 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.

```
Usage: ./gto_fastq_mutate [options] [[--] args]
or: ./gto_fastq_mutate [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: ./gto_fastq_mutate -s <seed> -m <mutation rate> -d <deletion rate> -i
<insertion rate> -a < input.fastq > output.fastq
```

### Output

The output of the gto\_fastq\_mutate 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:

# 3.12 Program gto fastq split

The gto\_fastq\_split splits Paired End files according to the direction of the strand ( $^{\prime}/1^{\prime}$  or  $^{\prime}/2^{\prime}$ ). It writes by default singleton reads as forward stands.

For help type:

```
./gto_fastq_split -h
```

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

## Input parameters

The gto\_fastq\_split program needs a stream for the computation, namely the input standard. The input stream is a FASTQ file.

The attribution is given according to:

```
Usage: ./gto_fastq_split [options] [[--] args]
  or: ./gto_fastq_split [options]
It writes by default singleton reads as forward stands.
   -h, --help
                         show this help message and exit
Basic options
   -f, --forward=<str> Output forward file
   -r, --reverse=<str> Output reverse file
   < input.fastq</pre>
                         Input FASTQ file format (stdin)
   > output
                         Output read information (stdout)
Example: ./gto_fastq_split -t <output_forward.fastq> -r <output_reverse.fastq> < input.fastq > output
Output example :
Total reads : value
Singleton reads : value
Forward reads : value
Reverse reads : value
```

An example on such an input file is:

## Output

The output of the gto\_fastq\_split program is a set of information related to the file read. An example, for the input, is:

```
Total reads : 2
Singleton reads : 0
Forward reads : 65536
Reverse reads : 1
```

Also, this program generates two FASTQ files, with the reverse and forward reads.

An example of the forward reads, for the input, is:

## 3.13 Program gto fastq pack

The gto\_fastq\_pack packages each FASTQ read in a single line. It can show the read score first or the dna sequence, depending on the execution mode.

For help type:

```
./gto_fastq_pack -h
```

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

### Input parameters

The gto\_fastq\_pack 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 gto\_fastq\_pack program is a packaged FASTQ file.

An example, for the input, is:

Another example for the same input, but using the scores first (flag "s"), is:

# 3.14 Program gto fastq unpack

The gto\_fastq\_unpack unpacks the FASTQ reads packaged using the gto\_fastq\_pack tool. For help type:

```
./gto_fastq_unpack -h
```

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

#### Input parameters

The gto\_fastq\_unpack program needs two streams for the computation, namely the input and output standard. The input stream is a FASTQ file.

```
Example: ./gto_fastq_unpack -s < input.fastq > output.fastq
```

### Output

The output of the gto\_fastq\_unpack program is a packaged FASTQ file.

An example, for the input, is:

# 3.15 Program gto\_fastq\_quality\_score\_info

The gto\_fastq\_quality\_score\_info analyses the quality-scores of a FASTQ file.

For help type:

```
./gto_fastq_quality_score_info -h
```

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

#### Input parameters

The gto\_fastq\_quality\_score\_info program needs two streams for the computation, namely the input and output standard. The input stream is a FASTQ file.

```
Usage: ./gto_fastq_quality_score_info [options] [[--] args]
or: ./gto_fastq_quality_score_info [options]

It analyses the quality-scores of a FASTQ file.

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

Basic options
```

```
< input.fastq</pre>
                          Input FASTQ file format (stdin)
    > output
                          Output read information (stdout)
Optional
    -m, --max = < int >
                          The lenght of the maximum window
Example: ./gto_fastq_quality_score_info -m <max> < input.fastq > output
Output example :
Total reads
              : value
Max read length : value
Min read length : value
Min QS value : value
Max QS value : value
              : value
QS range
```

## Output

The output of the gto\_fastq\_quality\_score\_info program is a set of information related to the file read. Using the max window value as 30, an example for this input, is:

# 3.16 Program gto\_fastq\_quality\_score\_max

The gto\_fastq\_quality\_score\_max analyses the maximal quality-scores of a FASTQ file. For help type:

```
./gto_fastq_quality_score_max -h
```

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

## Input parameters

The gto\_fastq\_quality\_score\_max 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 gto\_fastq\_quality\_score\_max program is a set of information related to the file read, considering the maximal quality scores.

