



The Genomics Toolkit

João Rafael Almeida^{1,2,3} (joao.rafael.almeida@ua.pt)

Armando José Pinho^{2,3} (ap@ua.pt)

José Luís Oliveira^{2,3} (jlo@ua.pt)

Olga Fajarda^{2,3} (olga.oliveira@ua.pt)

Diogo Pratas^{1,2,4} (pratas@ua.pt)

¹Institute of Electronics and Informatics Engineering of Aveiro, University of Aveiro, Aveiro, Portugal

²Department of Electronics, Telecommunications and Informatics, University of Aveiro, Aveiro, Portugal

³Department of Information and Communications Technologies, University of A Coruña, A Coruña, Spain

⁴Department of Virology, University of Helsinki, Helsinki, Finland

Version 1.4

Contents

| | | |
|----------|---|----------|
| 1 | Introduction | 4 |
| 1.1 | Installation | 5 |
| 1.2 | Testing | 6 |
| 1.3 | License | 6 |
| 2 | FASTQ tools | 7 |
| 2.1 | Program gto_fastq_to_fasta | 8 |
| 2.2 | Program gto_fastq_to_mfasta | 9 |
| 2.3 | Program gto_fastq_exclude_n | 10 |
| 2.4 | Program gto_fastq_extract_quality_scores | 12 |
| 2.5 | Program gto_fastq_info | 13 |
| 2.6 | Program gto_fastq_maximum_read_size | 14 |
| 2.7 | Program gto_fastq_minimum_quality_score | 15 |
| 2.8 | Program gto_fastq_minimum_read_size | 16 |
| 2.9 | Program gto_fastq_rand_extra_chars | 18 |
| 2.10 | Program gto_fastq_from_seq | 19 |
| 2.11 | Program gto_fastq_mutate | 21 |
| 2.12 | Program gto_fastq_split | 22 |
| 2.13 | Program gto_fastq_pack | 23 |
| 2.14 | Program gto_fastq_unpack | 25 |
| 2.15 | Program gto_fastq_quality_score_info | 26 |
| 2.16 | Program gto_fastq_quality_score_max | 27 |
| 2.17 | Program gto_fastq_quality_score_min | 28 |
| 2.18 | Program gto_fastq_cut | 29 |
| 2.19 | Program gto_fastq_minimum_local_quality_score_forward | 30 |
| 2.20 | Program gto_fastq_minimum_local_quality_score_reverse | 32 |
| 2.21 | Program gto_fastq_xs | 33 |
| 2.22 | Program gto_fastq_clust_reads | 35 |
| 2.23 | Program gto_fastq_complement | 37 |
| 2.24 | Program gto_fastq_reverse | 38 |

| | | |
|----------|---|-----------|
| 3 | FASTA tools | 40 |
| 3.1 | Program gto_fasta_to_seq | 41 |
| 3.2 | Program gto_fasta_from_seq | 42 |
| 3.3 | Program gto_fasta_extract | 43 |
| 3.4 | Program gto_fasta_extract_by_read | 44 |
| 3.5 | Program gto_fasta_info | 45 |
| 3.6 | Program gto_fasta_mutate | 47 |
| 3.7 | Program gto_fasta_rand_extra_chars | 48 |
| 3.8 | Program gto_fasta_extract_read_by_pattern | 50 |
| 3.9 | Program gto_fasta_find_n_pos | 51 |
| 3.10 | Program gto_fasta_split_reads | 52 |
| 3.11 | Program gto_fasta_rename_human_headers | 53 |
| 3.12 | Program gto_fasta_extract_pattern_coords | 54 |
| 3.13 | Program gto_fasta_complement | 55 |
| 3.14 | Program gto_fasta_reverse | 57 |
| 4 | Genomic sequence tools | 59 |
| 4.1 | Program gto_genomic_gen_random_dna | 59 |
| 4.2 | Program gto_genomic_rand_seq_extra_chars | 60 |
| 4.3 | Program gto_genomic_dna_mutate | 62 |
| 4.4 | Program gto_genomic_extract | 63 |
| 4.5 | Program gto_genomic_period | 64 |
| 4.6 | Program gto_genomic_count_bases | 66 |
| 4.7 | Program gto_genomic_compressor | 68 |
| 4.8 | Program gto_genomic_complement | 70 |
| 4.9 | Program gto_genomic_reverse | 71 |
| 5 | Amino acid sequence tools | 73 |
| 5.1 | Program gto_amino_acid_to_group | 73 |
| 5.2 | Program gto_amino_acid_to_pseudo_dna | 75 |
| 5.3 | Program gto_amino_acid_compressor | 76 |
| 6 | General purpose tools | 78 |
| 6.1 | Program gto_char_to_line | 79 |
| 6.2 | Program gto_new_line_on_new_x | 80 |
| 6.3 | Program gto_upper_bound | 81 |
| 6.4 | Program gto_lower_bound | 83 |
| 6.5 | Program gto_brute_force_string | 84 |
| 6.6 | Program gto_real_to_binary_with_threshold | 85 |
| 6.7 | Program gto_sum | 86 |

