

### The Genomics Toolkit

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## Chapter 1

## Introduction

Recent advances in DNA sequencing, specifically in next-generation sequencing (NGS), revolutionised the field of genomics, making possible the generation of large amounts of sequencing data very rapidly and at substantially low cost [1]. This new technology also brought with it several challenges, namely in what concerns the analysis, storage, and transmission of the generated sequences [2, 3]. As a consequence, several specialised tools were developed throughout the years in order to deal with these challenges.

Firstly, the storage of the raw data generated by NGS experiments is possible by using several file formats, the FASTQ and FASTA are the most commonly used [4]. FASTQ is an extension of the FASTA format, that besides the nucleotide sequence, also stores associated per base quality score and it is considered the standard format for sequencing data storage and exchange [5].

Regarding the analysis and manipulation of these sequencing data files many software applications emerged, including fqtools [6], FASTX-Toolkit [7], GALAXY [8], GATK [9], MEGA [10], SeqKit [11], among others. Fqtools is a suite of tools to view, manipulate and summarise FASTQ data. This software also identifies invalid FASTQ files [6]. GALAXY, in its turn, is an open, web-based scientific platform for analysing genomic data [12]. This platform integrates several specialised sets of tools, e.g. for manipulating FASTQ files [13]. FASTX-Toolkit is a collection of command-line tools to process FASTA and FASTQ files. This toolkit is available in two forms: as a command-line, or integrated into the web-based platform GALAXY [7]. SeqKit is another toolkit used to process FASTA and FASTQ files and is available for all major operating systems [11]. The Genome Analysis Toolkit (GATK) was designed as a structured programming framework to simplify the development of analysis tools. However, nowadays, it is a suite of tools focused on variant discovering and genotyping [14]. More towards the evolutionary perspectives, Molecular Evolutionary Genetics Analysis (MEGA) software provides tools to analyse DNA and protein sequences statistically [15]. Several of these frameworks lack on variety, namely the ability to perform multiple tasks using only one toolkit.

Compression is another important aspect when dealing with high-throughput sequencing data, as it reduces storage space and accelerates data transmission. A survey on DNA compressors and amino acid sequence compression can be found in [16]. Currently, the DNA sequence compressors HiRGC [17], iDo-Comp [18], GeCo [19], and GDC [20] are considered to have the best performance [21]. Of these four

approaches, GeCo is the only one that can be used for reference-free and reference-based compression. Furthermore, GeCo can be used as an analysis tool to determine absolute measures for many distance computations and local measures [19].

Amino acid sequences are known to be very hard to compress [22], however, Hosseini et al. [23] recently developed AC, a state-of-the-art for lossless amino acid sequence compression. In [24] the authors compared the performance of AC, in terms of bit-rate, to several general-purpose lossless compressors and several protein compressors, using different proteomes. They concluded that in average AC provides the best bit-rates.

Another relevant subject is genomic data simulation. Read simulations tools are fundamental for the development, testing and evaluation of methods and computational tools [25, 26]. Despite the availability of a large number of real sequence reads, read simulation data is necessary due to the inability to know the ground truth of real data [27]. Escalona et al. [28], recently, reviewed 23 NGS simulation tools. XS [29], a FASTQ read simulation tool, stands out in relation to the other 22 simulation tools because it is the only one that does not need a reference sequence. Furthermore, XS is the only open-source tool for simulation of FASTQ reads produced by the four most used sequencing machines, Roche-454, Illumina, ABI SOLiD and Ion Torrent.

Although a large number of tools are available for analysing, compressing, and simulation, these tools are specialised in only a specific task. Besides, in many cases the output of one tool cannot be used directly as input for another tool, e.g. the output of a simulation tool cannot always be used directly as input for an analysis tool. Thus, unique software that includes several specialised tools is necessary.

In this document, we describe  $\mathtt{GTO}$ , a complete toolkit for genomics, namely for  $\mathtt{FASTQ/A}$  formats and sequences (DNA, amino acids, text), with many complementary tools. The toolkit is for Unix-based systems, built for ultra-fast computations.  $\mathtt{GTO}$  supports pipes for easy integration with the sub-programs belonging to  $\mathtt{GTO}$  as well as external tools.  $\mathtt{GTO}$  works as  $\mathtt{LEGOs}$ , since it allows the construction of multiple pipelines with many combinations.

GTO includes tools for information display, randomisation, edition, conversion, extraction, search, calculation, compression, simulation and visualisation. GTO is prepared to deal with very large datasets, typically in the scale of Gigabytes or Terabytes (but not limited). The complete toolkit is an optimised 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:

```
http://bioinformatics.ua.pt/gto
```

### 1.1 Installation

For GTO installation, run:

```
git clone https://github.com/bioinformatics-ua/gto.git
cd gto/src/
make
```

### 1.2 Testing

The examples provided in this document are available in the repository. Therefore, each example can be easily reproduced, which it will also test and validate each tool. It is possible to access the demo files for each tool or run all the tests, as follows:

• Run one test for a specific tool:

```
cd gto/tester/gto_{tool}
sh runExample.sh
```

• Run the tests for all the tools:

```
cd gto/tester/
sh runAllTests.sh
```

Some of this tests require internet connection to download external files and it will create new files.

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## Chapter 2

## FASTQ tools

The toolkit has a set of tools dedicated to manipulating FASTQ files. Some of these tools allow the conversion to/from different formats, i. e., there are tools design to convert a FASTQ file into a sequence or a FASTA file, or receive some of these file types and convert to FASTQ.

There are also tools for data manipulation in this format, which are designed to exclude 'N', remove low quality scored reads, following different metrics and randomize some DNA sequences. Succeeding the manipulation, it is possible also to perform analyses over these files, simulations and mutations. 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.
- 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.
- 21. gto\_fastq\_xs: it is a skilled FASTQ read simulation tool, flexible, portable and tunable in terms of sequence complexity.
- 22. gto\_fastq\_clust\_reads: it agroups reads and creates an index file.

### $2.1 \quad Program\ gto\_fastq\_to\_fasta$

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.

```
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.
```

### Output

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

Using the input above, an output example for this is the following:

```
> Computed with Fastq2Fasta

GGGTGATGGCCGCTGCCGATGGCGTCAAATCCCACCAAGTTACCCTTAACAACTTAAGGGTTTTCAAATAGA

GTTCAGGGATACGACGTTTGTATTTTAAGAATCTGAAGCAGAAGTCGATGATAATACGCGTCGTTTTATCAT
```

### $2.2 \quad Program\ gto\_fastq\_to\_mfasta$

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.
```

### Output

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

Using the input above, an output example for this is the following:

```
>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
GTTCAGGGATACGACGTTTGTATTTTAAGAATCTGAAGCAGAAGTCGATGATAATACGCGTCGTTTTATCAT
```

### 2.3 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</pre>
                         Input FASTQ file format (stdin)
    > output.fastq
                          Output FASTQ file format (stdout)
Example: ./gto_fastq_exclude_n -m <max> < 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 input above with the max value as 5, an output example for this is the following:

### 2.4 Program gto\_fastq\_extract\_quality\_scores

The 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.

The attribution is given according to:

```
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 file format (stdout)
    > output.fastq
Example: ./gto_fastq_extract_quality_scores < input.fastq > output.fastq
Console output example:
<FASTQ quality scores>
Total reads
             : value
Total Quality-Scores : value
```

An example of such an input file is:

### 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. Using the input above, an output example for this is the following:

```
Total reads : 2
Total Quality-Scores : 144
```

### 2.5 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
                         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 of such an input file is:

### Output

The output of the gto\_fastq\_info program is a set of information related to the file read. Using the input above, an output example for this is the following:

```
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 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 of 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 input above with the size values as 60, an output example for this is the following:

### 2.7 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.

```
> output.fastq Output FASTQ file format (stdout)

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
```

### 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 input above with the minimum averge value as 30, an output example for this is the following:

### 2.8 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 file format (stdin)
   < input.fastq
   > 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 input above with the size values as 65, an output example for this is the following:

### 2.9 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.

The attribution is given according to:

```
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

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

Example: ./gto_fastq_rand_extra_chars < input.fastq > output.fastq
```

An example of such an input file is:

### Output

The output of the gto\_fastq\_rand\_extra\_chars program is a FASTQ file.