Using the max window value as 30, an example for this input, is:

# $3.17 \quad {\bf Program~gto\_fastq\_quality\_score\_min}$

The gto\_fastq\_quality\_score\_min analyses the minimal quality-scores of a FASTQ file. For help type:

```
./gto_fastq_quality_score_min -h
```

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

### Input parameters

The gto\_fastq\_quality\_score\_min 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 gto\_fastq\_quality\_score\_min program is a set of information related to the file read, considering the minimum quality scores.

Using the max window value as 30, an example for this input, is:

# 3.18 Program gto fastq cut

The gto\_fastq\_cut cuts read sequences in a FASTQ file. It requires that the initial and end positions for the cut.

For help type:

```
./gto_fastq_cut -h
```

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

## Input parameters

The gto\_fastq\_cut program needs 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 gto\_fastq\_cut program is a FASTQ file cut.

Using the initial value as 10 and the end value as 30, an example for this input, is:

```
@SRR001666.1 071112_SLXA - EAS1_s_7:5:1:817:345 length=72
CGCTGCCGATGGCGTCAAATC
+
IIIIIIIIIIIIIIIIIII
@SRR001666.2 071112_SLXA - EAS1_s_7:5:1:801:338 length=72
ACGACGTTTGTATTTTAAGAA
+
IIIIIIIIIIIIIIIIIIII
```

## 3.19 Program gto fastq minimum local quality score forward

The gto\_fastq\_minimum\_local\_quality\_score\_forward filters the reads considering the quality score average of a defined window size of bases.

For help type:

```
./gto_fastq_minimum_local_quality_score_forward -h
```

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

### Input parameters

The gto\_fastq\_minimum\_local\_quality\_score\_forward program needs program needs two streams for the computation, namely the input and output standard. The input stream is a FASTQ file.

```
Usage: ./gto_fastq_minimum_local_quality_score_forward [options] [[--] args]
   or: ./gto_fastq_minimum_local_quality_score_forward [options]
It filters the reads considering the quality score average of a defined window size
of bases.
   -h, --help
                             show this help message and exit
Basic options
   -k, --windowsize = <int> The window size of bases (default 5)
   -w, --minavg=<int> The minimum average of quality score (default 25)
   -m, --minqs=<int>
                            The minimum value of the quality score (default 33)
   < input.fastq</pre>
                            Input FASTQ file format (stdin)
                             Output FASTQ file format (stdout)
   > output.fastq
Example: ./gto_fastq_minimum_local_quality_score_forward -k <windowsize> -w <minavg>
-m <minqs> < input.fastq > output.fastq
Console output example:
Minimum QS
            : value
<FASTQ output>
Total reads
            : value
Trimmed reads : value
```

## Output

The output of the gto\_fastq\_minimum\_local\_quality\_score\_forward program is a FASTQ file with the reads filtered following a quality score average of a defined window of bases. The execution report only appears in the console.

An example using the default values, for the input, is:

# 3.20 Program gto\_fastq\_minimum\_local\_quality\_score\_reverse

The gto\_fastq\_minimum\_local\_quality\_score\_reverse filters the reverse reads, considering the quality score average of a defined window size of bases.

For help type:

```
./gto_fastq_minimum_local_quality_score_reverse -h
```

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

#### Input parameters

The gto\_fastq\_minimum\_local\_quality\_score\_reverse program needs program needs two streams for the computation, namely the input and output standard. The input stream is a FASTQ file.

```
Usage: ./gto_fastq_minimum_local_quality_score_reverse [options] [[--] args]
   or: ./gto_fastq_minimum_local_quality_score_reverse [options]
It filters the reverse reads, considering the quality score average of a defined
window size of bases.
   -h, --help
                             show this help message and exit
Basic options
   -k, --windowsize = <int> The window size of bases (default 5)
   -w, --minavg=<int>
                            The minimum average of quality score (default 25)
   -m, --minqs=<int>
                            The minimum value of the quality score (default 33)
   < input.fastq</pre>
                             Input FASTQ file format (stdin)
                             Output FASTQ file format (stdout)
   > output.fastq
Example: ./gto_fastq_minimum_local_quality_score_reverse -k <windowsize> -w <minavg>
-m <minqs> < input.fastq > output.fastq
Console output example:
Minimum QS
             : value
<FASTQ output>
Total reads
            : value
Trimmed reads : value
```