| | | |
|------|---|----|
| 6.8 | Program gto_filter | 87 |
| 6.9 | Program gto_word_search | 88 |
| 6.10 | Program gto_permute_by_blocks | 89 |
| 6.11 | Program gto_info | 90 |
| 6.12 | Program gto_segment | 92 |
| 6.13 | Program gto_comparative_map | 93 |
| 6.14 | Program gto_max | 94 |
| 6.15 | Program gto_min | 96 |

| | |
|---------------------|-----------|
| Bibliography | 97 |
|---------------------|-----------|

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 **GTO**, a complete toolkit for genomics, namely for 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. **GTO** supports pipes for easy integration with the sub-programs belonging to **GTO** as well as external tools. **GTO** works as *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.

1.3 License

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

Permissions:

- commercial use;
- modification;
- distribution;
- private use.

Limitations:

- liability;
- warranty.

Conditions:

- License and copyright notice.

For details on the license, consult: <https://opensource.org/licenses/MIT>.

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. The current available tools for FASTQ format analysis and manipulation include:

1. `gto_fastq_to_fasta`: it converts a FASTQ file format to a pseudo FASTA file.
2. `gto_fastq_to_mfasta`: it converts a FASTQ file format to a pseudo Multi-FASTA file.
3. `gto_fastq_exclude_n`: it discards the FASTQ reads with the minimum number of "N" symbols.
4. `gto_fastq_extract_quality_scores`: it extracts all the quality-scores from FASTQ reads.
5. `gto_fastq_info`: it analyses the basic information of FASTQ file format.
6. `gto_fastq_maximum_read_size`: it filters the FASTQ reads with the length higher than the value defined.
7. `gto_fastq_minimum_quality_score`: it discards reads with average quality-score below of the defined.
8. `gto_fastq_minimum_read_size`: it filters the FASTQ reads with the length smaller than the value defined.
9. `gto_fastq_rand_extra_chars`: it substitutes 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.
23. `gto_fastq_complement`: it replaces the ACGT bases with their complements in a FASTQ file format.
24. `gto_fastq_reverse`: it reverses the ACGT bases order for each read in a FASTQ file format.

2.1 Program `gto_fastq_to_fasta`

The `gto_fastq_to_fasta` converts a FASTQ file format to a pseudo FASTA file. However, it does not align the sequence. Also, it extracts the sequence and adds a pseudo header.

For help type:

```
./gto_fastq_to_fasta -h
```

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

Input parameters

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

The attribution is given according to:

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

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

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

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

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

An example of such an input file is:

```
@SRR001666.1 071112_SLXA-EAS1_s_7:5:1:817:345 length=72
GGGTGATGGCCGCTGCCGATGGCGTCAAATCCCACCAAGTTACCCTTAACAACCTTAAGGGTTTTCAAATAGA
+SRR001666.1 071112_SLXA-EAS1_s_7:5:1:817:345 length=72
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII>IIIIII/
@SRR001666.2 071112_SLXA-EAS1_s_7:5:1:801:338 length=72
GTTTCAGGGATACGACGTTTGTATTTTAAGAATCTGAAGCAGAAGTCGATGATAATACGCGTCGTTTATCAT
+SRR001666.2 071112_SLXA-EAS1_s_7:5:1:801:338 length=72
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII>IIIII-I)8I
```