Using the input above, an output example for this is the following:

### 2.10 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.

The attribution is given according to:

An example of such an input file is:

#### 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:

```
@SeqToFastq1
@SeqToFastq2
@SeqToFastq3
@SeqToFastq4
\tt GCGAATCCGCGCGGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCACCCCCCAGC
@SeqToFastq5
@SeqToFastq6
@SeqToFastq7
@SeqToFastq8
@SegToFastg9
\tt AGAATGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCTCCTGCAAATAAAACCTCACCCATGAATGCTCACGCAAGTT
@SeqToFastq10
TAATTACAGACCTGAA
FFFFFFFFFFFFFFF
```

### 2.11 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.

The attribution is given according to:

```
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
```

An example of such an input file is:

#### Output

The output of the gto\_fastq\_mutate program is a FASTQ file whith the synthetic mutation of input file. Using the input above with the seed value as 1 and the mutation rate as 0.5, an output example for this is the following:

```
QSRR001666.1 071112_SLXA-EAS1_s_7:5:1:817:345 length=72
GGACTTTGAGGTGTGGCGATAGACTGAAAACACTTCAGGGTAAAATCACTCGCAAAAGTGCTATGGTTATGG
+SRR001666.1 071112_SLXA-EAS1_s_7:5:1:817:345 length=72
```

### 2.12 Program gto fastq split

The gto\_fastq\_split splits Paired End files according to the direction of the strand ('/1' or '/2'). 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 -f <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 of such an input file is:

### Output

The output of the gto\_fastq\_split program is a set of information related to the file read. Using the input above, an output example for this is the following:

```
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:

### 2.13 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.

```
Optional
-s, --scores When active, the application show the scores first

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

### Output

The output of the gto\_fastq\_pack program is a packaged FASTQ file. Using the input above, an output example for this is the following:

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

### 2.14 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 packaged FASTQ file.

The attribution is given according to:

An example of such an input file is:

### Output

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

Using the input above, an output example for this is the following:

### 2.15 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.

The attribution is given according to:

```
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 file format (stdin)
   < input.fastq
                         Output read information (stdout)
   > output
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
```

An example of such an input file is:

### Output

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

### 2.16 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 of 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 input above with the max window value as 30, an output example for this is the following:

### 2.17 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 of 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 input above with the max window value as 30, an output example for this is the following:

### 2.18 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.

```
Example: ./gto_fastq_cut -i <initial> -e <end> < input.fastq > output.fastq
```

### 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
+
IIIIIIIIIIIIIIIIIIIII
```

### $2.19 \quad Program\ gto\_fastq\_minimum\_local\_quality\_score\_forward$

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)
                           The minimum value of the quality score (default 33)
   -m, --minqs = <int>
                           Input FASTQ file format (stdin)
   < input.fastq</pre>
   > output.fastq
                            Output FASTQ file format (stdout)
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.

Using the input above with the default values, an output example for this is the following:

### 2.20 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.

The attribution is given according to:

```
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)
                          The minimum value of the quality score (default 33)
   -m, --minqs=<int>
   < input.fastq</pre>
                           Input FASTQ file format (stdin)
   > output.fastq
                            Output FASTQ file format (stdout)
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
```

An example of such an input file is:

### 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.

Using the input above with the default values, an output example for this is the following:

### 2.21 Program gto fastq xs

The gto\_fastq\_xs is a skilled FASTQ read simulation tool, flexible, portable (does not need a reference sequence) and tunable in terms of sequence complexity. XS handles Ion Torrent, Roche-454, Illumina and ABI-SOLiD simulation sequencing types. It has several running modes, depending on the time and memory available, and is aimed at testing computing infrastructures, namely cloud computing of large-scale projects, and testing FASTQ compression algorithms. Moreover, XS offers the possibility of simulating the three main FASTQ components individually (headers, DNA sequences and quality-scores). Quality-scores can be simulated using uniform and Gaussian distributions.

For help type:

```
./gto_xs -h
```

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

### Input parameters

The gto\_fastq\_xs program needs program needs a FASTQ file to compute.

```
Usage: XS
            [OPTION]... [FILE]
System options:
 -h
                          give this help
 - v
                          verbose mode
Main FASTQ options:
                          type: 1=Roche-454, 2=Illumina, 3=ABI SOLiD, 4=Ion Torrent
 -t <sequencingType>
 -hf <headerFormat>
                          header format: 1=Length appendix, 2=Pair End
 -i n=<instrumentName>
                          the unique instrument name (use n= before name)
                          use the same header in third line of the read
                          static line (bases/quality scores) size
 -ls <lineSize>
```

```
-ld <minSize>:<maxSize> dynamic line (bases/quality scores) size
 -n <numberOfReads>
                           number of reads per file
DNA options:
 -f \langle A \rangle,\langle C \rangle,\langle G \rangle,\langle T \rangle,\langle N \rangle symbols frequency
-rn <numberOfRepeats> repeats: number (default: 0)
-ri <repeatsMinSize> repeats: minimum size
 -ra <repeatsMaxSize>
                           repeats: maximum size
-rm <mutationRate>
                         repeats: mutation frequency
                           repeats: use reverse complement repeats
 -rr
Quality scores options:
 -qt <assignmentType>
                           quality scores distribution: 1=uniform, 2=gaussian
 -qf <statsFile>
                           load file: mean, standard deviation (when: -qt 2)
                           custom template ascii alphabet
-qc <template>
Filtering options:
                           excludes the use of headers from output
 -eh
 -eo
                           excludes the use of optional headers (+) from output
 -ed
                           excludes the use of DNA bases from output
 -edb
                           excludes '\n' when DNA bases line size is reached
                           excludes the use of quality scores from output
 -es
Stochastic options:
-s <seed>
                           generation seed
<genFile>
                           simulated output file
Common usage:
 ./XS -v -t 1 -i n=MySeq -ld 30:80 -n 20000 -qt=1 -qc 33,36,39:43 File
 ./XS -v -ls 100 -n 10000 -eh -eo -es -edb -f 0.3,0.2,0.2,0.3,0.0 -rn 50 -ri 300 -ra 3000 -rm 0.1 File
```

### Output

The output of the gto\_fastq\_xs program is a FASTQ file

Using the input above using the common usage with 5 reads (-n 5), an output example for this is the following:

```
@output.fastq.598 LQGQLWH01D5WVZ length=62
TTCNTNCCAGGTAAAGAACATNCCGNCGCACTACTCGTAAGACTTGCTGGNCGAGAAAGG
+
)(+!*!$')($(()+'))$$()'!)!$!!$*+)+''('!)))!+!)(!+!*$!'$*)**++!
```