#### Output

The output of the gto\_fastq\_minimum\_local\_quality\_score\_reverse program is a FASTQ file with the reads filtered following a quality score average of a defined window of bases. The execution report only appears in the console.

An example using the default values, for the input, is:

# Chapter 4

# FASTA tools

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

- 1. gto\_fasta\_to\_seq: it converts a FASTA or Multi-FASTA file format to a seq.
- 2. gto\_fasta\_from\_seq: it converts a genomic sequence to pseudo FASTA file format.
- 3. gto\_fasta\_extract: it extracts sequences from a FASTA file, which the range is defined by the user in the parameters.
- 4. gto\_fasta\_extract\_by\_read: 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.
- 5. gto\_fasta\_info: it shows the readed information of a FASTA or Multi-FASTA file format.
- 6. gto\_fasta\_mutate: it reates a synthetic mutation of a fasta file given specific rates of editions, deletions and additions.
- 7. gto\_fasta\_rand\_extra\_chars: it substitues in the DNA sequence the outside ACGT chars by random ACGT symbols.
- 8. gto\_fasta\_extract\_read\_by\_pattern: it extracts reads from a Multi-FASTA file format given a pattern in the header.
- 9. gto\_fasta\_find\_n\_pos: it reports the "N" regions in a sequence or FASTA (seq) file.
- 10. gto\_fasta\_split\_reads: it splits a Multi-FASTA file to multiple FASTA files.
- 11. gto\_fasta\_rename\_human\_headers: it changes the headers of FASTA or Multi-FASTA file to simple chrX by order, where X is the number.

# 4.1 Program gto\_fasta\_to\_seq

The gto\_fasta\_to\_seq converts a FASTA or Multi-FASTA file format to a sequence. For help type:

```
./gto_fasta_to_seq -h
```

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

#### Input parameters

The gto\_fasta\_to\_seq 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 gto\_fasta\_to\_seq program is a group sequence.

An example, for the input, is:

 

### 4.2 Program gto fasta from seq

The gto\_fasta\_from\_seq converts a genomic sequence to pseudo FASTA file format. For help type:

```
./gto_fasta_from_seq -h
```

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

#### Input parameters

The gto\_fasta\_from\_seq program needs two streams for the computation, namely the input and output standard. The input stream is a sequence group file.

The attribution is given according to:

An example on such an input file is:

#### Output

The output of the gto\_fasta\_from\_seq program is a pseudo FASTA file.

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

### 4.3 Program gto fasta extract

The gto\_fasta\_extract extracts sequences from a FASTA file, which the range is defined by the user in the parameters.

For help type:

```
./gto_fasta_extract -h
```

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

#### Input parameters

The gto\_fasta\_extract 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.

```
> ABOOO 264 | acc = ABOOO 264 | descr = Homo sapiens mRNA
ACAAGACGGCCTCCTGCTGCTGCTGCTCCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGCCCCTCATAGGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGTGCC
GCGAATCCGCGCGCCCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCACCCCCCCAGC
TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
```

#### Output

The output of the gto\_fasta\_extract 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, is:

ACAAGACGGCCTCCTGCTGCTGCTCTCCGGGGCCACGGCCCTGGAGG

### 4.4 Program gto fasta extract by read

The gto\_fasta\_extract\_by\_read extracts sequences from a FASTA or Multi-FASTA file, which the range is defined by the user in the parameters.