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
GGGTGATGGCCGCTGCCGATGGCGTCAAATCCCACCAAGTTACCCTTAACAACCTTAAGGGTTTTCAAATAGA
GTTTCAGGGATACGACGTTTGTATTTTAAGAATCTGAAGCAGAAGTCGATGATAATACGCGTCGTTTATCAT
```

2.2 Program `gto_fastq_to_mfasta`

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

For help type:

```
./gto_fastq_to_mfasta -h
```

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

Input parameters

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

The attribution is given according to:

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

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


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

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

Example: ./gto_fastq_to_mfasta < input.fastq > output.mfasta
```

An example of such an input file is:

```
@SRR001666.1 071112_SLXA-EAS1_s_7:5:1:817:345 length=72
GGGTGATGGCCGCTGCCGATGGCGTCAAATCCCACCAAGTTACCCTTAACAACCTTAAGGGTTTTCAAATAGA
+SRR001666.1 071112_SLXA-EAS1_s_7:5:1:817:345 length=72
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII>IIIIII/
@SRR001666.2 071112_SLXA-EAS1_s_7:5:1:801:338 length=72
G TTCAGGGATACGACGTTTGTATTTTAAAGATCTGAAGCAGAAGTCGATGATAATACGCGTCGTTTATCAT
+SRR001666.2 071112_SLXA-EAS1_s_7:5:1:801:338 length=72
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII>IIIII-I)8I
```

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
GGGTGATGGCCGCTGCCGATGGCGTCAAATCCCACCAAGTTACCCTTAACAACCTTAAGGGTTTTCAAATAGA
>SRR001666.2 071112_SLXA-EAS1_s_7:5:1:801:338 length=72
G TTCAGGGATACGACGTTTGTATTTTAAAGATCTGAAGCAGAAGTCGATGATAATACGCGTCGTTTATCAT
```

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 parameters.

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.

The attribution is given according to:

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

It discards the FASTQ reads with the minimum number of "N" symbols.
If present, it will erase the second header (after +).

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

Basic options
    -m, --max=<int>          The maximum of of "N" symbols in the read
    < input.fastq            Input FASTQ file format (stdin)
    > output.fastq           Output FASTQ file format (stdout)

Example: ./gto_fastq_exclude_n -m <max> < 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:

```
@SRR001666.1 071112_SLXA-EAS1_s_7:5:1:817:345 length=72
GNNTGATGGCCGCTGCCGATGGCGNANAATCCCAACCAANATACCCTTAACAACCTTAAGGGTTNTCAAATAGA
+
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII>IIIIII/
@SRR001666.2 071112_SLXA-EAS1_s_7:5:1:801:338 length=72
NTTCAGGGATACGACGNTTGTATTTTAAAGAAATCTGNAGCAGAAGTCGATGATAATACGCGNCGTTTTATCAN
+
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII>IIIII-I)8I
```

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:

```
@SRR001666.2 071112_SLXA-EAS1_s_7:5:1:801:338 length=72
NTTCAGGGATACGACGNTTGTATTTTAAGAATCTGNAGCAGAAAGTCGATGATAATACGCNCGTTTTATCAN
+
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIBIIIIIIIIIIIIIIIIIIGII>IIIII-I)8I
Total reads      : 2
Filtered reads   : 1
```

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

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

Console output example:

<FASTQ quality scores>

```
Total reads      : value
```

Total Quality-Scores : value

An example of such an input file is:

```
@SRR001666.1 071112_SLXA-EAS1_s_7:5:1:817:345 length=72  
GGGTGATGGCCGCTGCCGATGGCGTCAAATCCCACCAAGTTACCCTTAACAACCTAAGGGTTTTCAAATAGA  
+SRR001666.1 071112_SLXA-EAS1_s_7:5:1:817:345 length=72  
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII9IG9ICIIIIIIIIIIIIIIIIIIIDIIIIIII>IIIIII/  
@SRR001666.2 071112_SLXA-EAS1_s_7:5:1:801:338 length=72  
GTTTCAGGGATACGACGTTTTGTATTTTAAGAATCTGAAGCAGAAGTCGATGATAATACGCGTCGTTTTATCAT  
+SRR001666.2 071112_SLXA-EAS1_s_7:5:1:801:338 length=72
```

[illegible]

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:

[illegible]

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:

```
@SRR001666.1 071112_SLXA-EAS1_s_7:5:1:817:345 length=72
GGGTGATGGCCGCTGCCGATGGCGTCAAATCCCACCAAGTTACCCTTAACCACTTAAGGGTTTTCAAATAGA
+SRR001666.1 071112_SLXA-EAS1_s_7:5:1:817:345 length=72
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII>IIIIII/
@SRR001666.2 071112_SLXA-EAS1_s_7:5:1:801:338 length=72
GTTTCAGGATACGACGTTTGTATTTTAAAGAATCTGAAGCAGAAGTCGATGATAATACGCGTCGTTTATCAT
+SRR001666.2 071112_SLXA-EAS1_s_7:5:1:801:338 length=72
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII>IIIII-I)8I
```

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 parameters.

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
```


The attribution is given according to:

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

It discards reads with average quality-score below value.

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

Basic options
-m, --min=<int>           The minimum average quality-score (Value 25 or 30 is commonly used)
< input.fastq            Input FASTQ file format (stdin)
> 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
```

An example of such an input file is:

```
@SRR001666.1 071112_SLXA-EAS1_s_7:5:1:817:345 length=72
GGGTGATGGCCGCTGCCGATGGCGTCAAATCCCACCAAGTTACCCTTAACAACCTTAAGGGTTTTCAAATAGA
+SRR001666.1 071112_SLXA-EAS1_s_7:5:1:817:345 length=72
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII>IIIIII/
@SRR001666.2 071112_SLXA-EAS1_s_7:5:1:801:338 length=72
GTTTCAGGGATACGACGTTTGTATTTTAAAGATCTGAAGCAGAAGTCGATGATAATACGCGTCGTTTTATCAT
+SRR001666.2 071112_SLXA-EAS1_s_7:5:1:801:338 length=72
54599<>77977==6=?I6IBI::33344235521677999>>><<@@A@BBBCDGGGBFFH>IIIII-I)8I
```

Output

The output of the `gto_fastq_minimum_quality_score` program is a set of all the filtered FASTQ reads, followed by the execution report.

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

```
@SRR001666.1 071112_SLXA-EAS1_s_7:5:1:817:345 length=72
GGGTGATGGCCGCTGCCGATGGCGTCAAATCCCACCAAGTTACCCTTAACAACCTTAAGGGTTTTCAAATAGA
+
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII>IIIIII/
Total reads      : 2
Filtered reads   : 1
```

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 parameters.

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.

The attribution is given according to:

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

Example: ./gto_fastq_minimum_read_size -s <size> < input.fastq > output.fastq

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

An example of such an input file is:

[illegible]

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:

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:

```
@SRR001666.1 071112_SLXA-EAS1_s_7:5:1:817:345 length=72
GTGTGATGGCCGCTGCCGATGGCGCATAATCCCACCAACATACCCTTAACAACCTTAAGGGTTTTCAAATAGA
+SRR001666.1 071112_SLXA-EAS1_s_7:5:1:817:345 length=72
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII>IIIIII/
@SRR001666.2 071112_SLXA-EAS1_s_7:5:1:801:338 length=72
GTTTCAGGATACGACGATTGTATTTTAAAGATCTGCAGCAGAAGTCGATGATAATACGCGCCGTTTATCAG
+SRR001666.2 071112_SLXA-EAS1_s_7:5:1:801:338 length=72
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII>IIIII-I)8I
```

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 parameters.

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:

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

It converts a genomic sequence to pseudo FASTQ file format.

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

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

Optional options
    -n, --name=<str>         The read's header
    -l, --lineSize=<int>     The maximum of chars for line

Example: ./gto_fastq_from_seq -l <lineSize> -n <name> < input.seq > output.fastq
```