```
@output.fastq.1510 LQGQLWH01A7LJI length=57
CTAGACTACTCGAGCACTAGGCTCGCGTNTACCANGGGGNCTGCGNGTTGGCNCGGT
+
)+(*(+**)!*!!(!(!!(*'*!+*!!))!*!')****!**!**!**'
@output.fastq.2153 LQGQLWH01CHBQJ length=33
ACTTTTGCTCAAGCAGGGTTGCCTAGCAANAC
+
*)++!+*'')***!+)*(*((*)*!'!!!**
@output.fastq.3251 LQGQLWH01C80Y4 length=75
TCTTTCCTTCNCGNCCNAATTCCCCATAANAACTTAAAATCNCNNGCTGCGCGTGATCAACAATATTAATACTCC
+
!*''+*'!''!!!!*!'!+(++)*(*(*!!*((')**!*(!'!!'+)***!**!'()*!*'+'**'!!+'(
@output.fastq.3934 LQGQLWH01AQDXM length=36
GGTAACNNGGAATTCTCCAATTANCCNTGTCCGGC
+
**''!'!!)+)+!''*****!!')!+)!)*()!))$
```

### 2.22 Program gto fastq clust reads

The gto\_fastq\_clust\_reads agroups reads and creates an index file. It cluster reads in therms of Seq k-mer Lexicographical order.

For help type:

```
./gto_fastq_clust_reads -h
```

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

### Input parameters

The gto\_fastq\_clust\_reads program needs two streams for the computation, namely the input and output standard. The input stream is a FASTQ file. The program sorts the FASTQ reads according to the lexicographic order of the genomic sequences.

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

It agroups reads and creates an index file.
It cluster reads in therms of Seq k-mer Lexicographical order

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

Basic options
    -c, --ctx=<int>
    < input.fastq Input FASTQ file format (stdin)
    > output.fastq Output FASTQ file format (stdout)

Example: ./gto_fastq_clust_reads -c <ctx> < input.fastq > output.fastq
```

#### Output

The output of the gto\_fastq\_clust\_reads program is a FASTQ file with clustered reads in therms of the genomic sequence k-mer Lexicographical order. An example, for the output, is:

## Chapter 3

## FASTA tools

The FASTA tool subset has similar goals to the FASTQ tools. With these tools, it is possible convert data from different formats to the FASTA and multi-FASTA files, or the opposite. In these tools, there are also features to extract and filter reads based on patterns, which can solve specific problems in genomic analytic workflows. The currently 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.
- 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.
- 12. gto\_fasta\_extract\_pattern\_coords: it extracts the header and coordinates from a Multi-FASTA file format given a pattern/motif in the sequence.

## 3.1 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 of such an input file is:

```
>ABOOO264 |acc=ABOOO264|descr=Homo sapiens mRNA

ACAAGACGGCCTCCTGCTGCTGCTCTCCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC

GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA

GTGGTTTGAGTGGACCTCCGGGCCCCTCATAGGAAGAGCACCGGGAGGTGGCCAGGCGAGGAAGCAGGCCAGTGCC

GCGAATCCGCGCGCGCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCACCCCCCCAGC

TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA

>ABOOO263 |acc=ABOOO263|descr=Homo sapiens mRNA

ACAAGATGCCATTGTCCCCCGGGCTCCTGCTGCTGCTGCTCCTCCGGGGCCACCGCCACCGCTGCCCTTGCCCCTGGAGGGT

GGCCCCACCGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAATAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTG

GTGGTTTGAGTGGACCTCCCAGGCCAGTGCCGGGCCCCTCATAGGAGAGCTCCGGGAGGTGGCCAGGCGGCAGGAAG

GCGCACCCCCCCAGCAATCCGCGCGCCCGGGACAGAATGCCCTGCAGGAACTTCTTCTCGGAAGACCTTCTCCTCCTCCTGCAAA

TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
```

#### Output

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

Using the input above, an output example for this is the following:

### 3.2 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 of such an input file is:

#### Output

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

Using the input above with the size line as 80 and the read's header as "SeqToFasta", an output example for this is the following:

## 3.3 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.

The attribution is given according to:

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

```
>ABO00264 |acc=ABO00264|descr=Homo sapiens mRNA
ACAAGACGGCCTCCTGCTGCTGCTCCTCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGCCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGCCAGTGCC
GCGAATCCGCGCGCGCGGGACAGATCTCCTGCAAAGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCACCCCCCCAGC
TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
```

#### Output

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

Using the input above with the value 0 as the extraction starting point and the 50 as the ending, an output example for this is the following:

```
ACAAGACGGCCTCCTGCTGCTCCCGGGGCCACGGCCCTGGAGG
```

## 3.4 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.

The attribution is given according to:

```
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.mfasta > output.mfasta
```

#### Output

The output of the gto\_fasta\_extract\_by\_read program is FASTA or Multi-FASTA file wiht the extracted sequences.

Using the input above with the value 0 as the extraction starting point and the 50 as the ending, an output example for this is the following:

```
>AB000264 |acc=AB000264|descr=Homo sapiens mRNA
ACAAGACGGCCTCCTGCTGCTGCTCCCGGGGCCACGGCCCTGGAGG
>AB000263 |acc=AB000263|descr=Homo sapiens mRNA
ACAAGATGCCATTGTCCCCCGGCCTCCTGCTGCTGCTCCTCCCGGGGCC
```

## 3.5 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.

The attribution is given according to:

```
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.mfasta > output
Output example :
Number of reads
                    : value
Number of bases
MIN of bases in read : value
MAX of bases in read : value
AVG of bases in read : value
```

An example of such an input file is:

```
>ABOOO264 | acc=ABOOO264 | descr=Homo sapiens mRNA

ACAAGACGCCTCCTGCTGCTGCTCCTCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCCATTGTCCCC

GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA

GTGGTTTGAGTGGACCTCCGGGCCCCTCATAGGAAGGCAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGTGCC

GCGAATCCGCGCGCGCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCACCCCCCCAGC

TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA

>ABOOO263 | acc=ABOOO263 | descr=Homo sapiens mRNA

ACAAGATGCCATTGTCCCCCGGGCCTCCTGCTGCTGCTCCTCCCGGGGCCCACCGCTGCCCTGCCCCTGGAGGGT

GGCCCCACCGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAATAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTG

GTGGTTTGAGTGGACCTCCCAGGCCAGTGCCGGGCCCCTCATAGGAGAGGAACTTCTTCTTGGAAGACCTTCTCCTCCTGCAAA

TAAAACCTCACCCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
```

#### Output

The output of the gto\_fasta\_info program is a set of information related to the file read. Using the input above, an output example for this is the following:

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

```
MAX of bases in read : 368.0000
```

## 3.6 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
                                   show this help message and exit
    -h, --help
Basic options
                                  Input FASTA or Multi-FASTA file format (stdin)
   < input.fasta</pre>
    > output.fasta
                                   Output FASTA or Multi-FASTA file format (stdout)
Optional
    -, --seed=<int>
-e, --edit-rate=<dbl>
                                  Starting point to the random generator
                                 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.mfasta > output.fasta
```

An example of such an input file is:

```
>AB000264 |acc=AB000264|descr=Homo sapiens mRNA
ACAAGACGGCCTCCTGCTGCTGCTCTCCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCCGATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGCCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCCAGTGCC
```

```
GCGAATCCGCGCGCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCACCCCCCAGC
TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
>ABOOO263 |acc=ABOOO263|descr=Homo sapiens mRNA
ACAAGATGCCATTGTCCCCCGGCCTCCTGCTGCTGCTCTCCCGGGGCCACCGCCACCGCTGCCCTTGCCCTTGAGGGT
GGCCCCACCGGCCGAGACAGCGATATGCAGGAAGCGGCAGGAATAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTG
GTGGTTTGAGTGGACCTCCCAGGCCAGTGCCGGGCCCCTCATAGGAGAGGCTCGGGAGGTGGCCAGGGGGAAG
GCGCACCCCCCCAGCAATCCGCGCGCCGGGACAGAATGCCCTGCAGGAACTTCTTCTTGGAAGACCTTCTCCTCCTCCAAA
```

#### 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 input above with the seed value as 1 and the edition rate as 0.5, an output example for this is the following:

```
> ABOOO264 | acc = ABOOO264 | descr = Homo sapiens mRNA
ACGCAACGNATTCCTGCTGATCATANTGTNCCGCNCCCCNGCGACGGGGNCTCNCNNGCACACATNGTACCATTGTCCAC
NCTTNCANGTNANCGCTAGCAGGCTACNGTTTNTCCTCNCCTANNCCAANCNGGCGTNNNTACACTGGCACGTGCAGGCA
TNGGTCGGCNGGNNCCTCCGGNAACGGCACCGGAGACCGAGGCTCGGNGGNTATACAGGTGTCANGAAACATCCCCGCGNC
GNGTGNCCNNGAANCCANAGAGTATCTCACTCACAACCCTGCGTGCACNTCTAGAGNANGACCTTACNCACCNTCCCNTT
NNGTACCACACCAATGAACGCTGCAGAAAGTCTGTTTNNAGGNGNGCA
> ABOOO263 | acc = ABOOO263 | descr = Homo sapiens mRNA
ATTTGAAGGCAANCGGNCCAGNAATNCGGNGGGTGCNGCTCNTGTNGGCTACGGNCATCGCGGCCCTGCTNTANTAAGCN
TGAACCACCGNTCGNNGCACTTAGCAATNGCGNAANCCGTCGGCACGGCGGAGACNAANCCGCTANTNNTTTCCCGCTNA
ATGGNTGTACAAGACCNACTANACCANCCTCCGTCACCACACTGGAGCGCANGATGGNNCGCTGNCTAGNAGNCNNTGAG
GCGCTCCNTCCTANAAANCCGTGGNCGAGCNCCCTATGGNAGNGTGGGGGTTTTACCGGGAAGACCNTCGNGCCCTATGGG
AGCAATCANAANCTAGAAAGCTTACNGATGGTGANGAANTAGACTANG
```

## 3.7 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.

The attribution is given according to:

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

#### Output

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

## 3.8 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.

The attribution is given according to:

An example of such an input file is:

#### Output

The output of the gto\_fasta\_extract\_read\_by\_pattern program is a Multi-FASTA file.

Using the input above with the pattern value as "264", an output example for this is the following:

```
>ABOOO264 |acc=ABOOO264|descr=Homo sapiens mRNA
ACAAGACGGCCTCCTGCTGCTGCTCCTCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGCCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGCCAGTGCC
GCGAATCCGCGCGCGCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCACCCCCCCAGC
TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
```

## 3.9 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 of such an input file is:

```
>ABO00264 |acc=ABO00264|descr=Homo sapiens mRNA
NCNNNACGGCCTCCTGCTGCTGCTCCTCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC
GNCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGGAGCATATGCAGGAAGCGGCAGGAA
GTNGTTTGAGTGGACCTCCGGGCCCCTCATAGGAGAGGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGTGCC
GCGAATCCGCGCGCGCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACNTCTTCTGGAAGACCTTCTCCACCCCCCCAGC
TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAN
```

#### 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.

Using the input above, an output example for this is the following:

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

## 3.10 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 of such an input file is:

```
>ABO00264 |acc=AB000264|descr=Homo sapiens mRNA

ACAAGACGGCCTCCTGCTGCTGCTCTCCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCCATTGTCCCC

GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA

GTGGTTTGAGTGGACCTCCGGGCCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGAGCAGGAAGCAGGCCAGTGCC

GCGAATCCGCGCGCGCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACTTCTTCTTGGAAGACCTTCTCCACCCCCCCAGC

TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA

>AB000263 |acc=AB000263|descr=Homo sapiens mRNA

ACAAGATGCCATTGTCCCCCGGGCCTCCTGCTGCTGCTCTCCCGGGGCCACCGCCACCGCTGCCCTTGCCCCTGGAGGGT

GGCCCCACCGGCCGAGACAGCCATATGCAGGAAGCGGCAGGAATAAGGAAAAGCAGCCTCCTGACTTTCCTCCCTTG

GTGGTTTGAGTGGACCTCCCAGGCCAGTGCCGGGCCCCTCATAGGAGAGACCTTCTTCTTGGAAGACCTTCTCCTCCTCCAAA

GCGCACCCCCCCAGCAATCCGCGCCCGGGACAGAATGCCCTGCAGGAACTTCTTCTTGGAAGACCTTCTCCTCCTCCAAA
```

#### 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.

Using the input above, an output example for this is the following:

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

## 3.11 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:

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

An example of such an input file is:

```
> ABO00264 | acc = ABO00264 | descr = Homo sapiens mRNA
ACAAGACGGCCTCCTGCTGCTGCTCCTCCGGGGCCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGCCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGTGCC
GCGAATCCGCGCGCGCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCACCCCCCCAGC
TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
```

```
> ABOOO263 | acc = ABOOO263 | descr = Homo sapiens mRNA
ACAAGATGCCATTGTCCCCCGGCCTCCTGCTGCTGCTCCCGGGGCCACCGGCCACCGCTGCCCTGCCCCTGGAGGGT
GGCCCCACCGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAATAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTG
GTGGTTTGAGTGGACCTCCCAGGCCAGTGCCGGGCCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAG
GCGCACCCCCCCAGCAATCCGCGCCCGGGACAGAATGCCCTGCAGGAACTTCTTCTTGGAAGACCTTCTCCTCCTGCAAA
TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
```

#### Output

The output of the gto\_fasta\_rename\_human\_headers program is a FASTA or Multi-FASTA file. Using the input above, an output example for this is the following:

## 3.12 Program gto fasta extract pattern coords

The gto\_fasta\_extract\_pattern\_coords extracts the header and coordinates from a Multi-FASTA file format given a pattern/motif in the sequence.

For help type:

```
./gto_fasta_extract_pattern_coords -h
```

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

#### Input parameters

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

The attribution is given according to:

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

It extracts the header and coordinates from a Multi-FASTA file format given a pattern/motif in the sequence.

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

```
>ABO00264 |acc=AB000264|descr=Homo sapiens mRNA
ACAAGACGGCCTCCTGCTGCTGCTCTCCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC
GGCCCCACCTAAGGAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGCCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGTGCC
GCGAATCCGCGCGCGCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACTTCTTCTTGGAAGACCTTCTCCACCCCCCAGC
TAAAACCTCACCCATGAATGCTCGCAACACGCAAGTTTAATTCGCAAGTTAGACCTGAACGGGAGGTGGCCACGCAAGTT
```

#### Output

The output of the gto\_fasta\_extract\_pattern\_coords program is a Multi-FASTA file. Using the input above, with the pattern ACA, an output example for this is the following:

```
1 3 >AB000264 |acc=AB000264|descr=Homo sapiens mRNA
131 133 >AB000264 |acc=AB000264|descr=Homo sapiens mRNA
259 261 >AB000264 |acc=AB000264|descr=Homo sapiens mRNA
347 349 >AB000264 |acc=AB000264|descr=Homo sapiens mRNA
```

## Chapter 4

# Genomic sequence tools

The Genomic Sequence subset works directly with the DNA sequences, without any standard format. These tools allow the data extraction, summarising and some mathematical operations over those files. Usually, these are used in the pipeline as a complementary tool. The 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. gto\_genomic\_period: it calculates the best order depth of a sequence, using FCMs.
- 6. gto\_genomic\_count\_bases: it counts the number of bases in sequence, FASTA or FASTQ files.

## $4.1 \quad Program\ gto\_genomic\_gen\_random\_dna$

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.

The attribution is given according to:

```
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)
    -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 -s <seed> -n <nsybomls> -f <fa,fc,fg,ft> > output.seq
```

#### Output

The output of the gto\_genomic\_gen\_random\_dna program is a sequence group file whith the synthetic DNA.

Using the input above with the seed value as 1 and the number of symbols as 400, an output example for this is the following:

## 4.2 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.

The attribution is given according to:

An example of such an input file is:

#### Output

The output of the gto\_genomic\_rand\_seq\_extra\_chars program is a sequence file. Using the input above, an output example for this is the following:

## 4.3 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
                                  Input sequence file (stdin)
    > output.seq
                                  Output sequence file (stdout)
Optional
                                 Starting point to the random generator
    -s. --seed=<int>
                                Defines the mutation rate (default 0.0)
    -m, --mutation-rate=<dbl>
    -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 of 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 input above with the seed value as 1 and the mutation rate as 0.5, an output example for this is the following:

## 4.4 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 of such an input file is:

#### Output

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

Using the input above with the value 0 as the extraction starting point and the 50 as the ending, an output example for this is the following:

TCTTTACTCGCGCGTTGGAGAATACAATAGTGCGGCTCTGTCTCCTTAT

## 4.5 Program gto genomic period

The gto\_genomic\_period calculates the best order depth of a sequence, using FCMs. It only works "ACGT", while the rest will be discarded.

This application has a dependency to represent the results. It requires the Gnuplot to show the execution result.

For help type:

```
./gto_genomic_period -h
```

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

#### Input parameters

The gto\_genomic\_period program needs 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_genomic_period [options] [[--] args]
    or: ./gto_genomic_period [options]

It calculates the best order depth of a sequence, using FCMs.It only works "ACGT",
while the rest will be discarded.

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

Basic options
    < input.seq Input sequence file format (stdin)
```

#### Output

The output of the gto\_genomic\_period program is a execution report, followed by the plot with this information.

Using the input above, an report example for this is the following:

```
Running order: 1 ... Done!
Running order: 2 ... Done!
Running order: 3 ... Done!
Running order: 4 ... Done!
Running order: 5 ... Done!
Running order: 6 ... Done!
Running order: 7 ... Done!
Running order: 8 ... Done!
Running order: 9 ... Done!
Running order: 10 ... Done!
Running order: 11 ... Done!
Running order: 12 ... Done!
Running order: 13 ... Done!
Running order: 14 ... Done!
Running order: 15 ... Done!
Running order: 16 ... Done!
Running order: 17 ... Done!
Running order: 18 ... Done!
Running order: 19 ... Done!
Running order: 20 ... Done!
1 2.246
2 2.225
 3 2.237
 4 2.079
 5 1.821
 6 1.733
7 1.717
8 1.708
9
   1.717
10 1.712
11 1.717
12
   1.721
13 1.725
14 1.729
```

In the Figure 4.1 is represented the plot for the execution above.

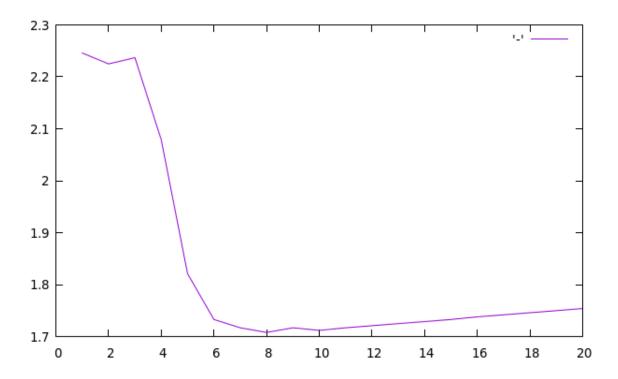


Figure 4.1: gto\_genomic\_period execution plot.

## 4.6 Program gto genomic count bases

The gto\_genomic\_count\_bases counts the number of bases in sequence, FASTA or FASTQ files. For help type:

```
./gto_genomic_count_bases -h
```

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

#### Input parameters

The gto\_genomic\_count\_bases program needs program needs two streams for the computation, namely the input and output standard. The input stream is a sequence, FASTA or FASTQ file.

The attribution is given according to:

```
Usage: ./gto_genomic_count_bases [options] [[--] args]
   or: ./gto_genomic_count_bases [options]
It counts the number of bases in sequence, FASTA or FASTQ files.
   -h, --help Show this help message and exit
Basic options
   < input
                 Input sequence, FASTA or FASTQ file format (stdin)
   > output
                Output read information (stdout)
Example: ./gto_genomic_count_bases < input.seq > output
Output example :
File type : value
Number of bases : value
Number of a/A : value
Number of c/C : value
Number of g/G : value
Number of t/T : value
Number of n/N : value
Number of others : value
```

#### Output

The output of the gto\_genomic\_count\_bases program is report which describes the number of each base in the file, and the file type.

Using the input above, an output example for this is the following:

```
File type : DNA
Number of bases : 480
Number of a/A : 114
Number of c/C : 116
Number of g/G : 120
Number of t/T : 130
Number of n/N : 0
Number of others : 0
```

## Chapter 5

# Amino acid sequence tools

A more specific subset of tools is the Amino Acid Sequence tools, designed to manipulate amino acid sequences. The main features of those tools are grouping sequences, for instance by their properties, such as electric charge (positive and negative), uncharged side chains, hydrophobic side chains and special cases. It is also possible generating pseudo-DNA with characteristics passed by amino acid sequences, or for data compression, using cooperation between multiple contexts and substitutional tolerant context models. The 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.
- 3. gto\_amino\_acid\_compressor: it is a new lossless compressor to compress efficiently amino acid sequences (proteins).

## 5.1 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:

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

```
It converts a amino acid sequence to a group sequence.
    -h, --help
                          show this help message and exit
Basic options
   < input.prot</pre>
                          Input amino acid sequence file (stdin)
   > output.group
                          Output group sequence file (stdout)
Example: ./gto_amino_acid_to_group < input.prot > output.group
Table:
Prot
        Group
        Р
        P Amino acids with electric charged side chains: POSITIVE
Н
D
        N
Е
        N Amino acids with electric charged side chains: NEGATIVE
S
        U
Т
        U
          Amino acids with electric UNCHARGED side chains
N
        U
Q
        U
С
        S
U
        S
G
       S Special cases
Р
        S
        Н
        Н
V
Ι
        Н
L
        Η
М
        H Amino acids with hydrophobic side chains
F
        Н
Y
        Η
        Н
        * Others
Х
        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 of such an input file is:

```
IPFLLKKQFALADKLVLSKLRQLLGGRIKMMPCGGAKLEPAIGLFFHAIGINIKLGYGMTETTATVSCWHDFQFNPNSIG
TLMPKAEVKIGENNEILVRGGMVMKGYYKKPEETAQAFTEDGFLKTGDAGEFDEQGNLFITDRIKELMKTSNGKYIAPQY
IESKIGKDKFIEQIAIIADAKKYVSALIVPCFDSLEEYAKQLNIKYHDRLELLKNSDILKMFE
```

#### Output

The output of the gto\_amino\_acid\_to\_group program is a group sequence. Using the input above, an output example for this is the following:

HSHHHPPUHHHHNPHHHUPHPUHHSSPHPHHSSSSHPHNSHHSHHPHHSHUPHSHSHUNUUHUHUSHPNHUHUSUUHS UHHSPHNHPHSNUUNHHHPSSHHHPSHHPPSNNUHUHHUNNSHHPUSNHSNHNNUSUHHHUNPHPNHHPUUUSPHHHSUH HNUPHSPNPHHNUHHHHHNHPPHHUHHHHSSHNUHNNHHPUHUHPHPNPHNHHPUUNHHPHHN

## 5.2 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
Table:
Prot
        DNA
        GCA
С
        TGC
D
        GAC
Ε
        GAG
F
        TTT
G
        GGC
Н
        CAT
Ι
        ATC
K
        A\,A\,A
L
        CTG
        ATG
М
N
        AAC
Р
        CCG
Q
        CAG
R.
        CGT
```

```
S TCT
T ACG
V GTA
W TGG
Y TAC
* TAG
X GGG
```