For help type:

```
./gto_fasta_extract_by_read -h
```

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

#### Input parameters

The gto\_fasta\_extract\_by\_read 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.

```
Usage: ./gto_fasta_extract_by_read [options] [[--] args]
or: ./gto_fasta_extract_by_read [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: ./gto_fasta_extract_by_read -i <init> -e <end> < input.fasta > output.fasta
```

#### Output

The output of the gto\_fasta\_extract\_by\_read 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
```

### 4.5 Program gto\_fasta\_info

The gto\_fasta\_info shows the readed information of a FASTA or Multi-FASTA file format.

For help type:

```
./gto_fasta_info -h
```

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

#### Input parameters

The gto\_fasta\_info program needs two streams for the computation, namely the input and output standard. The input stream is a FASTA or Multi-FASTA file.

```
Usage: ./gto_fasta_info [options] [[--] args]
  or: ./gto_fasta_info [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: ./gto_fasta_info < 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
```

```
> ABOOO 264 | acc = ABOOO 264 | descr = Homo sapiens mRNA

ACAAGACGGCCTCCTGCTGCTGCTGCTCCCGGGGCCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC

GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA

GTGGTTTGAGTGGACCTCCGGGCCCCTCATAGGAGAGAGCTCGGGAGGTGGCCAGGCCAGGAAGCAGGCCAGTGCC

GCGAATCCGCGCGCGCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCACCCCCCCAGC

TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA

> ABOOO 263 | acc = ABOOO 263 | descr = Homo sapiens mRNA

ACAAGATGCCATTGTCCCCCGGGCCTCCTGCTGCTGCTGCTCTCCGGGGCCACGGCCACCGCTGCCCTTGCCCCTGGAGGGT

GGCCCCACCGGCCGAGACAGCGAACAGCAATTCCAGGAAAGCAGCTCCTGGAAAAAACCTCCACCCCCCAGGCCACCGCTGCCCTTGACTTTCCTCGCTTG

GTGGTTTGAGTGGACCTCCCAGGCCAGTGCCGGGCCCCTCATAGGAAAACCTCGGGAAGACCTTCTCCTCCTCCTGCAAA

TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
```

#### Output

The output of the gto\_fasta\_info program is a set of information related to the file read. 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
```

### 4.6 Program gto fasta mutate

The gto\_fasta\_mutate 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:

```
./gto_fasta_mutate -h
```

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

#### Input parameters

The gto\_fasta\_mutate 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.

The attribution is given according to:

```
Usage: ./gto_fasta_mutate [options] [[--] args]
   or: ./gto_fasta_mutate [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
                                 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: ./gto_fasta_mutate -s <seed> -e <edit rate> -d <deletion rate> -i
<insertion rate> -a < input.fasta > output.fasta
```

An example on such an input file is:

#### Output

The output of the gto\_fasta\_mutate 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

TNGGTCGGCNGGNNCCTCCGGNAACGGCACCGGAGACCGAAGCTCGGNGGNTATACAGGTGTCANGAAACATCCCCGCGNC

GNGTGNCCNNGAANCCANAGAGTATCTCACTCACAACCCTGCGTGCACNTCTAGAGNANGACCTTACNCACCNTCCCNTT

NNGTACCACACCAATGAACGCTGCAGAAAGTCTGTTTNNAGGNGNGCA

> ABOOO 263 | acc = ABOOO 263 | descr = Homo sapiens mRNA

ATTTGAAGGCAANCGGNCCAGNAATNCGGNGGGTGCNGCTCNTGTNGGCTACGGGNCATCGCGGCCCTGCTNTANTAAGCN

TGAACCACCGNTCGNNGCACTTAGCAATNGCGNAANCCGTCGGCACGGCGAGACNAANCCGCTANTNNTTTCCCGGCTNA

ATGGNTGTACAAGACCNACTANACCANCCTCCGTCACCACCTGGAGCGCANGATGGNNCGCTGNCTAGNAGNCNNTGAG

GCGCTCCNTCCTANAAANCCGTGGNCGAGCNCCCTATGGNAGNGTGGGGGTTTTTACCGGGAAGACCNTCGNGCCCTATGGG

AGCAATCANAANCTAGAAAGCTTACNGATGGTGANGAANTAGACTANG
```

### 4.7 Program gto fasta rand extra chars

The gto\_fasta\_rand\_extra\_chars 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:

```
./gto_fasta_rand_extra_chars -h
```

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

#### Input parameters

The gto\_fasta\_rand\_extra\_chars program needs two streams for the computation, namely the input and output standard. The input stream is a FASTA or Multi-FASTA file.

#### Output

The output of the gto\_fasta\_rand\_extra\_chars program is a FASTA or Multi-FASTA file. An example, for the input, is:

### 4.8 Program gto fasta extract read by pattern

The gto\_fasta\_extract\_read\_by\_pattern extracts reads from a Multi-FASTA file format given a pattern in the header. Also, this pattern is case insensitive.

For help type:

```
./gto_fasta_extract_read_by_pattern -h
```

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

#### Input parameters

The gto\_fasta\_extract\_read\_by\_pattern program needs two streams for the computation, namely the input and output standard. The input stream is a Multi-FASTA file.

```
> ABOOO 264 | acc = ABOOO 264 | descr = Homo sapiens mRNA

ACAAGACGGCCTCCTGCTGCTGCTGCTCCTCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC

GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA

GTGGTTTGAGTGGACCTCCGGGCCCCTCATAGGAGAGGAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGTGCC

GCGAATCCGCGCGCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACCTTCTTCTGGAAGACCTTCTCCCACCCCCCAGC

TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA

> ABOOO 263 | acc = ABOOO 263 | descr = Homo sapiens mRNA

ACAAGATGCCATTGTCCCCCGGCCTCCTGCTGCTGCTGCTCCTCCGGGGCCCACGGCCACCGCTGCCCTGCCCTGGAGGGT

GGCCCCACCGGCCGAGACAGCGAGCATATGCAGGAAGCGGCCAGGAAATAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTG

GTGGTTTGAGTGGACCTCCCAGGCCAGTGCCGGGCCCCTCATAGGAGAGGAAGCTCCTGGGAGGTGGCCAGGCGGCAGGAAG

TAAAACCTCACCCCATGAATCCGCGCGCCCGGGACAGAATGCCCTGCAAA
```

#### Output

The output of the gto\_fasta\_extract\_read\_by\_pattern program is a Multi-FASTA file. An example, using the pattern "264", for the provided input, is:

```
> ABOOO 264 | acc = ABOOO 264 | descr = Homo sapiens mRNA
ACAAGACGGCCTCCTGCTGCTGCTCCTCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGCCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGAGGAAGCAGCCAGTGCC
GCGAATCCGCGCGCGCGCGGACAGATCTCCTGCAAAGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCCACCCCCCAGC
TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
```

### 4.9 Program gto\_fasta\_find\_n\_pos

The gto\_fasta\_find\_n\_pos reports the "N" regions in a sequence or FASTA (seq) file. For help type:

```
./gto_fasta_find_n_pos -h
```

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

#### Input parameters

The gto\_fasta\_find\_n\_pos program needs two streams for the computation, namely the input and output standard. The input stream is a FASTA file or a sequence.

The attribution is given according to:

An example on such an input file is:

#### Output

The output of the gto\_fasta\_find\_n\_pos 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 1 3 5 3 82 82 1 163 163 1 289 289 1
```

### 4.10 Program gto\_fasta\_split\_reads

The gto\_fasta\_split\_reads splits a Multi-FASTA file to multiple FASTA files. For help type:

```
./gto_fasta_split_reads -h
```