An example of such an input file is:

```

ACAAGACGGCCTCCTGCTGCTGCTGCTCTCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGGCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGTGCC
GCGAATCCGCGCGCCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCACCCCCCAGC
TAAAACCTACCCATGAATGCTCAGCAAGTTTAATTACAGACCTGAAACAAGATGCCATTGTCCCCGGCCTCCTGCTG
CTGCTGCTCTCCGGGGCCACGGCCACCGCTGCCCTGCCCTGGAGGGTGGCCCCACGGCCGAGACAGCGAGCATATGCA
GGAAGCGGCAGGAATAAGGAAAAGCAGCCTCCTGACTTTCTCGCTTGGTGGTTTGAGTGGACCTCCAGGCCAGTGCCG
GGCCCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGGCGCACCCCCCAGCAATCCGCGCGCCGGGAC
AGAATGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCTCCTGCAAATAAAACCTACCCATGAATGCTCAGCAAGTT
TAATTACAGACCTGAA

```

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
ACAAGACGGCCTCCTGCTGCTGCTGCTCTCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC
+
FFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFF
@SeqToFastq2
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
+
FFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFF
@SeqToFastq3
GTGGTTTGAGTGGACCTCCGGGGCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGTGCC
+
FFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFF
@SeqToFastq4
GCGAATCCGCGCGCCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCACCCCCCAGC
+
FFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFF
@SeqToFastq5
TAAAACCTACCCATGAATGCTCAGCAAGTTTAATTACAGACCTGAAACAAGATGCCATTGTCCCCGGCCTCCTGCTG
+
FFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFF
@SeqToFastq6
CTGCTGCTCTCCGGGGCCACGGCCACCGCTGCCCTGCCCTGGAGGGTGGCCCCACGGCCGAGACAGCGAGCATATGCA
+
FFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFF
@SeqToFastq7
GGAAGCGGCAGGAATAAGGAAAAGCAGCCTCCTGACTTTCTCGCTTGGTGGTTTGAGTGGACCTCCAGGCCAGTGCCG
+
FFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFF
@SeqToFastq8
GGCCCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGGCGCACCCCCCAGCAATCCGCGCGCCGGGAC
+
FFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFF
@SeqToFastq9
AGAATGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCTCCTGCAAATAAAACCTACCCATGAATGCTCAGCAAGTT
+
FFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFF

```

```
@SeqToFastq10
TAATTACAGACCTGAA
+
FFFFFFFFFFFFFFFFFF
```

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 they are optional.

For help type:

```
./gto_fastq_mutate -h
```

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

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             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:

```
@SRR001666.1 071112_SLXA-EAS1_s_7:5:1:817:345 length=72
GGGTGATGGCCGCTGCCGATGGCGTCAAATCCCACCAAGTTACCCTTAACAACCTTAAGGGTTTTCAAATAGA
+SRR001666.1 071112_SLXA-EAS1_s_7:5:1:817:345 length=72
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII>IIIIII/
@SRR001666.2 071112_SLXA-EAS1_s_7:5:1:801:338 length=72
GTTTCAGGGATACGACGTTTGTATTTTAAAGAATCTGAAGCAGAAGTCGATGATAATACGCGTCGTTTATCAT
+SRR001666.2 071112_SLXA-EAS1_s_7:5:1:801:338 length=72
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII>IIIII-I)8I
```

Output

The output of the `gto_fastq_mutate` program is a FASTQ file with 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:

```
@SRR001666.1 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
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII>IIIIII/
@SRR001666.2 071112_SLXA-EAS1_s_7:5:1:801:338 length=72
GTTTCAGAGCCTTTACCGTAGGGGTGTAAGATTTTATACAAAAAGTCCAGGTCAAGAGGAATCGGACAACCGA
+SRR001666.2 071112_SLXA-EAS1_s_7:5:1:801:338 length=72
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII>IIIII-I)8I
```

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 parameters.

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         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:

```
@SRR001666.1 071112_SLXA-EAS1_s_7:5:1:817:345 length=72 1
GNNTGATGGCCGCTGCCGATGGCGNANAATCCCACCAANATACCCTTAACAACTTAAGGGTTNTCAAATAGA
+
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII9IG9ICIIIIIIIIIIIIIIIIIIIIIDIIIIIII>IIIIII/
@SRR001666.2 071112_SLXA-EAS1_s_7:5:1:801:338 length=72 2
NTTCAGGGATACGACGNTTGTATTTTAAGAATCTGNAGCAGAAGTCGATGATAATACGCGNCGTTTATCAN
+
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII6IBIIIIIIIIIIIIIIIIIIIIIGII>IIIII-I)8I
```

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:

```
@SRR001666.1 071112_SLXA-EAS1_s_7:5:1:817:345 length=72 1
GNNTGATGGCCGCTGCCGATGGCGNANAATCCCACCAANATACCCTTAACAACTTAAGGGTTNTCAAATAGA
+
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII9IG9ICIIIIIIIIIIIIIIIIIIIIIDIIIIIII>IIIIII/
```

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 parameters.

Input parameters

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

The attribution is given according to:

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

It packages each FASTQ read in a single line.

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

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

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

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

An example of such an input file is:

```
@SRR001666.1 071112_SLXA-EAS1_s_7:5:1:817:345 length=72
GNNTGATGGCCGCTGCCGATGGCGNANAATCCCACCAANATACCCTTAACAACCTAAGGGTTNTCAAATAGA
+
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII9IG9ICIIIIIIIIIIIIIIIIIIIIIIIDIIIIII>IIIIII/
@SRR001666.2 071112_SLXA-EAS1_s_7:5:1:801:338 length=72
NTTCAGGGATACGACGNTTGTATTTTAAGAATCTGNAGCAGAAGTCGATGATAATACGCGNCGTTTATCAN
+
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII6IBIIIIIIIIIIIIIIIIIIIIIIIGII>IIIII-I)8I
```

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:

```
GNNTGATGGCCGCTGCCGATGGCGNANAATCCCACCAANATACCCTTAACAACCTAAGGGTTNTCAAATAGA
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII9IG9ICIIIIIIIIIIIIIIIIIIIIIIIDIIIIII>IIIIII/
SRR001666.1 071112_SLXA-EAS1_s_7:5:1:817:345 length=72+ 0
NTTCAGGGATACGACGNTTGTATTTTAAGAATCTGNAGCAGAAGTCGATGATAATACGCGNCGTTTATCAN
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII6IBIIIIIIIIIIIIIIIIIIIIIIIGII>IIIII-I)8I
SRR001666.2 071112_SLXA-EAS1_s_7:5:1:801:338 length=72+ 1
```

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

```

IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII9IG9ICIIIIIIIIIIIIIIIIIIIIIIIDIIIIIII>IIIIII/
GNNTGATGGCCGCTGCCGATGGCGNANAATCCCACCAANATACCCTTAACAACCTTAAGGGTTNTCAAATAGA
SRR001666.1 071112_SLXA-EAS1_s_7:5:1:817:345 length=72+ 0
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII6IBIIIIIIIIIIIIIIIIIIIIIIIGII>IIIII-I)8I
NTTCAGGGATACGACGNTTGTATTTTAAAGAATCTGNAGCAGAAGTCGATGATAATACGCGNCGTTTATCAN
SRR001666.2 071112_SLXA-EAS1_s_7:5:1:801:338 length=72+ 1

```

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 parameters.

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:

```

Usage: ./gto_fastq_unpack [options] [--] args]
       or: ./gto_fastq_unpack [options]

It unpacks the FASTQ reads packaged using the gto_fastq_pack tool.

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

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

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

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

```

An example of such an input file is:

```

GNNTGATGGCCGCTGCCGATGGCGNANAATCCCACCAANATACCCTTAACAACCTTAAGGGTTNTCAAATAGA
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII9IG9ICIIIIIIIIIIIIIIIIIIIIIIIDIIIIIII>IIIIII/
SRR001666.1 071112_SLXA-EAS1_s_7:5:1:817:345 length=72+ 0
NTTCAGGGATACGACGNTTGTATTTTAAAGAATCTGNAGCAGAAGTCGATGATAATACGCGNCGTTTATCAN
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII6IBIIIIIIIIIIIIIIIIIIIIIIIGII>IIIII-I)8I
SRR001666.2 071112_SLXA-EAS1_s_7:5:1:801:338 length=72+ 1

```

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:

```
@SRR001666.1 071112_SLXA-EAS1_s_7:5:1:817:345 length=72
GNNTGATGGCCGCTGCCGATGGCGNANAATCCCACCAANATACCCTTAACAACCTTAAGGGTTNTCAAATAGA
+
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII>IIIIII/
@SRR001666.2 071112_SLXA-EAS1_s_7:5:1:801:338 length=72
NTTCAGGGATACGACGNTTGTATTTTAAGAATCTGNAGCAGAAGTCGATGATAATACGCGNCGTTTATCAN
+
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII>IIIII-I)8I
```

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 parameters.

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` Input FASTQ file format (stdin)
`> output` Output read information (stdout)

Optional

`-m, --max=<int>` The length 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
QS range          : value
```

An example of such an input file is:

```
@111 071112_SLXA-EAS1_s_7:5:1:817:345 length=72 1
GNNTGATGGCCGCTGCCGATGGCGNANAATCCCACCAANATACCCTTAACAACTTAAGGGTTNTCAAATAGA
+111
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII>IIIIII/
@222 071112_SLXA-EAS1_s_7:5:1:801:338 length=72 2
NTTCAGGGATACGACGNTTGTATTTTAAAGAATCTGNAGCAGAAGTCGATGATAATACGCGNCGTTTATCAN
+222
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII>IIIII-I)8I
```

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:

```
Total reads      : 2
Max read length  : 72
Min read length  : 72
Min QS value     : 41
Max QS value     : 73
QS range         : 33
 1  2  3  4  5  6  7  8  9 10 11 12 13 14 15 16 17 18 19 20 21 22 23 24 25 26 27 28 29 30
--+-+--+--+--+--+--+--+--+--+--+--+--+--+--+--+--+--+--+--+--+--+--+--+--+--+--+--+
73 73 73 73 73 73 73 73 73 73 73 73 73 73 73 73 73 73 73 73 73 73 73 73 73 73 73 73
```

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 parameters.

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:

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

It analyses the maximal quality-scores of a FASTQ file.

```

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

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

Optional
    -m, --max=<int>      The maximum window length (default 40)

Example: ./gto_fastq_quality_score_max -m <max> < input.fastq > output

```

An example of such an input file is:

```

@111 071112_SLXA-EAS1_s_7:5:1:817:345 length=72 1
GNNTGATGGCCGCTGCCGATGGCGNANAATCCCACCAANATACCCTTAACAACCTTAAGGGTTNTCAAATAGA
+
IIIIIIIIII9IG9ICIIIIIIIIIIABAAABCIIIIFFGIIAACBBIIII6IBIIIIII>IIIIII/
@222 071112_SLXA-EAS1_s_7:5:1:801:338 length=72 2
NTTCAGGGATACGACGNTTGTATTTAAGAATCTGNAGCAGAAGTCGATGATAATACGCGNCGTTTTATCAN
+
IIIIIIIIABAAABCIIIIFFGIIAACBBIIII6IBIIIIIIIIIIIIIIIIIIIIIGII>IIIII-I)8I

```

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:

```

 1  2  3  4  5  6  7  8  9 10 11 12 13 14 15 16 17 18 19 20 21 22 23 24 25 26 27 28 29 30
--+-+--+--+--+--+--+--+--+--+--+--+--+--+--+--+--+--+--+--+--+--+--+--+--+--+--+--+
73 73 73 73 73 73 73 73 73 73 73 73 66 73 73 73 73 73 73 73 73 73 73 73 73 73 73 73

```

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 parameters.