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

```
IPFLLKKQFALADKLVLSKLRQLLGGRIKMMPCGGAKLEPAIGLFFHAIGINIKLGYGMTETTATVSCWHDFQFNPNSIG
TLMPKAEVKIGENNEILVRGGMVMKGYYKKPEETAQAFTEDGFLKTGDAGEFDEQGNLFITDRIKELMKTSNGKYIAPQY
IESKIGKDKFIEQIAIIADAKKYVSALIVPCFDSLEEYAKQLNIKYHDRLELLKNSDILKMFE
```

#### Output

The output of the gto\_amino\_acid\_to\_pseudo\_dna program is a DNA sequence. Using the input above, an output example for this is the following:

## 5.3 Program gto amino acid compressor

The gto\_amino\_acid\_compressor is a new lossless compressor to compress efficiently amino acid sequences (proteins). It uses a cooperation between multiple context and substitutional tolerant context models. The cooperation between models is balanced with weights that benefit the models with better performance according to a forgetting function specific for each model.

For help type:

```
./gto_amino_acid_compressor -h
```

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

#### Input parameters

The gto\_amino\_acid\_compressor program needs a file with amino acid sequences to compress.

The attribution is given according to:

```
Usage: ./gto_amino_acid_compressor [OPTION]... -r [FILE] [FILE]:[...]
Compression of amino acid sequences.
Non-mandatory arguments:
  -h
                          give this help,
  - s
                         show AC compression levels,
  - 17
                          verbose mode (more information),
  _ V
                          display version number,
  -f
                         force overwrite of output,
                          level of compression [1;7] (lazy -tm setup),
  -1 <level>
  -t <threshold>
                         threshold frequency to discard from alphabet,
                          it creates a file with the extension ".iae"
  - e
                          with the respective information content.
  -rm < c > : < d > : < g > / < m > : < e > : < a > reference model (-rm 1:10:0.9/0:0:0),
  -rm < c < d < g / m < e < e < a  reference model (-rm 5:90:0.9/1:50:0.8),
  -tm < c > : < d > : < g > / < m > : < e > : < a > target model (-tm 1:1:0.8/0:0:0),
  target and reference templates use <c> for
                          context-order size, \langle d \rangle for alpha (1/\langle d \rangle), \langle g \rangle
                          for gamma (decayment forgetting factor) [0;1),
                          \mbox{\ensuremath{\mbox{cm}}>} to the maximum sets the allowed mutations,
                          on the context without being discarded (for
                          deep contexts), under the estimator <e>, using
                          <a>> for gamma (decayment forgetting factor)</a>
                          [0;1) (tolerant model),
  -r <FILE>
                          reference file ("-rm" are loaded here),
Mandatory arguments:
  <FILE>:<...>
                        file to compress (last argument). For more
                         files use splitting ":" characters.
Example:
               ./gto_amino_acid_compressor -v -tm 1:1:0.8/0:0:0 -tm 5:20:0.9/3:20:0.9 seq.txt
  [Decompress] ./gto_amino_acid_decompressor -v seq.txt.co
```

In the following example, it will be downloaded nine amino acid sequences and compress and decompress one of the smallest (HI). Finally, it compares if the uncompressed sequence is equal to the original.

```
wget http://sweet.ua.pt/pratas/datasets/AminoAcidsCorpus.zip
unzip AminoAcidsCorpus.zip
cp AminoAcidsCorpus/HI .
./gto_amino_acid_compressor -v -1 2 HI
./gto_amino_acid_decompressor -v HI.co
cmp HI HI.de
```

## Chapter 6

# General purpose tools

The toolkit also has a set of tools with a more general-purpose, which were not designed to work with a specific data format. Instead, it was developed as an auxiliary component to help the construction of pipelines combining all the described subsets. This contains tools for char manipulations, such as reversing, segmentation and permutation, for manipulating numerical scores, such sum, filter, calculate the min and the max of a numeric matrix mainly originated from the tools' outputs. The current available tools for general purposes are:

- 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.
- 8. gto\_sum: it adds decimal values in file, line by line, splitted by spaces or tabs.
- 9. gto\_filter: it filters numerical sequences.
- 10. gto\_word\_search: it search for a word in a file.
- 11. gto\_permute\_by\_blocks: it permutates by block sequence, FASTA and Multi-FASTA files.
- 12. gto\_info: it gives the basic properties of the file, namely size, cardinality, distribution percentage of the symbols, among others.

- 13. gto\_segment: it segments a filtered sequence.
- 14. gto\_comparative\_map: it creates a visualization for comparative maps.
- 15. gto\_max: it computes the maximum value in each row between two files.
- 16. gto\_min: it computes the minimum value in each row between two files.
- 17. gto\_geco: it compress and decompress genomic sequences for storage purposes.

### 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.

The attribution is given according to:

An example of such an input file is:

#### Output

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

```
Α
С
Α
Α
Α
С
G
С
T
С
С
T
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.
```

#### Output

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

Using the input above, an output example for this is the following:

## 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 of 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

      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.

Using the input above, an output example for this is the following:

```
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 47.000000 3.000000
```

```
55.000000 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.

The attribution is given according to:

An example of such an input file is:

```
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. Using the input above, an output example for this is the following:

```
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.

The attribution is given according to:

An example of such an input file is:

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

```
0.654
5.633
```

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

```
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.

```
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, --alphabet=<str>
    -s, --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
```

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 input above with the alphabet "abAB" with the word size of 3, an output example for this is the following:

```
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.

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

It converts a sequence of real numbers into a binary sequence given a threshold.

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

Basic options
-t, --threshold=<dbl> The threshold in real format
< input.num Input numeric file (stdin)
> output.bin Output binary file (stdout)
```

```
Example: ./gto_real_to_binary_with_threshold -t <threshold> < input.num > output.bin
```

An example of 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.

Using the input above with the threshold of 5.5, an output example for this is the following:

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

### 6.8 Program gto sum

The gto\_sum adds decimal values in file, line by line, splitted by spaces or tabs.

For help type:

```
./gto_sum -h
```

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

#### Input parameters

The gto\_sum program needs program needs two streams for the computation, namely the input, which is a decimal file.

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

It adds decimal values in file, line by line, splitted by spaces or tabs.