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

#### Input parameters

The gto\_fasta\_split\_reads 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:

```
> ABOOO 264 | acc = ABOOO 264 | descr = Homo sapiens mRNA

ACAAGACGGCCTCCTGCTGCTGCTCTCCCGGGGCCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC

GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA

GTGGTTTGAGTGGACCTCCGGGCCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCCAGGAAGCAGGCCAGGCCAGGCC

GCGAATCCGCGCGCCGGGACAGATCTCCTGCAAAGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCACCCCCCCAGC

TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA

> ABOOO 263 | acc = ABOOO 263 | descr = Homo sapiens mRNA

ACAAGATGCCATTGTCCCCCGGGCCTCCTGCTGCTGCTGCTCTCCGGGGCCACGGCCACCGCTGCCCTGCCCCTGGAGGGT

GGCCCCACCGGCCGAGACAGCCATTTCCAGGAAGCCGCAGGAAAAAGCAGCCTCCTGACTTTCCTCCCTTG

GTGGTTTGAGTGGACCTCCCAGGCCAGTGCCGGGCCCCTCATAGGAAAAGCAGCTCCGGGAGGTGGCCAGGCGCAGGAAG

GCGCACCCCCCCAGCAATCCGCGCGCCGGGACAGAATGCCCTGCAGGAACTTCTTCTTCTGGAAGACCTTCTCCTCCTGCAAA

TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
```

#### Output

The output of the gto\_fasta\_split\_reads 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
```

### 4.11 Program gto fasta rename human headers

The gto\_fasta\_rename\_human\_headers changes the headers of FASTA or Multi-FASTA file to simple chrX by order, where X is the number.

For help type:

```
./gto_fasta_rename_human_headers -h
```

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

#### Input parameters

The gto\_fasta\_rename\_human\_headers 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 gto\_fasta\_rename\_human\_headers program is a FASTA or Multi-FASTA file. An example, for the input, is:

#### >chr1

ACAAGACGCCTCCTGCTGCTGCTGCTCTCCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGCCCCTCATAGGAGAGGCCTCGGGAGGTGGCCAGGCGAGCAGAAGCAGGCCAGTGCC
GCGAATCCGCGCGCCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCACCCCCCCAGC
TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA

#### >chr2

ACAAGATGCCATTGTCCCCCGGCCTCCTGCTGCTGCTGCTCCTCCCGGGGCCACGGCCACCGCTGCCCTGCCCTGGAGGGT
GGCCCCACCGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGGAATAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTG
GTGGTTTGAGTGGACCTCCCAGGCCAGTGCCGGGCCCCTCATAGGAGAGGCTCGGGAGGTGGCCAGGCGGCAGGAAG
GCGCACCCCCCCAGCAATCCGCGCGCCCGGGACAGATGCCCTGCAGGAACTTCTTCTTCTGGAAGACCTTCTCCTCCTGCAAA
TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA

## Chapter 5

# Genomic sequence tools

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

- 1. gto\_genomic\_gen\_random\_dna: it generates a synthetic DNA.
- 2. gto\_genomic\_rand\_seq\_extra\_chars: it substitues in the DNA sequence the outside ACGT chars by random ACGT symbols.
- 3. gto\_genomic\_dna\_mutate: it creates a synthetic mutation of a sequence file given specific rates of mutations, deletions and additions.
- 4. gto\_genomic\_extract: it extracts sequences from a sequence file, which the range is defined by the user in the parameters.

### $5.1 \quad Program\ gto\_genomic\_gen\_random\ dna$

The gto\_genomic\_gen\_random\_dna generates a synthetic DNA.

For help type:

```
./gto_genomic_gen_random_dna -h
```

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

#### Input parameters

The gto\_genomic\_gen\_random\_dna program needs one stream for the computation, namely the output standard.

```
Usage: ./gto_genomic_gen_random_dna [options] [[--] args]
  or: ./gto_genomic_gen_random_dna [options]

It generates a synthetic DNA.
```

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

Basic options
> output.seq Output synthetic DNA sequence (stdout)

Optional
-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: ./gto_genomic_gen_random_dna > output.seq
```

#### Output

The output of the gto\_genomic\_gen\_random\_dna 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:

### 5.2 Program gto genomic rand seq extra chars

The gto\_genomic\_rand\_seq\_extra\_chars substitues in the DNA sequence the outside ACGT chars by random ACGT symbols. It works in sequence file formats.

For help type:

```
./gto_genomic_rand_seq_extra_chars -h
```

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

#### Input parameters

The gto\_genomic\_rand\_seq\_extra\_chars program needs two streams for the computation, namely the input and output standard. The input stream is a sequence file.

```
Usage: ./gto_genomic_rand_seq_extra_chars [options] [[--] args]
or: ./gto_genomic_rand_seq_extra_chars [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: ./gto_genomic_rand_seq_extra_chars < input.seq > output.seq
```

#### Output

The output of the gto\_genomic\_rand\_seq\_extra\_chars program is a sequence file. An example, for the input, is:

## 5.3 Program gto\_genomic\_dna\_mutate

The gto\_genomic\_dna\_mutate creates a synthetic mutation of a sequence file given specific rates of mutations, deletions and additions. All these parameters are defined by the user, and their are optional. For help type:

```
./gto_genomic_dna_mutate -h
```

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

#### Input parameters

The gto\_genomic\_dna\_mutate 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 sequence File.

The attribution is given according to:

```
Usage: ./gto_genomic_dna_mutate [options] [[--] args]
   or: ./gto_genomic_dna_mutate [options]
Creates a synthetic mutation of a sequence file given specific rates of mutations,
deletions and additions
    -h, --help
                                 show this help message and exit
Basic options
   < input.seq</pre>
                                  Input sequence file (stdin)
   > output.seq
                                  Output sequence file (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: ./gto_genomic_dna_mutate -s <seed> -m <mutation rate> -d <deletion rate> -i
<insertion rate> -a < input.seq > output.seq
```

An example on such an input file is:

#### Output

The output of the gto\_genomic\_dna\_mutate program is a sequence 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:

```
C C C C A A C G A C C T A G T A T C C A A A G C C G C A T A C G A C T T A C A G A A C A G G C T G T A T C G T C A G G A G T G T G T A C A C G A A G A G T
A
```

### 5.4 Program gto genomic extract

The gto\_genomic\_extract extracts sequences from a sequence file, which the range is defined by the user in the parameters.

For help type:

```
./gto_genomic_extract -h
```

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

#### Input parameters

The gto\_genomic\_extract 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 sequence file.

The attribution is given according to:

An example on such an input file is:

#### Output

The output of the gto\_genomic\_extract 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, is:

TCTTTACTCGCGCGTTGGAGAAATACAATAGTGCGGCTCTGTCTCCTTAT

## Chapter 6

# General purpose tools

- 1. gto\_char\_to\_line: it splits a sequence into lines, creating an output sequence which has a char for each line.
- 2. gto\_reverse: it reverses the order of a sequence.
- 3. gto\_new\_line\_on\_new\_x: it splits different rows with a new empty row.
- 4. gto\_upper\_bound: it sets an upper bound in a file with a value per line.
- 5. gto\_lower\_bound: it sets an lower bound in a file with a value per line.
- 6. gto\_brute\_force\_string: it generates all combinations, line by line, for an inputted alphabet and specific size.
- 7. gto\_real\_to\_binary\_with\_threshold: it converts a sequence of real numbers into a binary sequence, given a threshold.

### 6.1 Program gto char to line

The gto\_char\_to\_line splits a sequence into lines, creating an output sequence which has a char for each line.

For help type:

```
./gto_char_to_line -h
```

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

#### Input parameters

The gto\_char\_to\_line program needs two streams for the computation, namely the input and output standard. The input stream is a sequence file.

#### Output

The output of the gto\_char\_to\_line program is a group sequence splited by \n foreach character. An example, for the input, is:

```
Α
C
A
G
A
С
G
G
С
С
Т
С
С
Т
G
С
Т
G
С
Т
```

. . .

### 6.2 Program gto reverse

The gto\_reverse reverses the order of a sequence file.

For help type:

```
./gto_reverse -h
```

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

#### Input parameters

The gto\_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:

```
Usage: ./gto_reverse [options] [[--] args]
  or: ./gto_reverse [options]

It reverses the order of a sequence file.

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

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

Example: ./gto_reverse < input.seq > output.seq
```

An example on such an input file is:

#### Output

The output of the gto\_reverse program is a group sequence.