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

Basic options
  < input.num Input numeric file (stdin)</pre>
```

```
Optional
-r, --sumrows When active, the application adds all the values line by line
-a, --sumall When active, the application adds all values

Example: ./gto_sum -a < input.num > output.num
```

An example of such an input file is:

```
      0.123
      5
      5

      3.432
      2

      1.323
      7.538
      5

      4.122
      0.242

      0.654
      5.633
      10
```

#### Output

The output of the gto\_sum program is a sum of the elements in the input file.

Executing the application with the provided input and with the flag to add only the elements in each row, the output of this execution is:

```
10.123000
3.432000
7.341000
1.323000
12.538000
4.122000
0.242000
0.654000
15.633000
```

## 6.9 Program gto filter

The gto\_filter filters numerical sequences using a low-pass filter.

For help type:

```
./gto_filter -h
```

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

#### Input parameters

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

The attribution is given according to:

```
Usage: ./gto_filter [options] [[--] args]
  or: ./gto_filter [options]
It filters numerical sequences using a low-pass filter.
   -h, --help
                        show this help message and exit
Basic options
   < input.num
                        Input numeric file (stdin)
   > output.num
                        Output numeric file (stdout)
Optional
   -w, --windowsize=<int> Window size (defaut 0)
   -d, --drop=<int> Discard elements (default 0.0)
   -t, --windowtype=<int> Window type (0=Hamm, 1=Hann, 2=Black, 3=rec) (default 0 (Hamm))
                      Read from one column
   -c, --onecolumn
   -p, --printone
                       Print one column
   -r, --reverse
                       Reverse mode
```

An example of such an input file is:

```
1 1.77
5 2.18
10 2.32
15 3.15
20 2.52
25 4.43
30 1.23
```

#### Output

The output of the gto\_filter program is a numeric file, identical of the input.

Using the input above with the window size of 3, an output example for this is the following:

```
Got 7 entries from file
1 2.085
5 2.256
10 2.507
15 2.757
20 2.905
25 2.860
30 2.674
```

## 6.10 Program gto\_word\_search

The gto\_word\_search search for a word in a file. It is case sensitive.

For help type:

```
./gto_word_search -h
```

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

#### Input parameters

The gto\_word\_search program needs program needs two streams for the computation, namely the input and output standard. The input stream is a text file.

The attribution is given according to:

An example of such an input file is:

```
No guts, no story. Chris Brady
My life is my message. Mahatma Gandhi
Screw it, letã Á Źs do it. Richard Branson
Boldness be my friend. William Shakespeare
Keep going. Be all in. Bryan Hutchinson
My life is my argument. Albert Schweitzer
Fight till the last gasp. William Shakespeare
Leave no stone unturned. Euripides
```

#### Output

The output of the gto\_word\_search program is a text file with the matching paragraphs and the location of the word found.

Using the input above with the word "Shakespeare", an output example for this is the following:

```
Found match in range [ 1536 : 2048 ]
Boldness be my friend. William Shakespeare

Found match in range [ 3072 : 3584 ]
Fight till the last gasp. William Shakespeare
```

## 6.11 Program gto permute by blocks

The gto\_permute\_by\_blocks permutates by block sequence, FASTA and Multi-FASTA files. For help type:

```
./gto_ -h
```

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

#### Input parameters

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

The attribution is given according to:

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

It permutates by block sequence, FASTA and Multi-FASTA files.

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

Basic options
-b, --numbases=<int> The number of bases in each block
-s, --seed=<int> Starting point to the random generator
< input Input sequence, FASTA or Multi-FASTA file format (stdin)
> output Output sequence, FASTA or Multi-FASTA file format (stdout)

Example: ./gto_permute_by_blocks -b <numbases> -s <seed> < input.fasta > output.fasta
```

An example of such an input file is:

```
>AB000264 |acc=AB000264|descr=Homo sapiens mRNA
ACAAGACGGCCTCCTGCTGCTGCTCTCCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCCATTGTCCCC
GGCCCCACCTAAGGAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGCCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGCCAGTGCC
GCGAATCCGCGCGCGCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCACCCCCCAGC
TAAAACCTCACCCATGAATGCTCGCAACACGCAAGTTTAATTCGCAAGTTAGACCTGAACGGGAGGTGGCCACGCAAGTT
```

#### Output

The output of the gto\_permute\_by\_blocks program is a sequence, FASTA or Multi-FASTA file permuted following some parameters.

Using the input above with the base number as 80, an output example for this is the following:

## 6.12 Program gto info

The gto\_info gives the basic properties of the file, namely size, cardinality, distribution percentage of the symbols, among others.

For help type:

```
./gto_info -h
```

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

#### Input parameters

The gto\_info program needs two streams for the computation, namely the input and output standard. The input stream is a file withou any specific format.

The attribution is given according to:

```
Usage: ./gto_info [options] [[--] args]
   or: ./gto_info [options]
It gives the basic properties of the file, namely size, cardinality, distribution
percentage of the symbols, among others.
   -h, --help
                show this help message and exit
Basic options
   < input
                Input file (stdin)
   > output
                 Output read information (stdout)
Optional
   -a, --ascii When active, the application shows the ASCII codes
Example: ./gto_info < input > output
Output example :
Number of symbols : value
Alphabet size
                 : value
Alphabet
                  : value
Symbol distribution:
<Symbol/Code ASCII> <Symbol count> <Distribution percentage>
```

An example of such an input file is:

```
>AB000264 |acc=AB000264|descr=Homo sapiens mRNA
ACAAGACGGCCTCCTGCTGCTGCTCCTCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCCGTTGCCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGCCCCTCATAGGAGGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGTGCC
```

The output of the gto\_info program is a set of information related to the file read. Using the input above, an output example for this is the following:

```
Number of symbols : 453
Alphabet size
Alphabet :|srponmiedcaTRNHGCBA>=6420 \n
Symbol distribution:
1 : 2 0.4415011
      3
           0.66225166
           0.22075055
      1
p :
     1
           0.22075055
      2
             0.4415011
           0.22075055
      1
m: 2
             0.4415011
i :
     1
           0.22075055
      2
           0.4415011
     1
             0.22075055
c :
      3
             0.66225166
      2
            0.4415011
T: 66
           14.569536
R :
      1
           0.22075055
      1
            0.22075055
      1
             0.22075055
G:
     117
             25.827815
             28.918322
      131
      2
             0.4415011
A :
           19.646799
      89
      1
            0.22075055
      2
             0.4415011
6 :
      2
             0.4415011
      2
            0.4415011
2 : 2
             0.4415011
0 : 6
           1.3245033
     4
             0.88300221
\n : 5
             1.1037528
```

## 6.13 Program gto segment

The gto\_segment segments a filtered sequence.

For help type:

```
./gto_segment -h
```

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

#### Input parameters

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

The attribution is given according to:

An example of such an input file is:

```
1 1.77
5 2.18
10 2.32
15 3.15
20 2.52
25 4.43
30 1.23
```

#### Output

The output of the gto\_segment program is the interval of values âĂŃâĂŃbelow the threshold. Using the input above with a threshold of 3, an output example for this is the following:

```
0:10
```

## 6.14 Program gto\_comparative\_map

The gto\_comparative\_map creates a visualization for comparative maps.

```
For help type:
```

```
./gto_comparative_map -h
```

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

#### Input parameters

The gto\_comparative\_map program needs an input file with the plot positions, respecting a defined structure.

The attribution is given according to:

```
Usage: ./gto_comparative_map [options] [[--] args]
  or: ./gto_comparative_map [options]
It creates a visualization for comparative maps.
   -h, --help
                        Show this help message and exit
Basic options
   <FILE>
                         Contigs filename with positions (.pos),
Optional
   -h
                         Give this help,
   – V
                         Display version number,
   – v
                        Verbose mode (more information),
   -1 <link>
                       Link type between maps [0;4],
   -w <width>
                        Chromosome width,
   -s <space>
                        Space between chromosomes,
   -m <mult>
                        Color id multiplication factor,
   -b <begin>
-c <minimum>
                        Color id beggining,
                        Minimum block size to consider,
   - i
                        Do NOT show inversion maps,
   -r
                         Do NOT show regular maps,
   -o <FILE>
                        Output image filename with map,
Example: ./gto_comparative_map -o map.svg map.config
```

An example of such an input file is:

```
#SCF 5000000 5000000

aaa 1 1000000 1 1000000 bbbb 3000000 4000000 3000000 4000000

bbb 1500000 2000000 1500000 2000000 cccc 1500000 2000000 1500000 2000000

aaa 2000000 3000000 2000000 3000000 bbbb 3000000 2000000 2000000
```

#### Output

The output of the gto\_comparative\_map program is a executing report, and a svg plot with the maps. Using the input above, an output example for this is the following:

In the Figure 6.1 is represented the plot for the execution above.

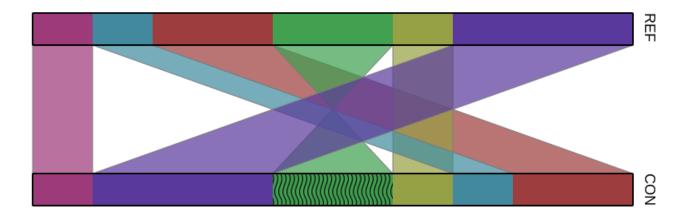


Figure 6.1: gto\_comparative\_map execution plot.

### 6.15 Program gto max

The gto\_max computes the maximum value in each row between two files.

For help type:

```
./gto_max -h
```

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

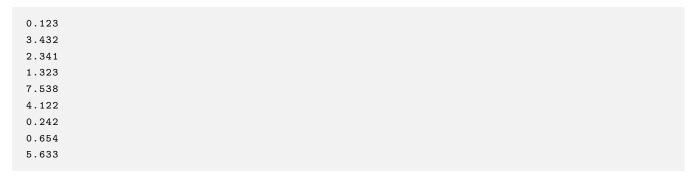
#### Input parameters

The gto\_max program needs program needs two streams for the computation, namely the input, which are two decimal files.

```
Example: ./gto_max -f input1.num -s input2.num > output.num
```

An example of such an input files are:

#### File 1:



#### File 2:

```
2.123
5.312
2.355
0.124
1.785
3.521
0.532
7.324
2.312
```

#### Output

The output of the gto\_max program is the numeric file with the maximum value for each row between both input files.

Executing the application with the provided input, the output of this execution is:

```
2.123000

5.312000

2.355000

1.323000

7.538000

4.122000

0.532000

7.324000

5.633000
```

## 6.16 Program gto\_min

The gto\_min computes the minium value in each row between two files.

For help type:

```
./gto_min -h
```

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

#### Input parameters

The gto\_min program needs program needs two streams for the computation, namely the input, which are two decimal files.

The attribution is given according to:

An example of such an input files are:

#### File 1:

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

#### File 2:

```
2.123
5.312
2.355
0.124
1.785
3.521
0.532
7.324
2.312
```

The output of the gto\_min program is the numeric file with the minimum value for each row between both input files.

Executing the application with the provided input, the output of this execution is:

```
0.123000
3.432000
2.341000
0.124000
1.785000
3.521000
0.242000
0.654000
2.312000
```

## 6.17 Program gto geco

The gto\_geco is able to provide additional compression gains over several top specific tools, while as an analysis tool, it is able to determine absolute measures, namely for many distance computations, and local measures, such as the information content contained in each element, providing a way to quantify and locate specific genomic events.

For help type:

```
./gto_geco -h
```

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

#### Input parameters

The gto\_geco program needs a sequence to compress.

```
./gto_geco [OPTION]... -r [FILE] [FILE]:[FILE]:[FILE]:[...]

SAMPLE

Run Compression : ./gto_geco -v -l 3 sequence.txt
Run Decompression : ./gto_gede -v sequence.txt.co
Run Information Profile : ./gto_geco -v -l 3 -e sequence.txt

DESCRIPTION

Compress and decompress genomic sequences for storage purposes.
Measure an upper bound of the sequences entropy.
Compute information profiles of genomic sequences.

-h, --help
usage guide (help menu).
```

- -V, --version Display program and version information.
- -F, --force force mode. Overwrites old files.
- -v, --verbose  $\qquad \qquad \text{verbose mode (more information)}.$
- -x, --examples
  show several running examples (parameter examples).
- -s, --show-levels show pre-computed compression levels (configured parameters).
- -e, --estimate it creates a file with the extension ".iae" with the respective information content. If the file is FASTA or FASTQ it will only use the "ACGT" (genomic) sequence.
- -1 [NUMBER], --level [NUMBER]

  Compression level (integer).

  Default level: 5.

  It defines compressibility in balance with computational resources (RAM & time). Use -s for levels perception.
- -tm  $[NB_C]:[NB_D]:[NB_I]:[NB_H]:[NB_G]/[NB_S]:[NB_E]:[NB_A]$ Template of a target context model.

Parameters:

- [NB\_C]: (integer [1;20]) order size of the regular context model. Higher values use more RAM but, usually, are related to a better compression score.
- [NB\_D]: (integer [1;5000]) denominator to build alpha, which
   is a parameter estimator. Alpha is given by 1/[NB\_D].
   Higher values are usually used with higher [NB\_C],
   and related to confiant bets. When [NB\_D] is one,
   the probabilities assume a Laplacian distribution.
- [NB\_I]: (integer {0,1,2}) number to define if a sub-program which addresses the specific properties of DNA sequences (Inverted repeats) is used or not. The number 2 turns ON this sub-program without the regular context model (only inverted repeats). The number 1 turns ON the sub-program using at the same time the regular context model. The number O does not contemple its use (Inverted repeats OFF). The use of this sub-program increases the necessary time to compress but it does not affect the RAM.
- [NB\_G]: (real [0;1)) real number to define gamma. This value

```
represents the decayment forgetting factor of the
             regular context model in definition.
     [NB_S]: (integer [0;20]) maximum number of editions allowed
             to use a substitutional tolerant model with the same
             memory model of the regular context model with
             order size equal to [NB_C]. The value 0 stands for
             turning the tolerant context model off. When the
             model is on, it pauses when the number of editions
             is higher that [NB_C], while it is turned on when
             a complete match of size [NB_C] is seen again. This
             is probabilistic-algorithmic model very usefull to
             handle the high substitutional nature of genomic
             sequences. When [NB_S] > 0, the compressor used more
             processing time, but uses the same RAM and, usually,
             achieves a substantial higher compression ratio. The
             impact of this model is usually only noticed for
             [NB_C] >= 14.
     [NB_E]: (integer [1;5000]) denominator to build alpha for
             substitutional tolerant context model. It is
             analogous to [NB_D], however to be only used in the
             probabilistic model for computing the statistics of
             the substitutional tolerant context model.
     [NB_A]: (real [0;1)) real number to define gamma. This value
             represents the decayment forgetting factor of the
             substitutional tolerant context model in definition.
             Its definition and use is analogus to [NB_G].
... (you may use several target models with custom parameters)
-rm [NB_C]:[NB_D]:[NB_I]:[NB_H]:[NB_G]/[NB_S]:[NB_E]:[NB_A]
     Template of a reference context model.
     Use only when -r [FILE] is set (referential compression).
     Parameters: the same as in -tm.
... (you may use several reference models with custom parameters)
-r [FILE], --reference [FILE]
     Reference sequence filename ("-rm" are trainned here).
     Example: -r file1.txt.
[FILE]
     Input sequence filename (to compress) -- MANDATORY.
    File(s) to compress (last argument).
     For more files use splitting ":" characters.
     Example: file1.txt:file2.txt:file3.txt.
```

In the following example, it will be downloaded seventeen DNA sequences, and compress and decompress one of the smallest (BuEb). Finally, it compares if the uncompressed sequence is equal to the original.

```
wget http://sweet.ua.pt/pratas/datasets/DNACorpus.zip
unzip DNACorpus.zip
cp DNACorpus/BuEb .
../../bin/gto_geco -v -1 2 BuEb
../../bin/gto_gede -v BuEb.co
```

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