An example, for the input, is:

### 6.3 Program gto new line on new x

The gto\_new\_line\_on\_new\_x splits different rows with a new empty row.

For help type:

```
./gto_new_line_on_new_x -h
```

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

#### Input parameters

The gto\_new\_line\_on\_new\_x 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: ./gto_new_line_on_new_x [options] [[--] args]
    or: ./gto_new_line_on_new_x [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: ./gto_new_line_on_new_x < 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 gto\_new\_line\_on\_new\_x 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

      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
      47.000000
      3.000000

      45.000000
      47.000000
      3.000000

      45.000000
      55.000000
      1.000000
```

### 6.4 Program gto\_upper\_bound

The gto\_upper\_bound sets an upper bound in a file with a value per line.

For help type:

```
./gto_upper_bound -h
```

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

#### Input parameters

The gto\_upper\_bound program needs two streams for the computation, namely the input and output standard. The input stream is a numeric file.

```
Usage: ./gto_upper_bound [options] [[--] args]
or: ./gto_upper_bound [options]

It sets an upper bound in a file with a value per line.

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

Basic options
-u, --upperbound=<int> The upper bound value
```

```
0.123
3.432
2.341
1.323
7.538
4.122
0.242
0.654
5.633
```

#### Output

The output of the gto\_upper\_bound program is a set of numbers truncated at the a defined upper bound. An example, for the input, is:

```
Using upper bound: 4
0.123000
3.432000
2.341000
1.323000
4.000000
4.000000
0.242000
0.654000
4.000000
```

### 6.5 Program gto\_lower\_bound

The gto\_lower\_bound sets an lower bound in a file with a value per line.

For help type:

```
./gto_lower_bound -h
```

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

#### Input parameters

The gto\_lower\_bound program needs two streams for the computation, namely the input and output standard. The input stream is a numeric file.

```
0.123
3.432
2.341
1.323
7.538
4.122
0.242
0.654
5.633
```

### Output

The output of the gto\_lower\_bound program is a set of numbers truncated at the a defined lower bound. An example, for the input, is:

```
Using lower bound: 2
2.000000
3.432000
2.341000
2.000000
7.538000
4.122000
2.000000
2.000000
5.633000
```

### 6.6 Program gto brute force string

The gto\_brute\_force\_string generates all combinations, line by line, for an inputted alphabet and specific size.

For help type:

```
./gto_brute_force_string -h
```

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

#### Input parameters

The gto\_brute\_force\_string program needs some parameters for the computation, namely the alphabet and the key size.

The attribution is given according to:

```
Usage: ./gto_brute_force_string [options] [[--] args]
  or: ./gto_brute_force_string [options]

It generates all combinations, line by line, for an inputted alphabet and specific size.

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

Basic options
  -a, --alphabet=<str>  -a, --size=<int> The input alphabet
  -s, --size=<int> The combinations size
  > output Output all the combinations (stdout)

Example: ./gto_brute_force_string-a <alphabet> -s <size> > output
```

#### Output

The output of the gto\_brute\_force\_string program is a set of all possible word combinations with a defined size, using the input alphabet.

Using the alphabet "abAB" with the word size of 3, an example of this program, the output is:

```
aaa
aab
aaA
aaB
aba
...
BBb
BBA
BBB
```

### 6.7 Program gto real to binary with threshold

The gto\_real\_to\_binary\_with\_threshold converts a sequence of real numbers into a binary sequence, given a threshold. The numbers below to the threshold will be 0.

For help type:

```
./gto_real_to_binary_with_threshold -h
```

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

#### Input parameters

The gto\_real\_to\_binary\_with\_threshold program needs program needs two streams for the computation, namely the real sequence as input. These numbers should be splitted by lines.

The attribution is given according to:

An example on such an input file is:

```
12.25

1.2

5.44

5.51

7.97

2.34

8.123
```

#### Output

The output of the  ${\tt gto\_real\_to\_binary\_with\_threshold}$  program is a binary sequence.

An example, for the input using the threshold of 5.5, is:

```
1
0
0
1
1
1
```