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:

```

Usage: ./gto_fastq_quality_score_min [options] [--] args]
       or: ./gto_fastq_quality_score_min [options]

It analyses the minimal quality-scores of a FASTQ file.

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

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

Optional
    -m, --max=<int>          The maximum window length (default 40)

Example: ./gto_fastq_quality_score_min -m <max> < input.fastq > output

```

An example of such an input file is:

```

@111 071112_SLXA-EAS1_s_7:5:1:817:345 length=72 1
GNNTGATGGCCGCTGCCGATGGCGNANAATCCCACCAANATACCCTTAACAACCTTAAGGGTTNTCAAATAGA
+
IIIIIIIIII9IG9ICIIIIIIIIIIABAAABCIIIFFGIIAACBBIIII6IBIIIIII>IIIIII/
@222 071112_SLXA-EAS1_s_7:5:1:801:338 length=72 2
NTTCAGGGATACGACGNTTGTATTTTAAGAATCTGNAGCAGAAGTCGATGATAATACGCGNCGTTTTATCAN
+
IIIIIIABAAABCIIIFFGIIAACBBIIII6IBIIIIIIIIIIIIIIIIIIIIIGII>IIIII-I)8I

```

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:

```

 1  2  3  4  5  6  7  8  9 10 11 12 13 14 15 16 17 18 19 20 21 22 23 24 25 26 27 28 29 30
--+-+--+--+--+--+--+--+--+--+--+--+--+--+--+--+--+--+--+--+--+--+--+--+--+--+--+--+
73 73 73 73 73 73 73 65 66 65 65 65 57 67 71 57 73 67 70 70 71 73 73 65 65 67 66 66 73 65

```

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 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_cut [options] [--] args]
      or: ./gto_fastq_cut [options]

It cuts read sequences in a FASTQ file.

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

Basic options
      -i, --initial=<int>      Starting position to the cut
      -e, --end=<int>          Ending position to the cut
      < input.fastq            Input FASTQ file format (stdin)
      > output.fastq           Output FASTQ file format (stdout)

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

An example of such an input file is:

```
@SRR001666.1 071112_SLXA-EAS1_s_7:5:1:817:345 length=72
GGGTGATGGCCGCTGCCGATGGCGTCAAATCCCACCAAGTTACCCTTAACAACCTTAAGGGTTTTCAAATAGA
+SRR001666.1 071112_SLXA-EAS1_s_7:5:1:817:345 length=72
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII>IIIIII/
@SRR001666.2 071112_SLXA-EAS1_s_7:5:1:801:338 length=72
GTTTCAGGGATACGACGTTTGTATTTTAAAGATCTGAAGCAGAAGTCGATGATAATACGCGTCGTTTTATCAT
+SRR001666.2 071112_SLXA-EAS1_s_7:5:1:801:338 length=72
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII>IIIIII-1)8I
```

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
+
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII9
@SRR001666.2 071112_SLXA-EAS1_s_7:5:1:801:338 length=72
ACGACGTTTGTATTTTAAAGAA
+
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII
```

2.19 Program `gto_fastq_minimum_local_quality_score_forward`

The `gto_fastq_minimum_local_quality_score_forward` filters the reads considering the quality score average of a defined window size of bases.

For help type:

```
./gto_fastq_minimum_local_quality_score_forward -h
```

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

Input parameters

The `gto_fastq_minimum_local_quality_score_forward` 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_forward [options] [--] args]
or: ./gto_fastq_minimum_local_quality_score_forward [options]
```

It filters the reads considering the quality score average of a defined window size of bases.

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

Basic options

`-k, --windowsize=<int>` The window size of bases (default 5)
`-w, --minavg=<int>` The minimum average of quality score (default 25)
`-m, --minqs=<int>` The minimum value of the quality score (default 33)
`< input.fastq` Input FASTQ file format (stdin)
`> 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
```

An example of such an input file is:

```
@SRR001666.1 071112_SLXA-EAS1_s_7:5:1:817:345 length=72
GGGTGATGGCCGCTGCCGATGGCGTCAAATCCCACCAAGTTACCCTTAACAACCTTAAGGGTTTTCAAATAGA
+SRR001666.1 071112_SLXA-EAS1_s_7:5:1:817:345 length=72
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII>IIIIII/
@SRR001666.2 071112_SLXA-EAS1_s_7:5:1:801:338 length=72
GTTTCAGGGATACGACGTTTGTATTTTAAAGATCTGAAGCAGAAGTCGATGATAATACGCGTCGTTTTATCAT
+SRR001666.2 071112_SLXA-EAS1_s_7:5:1:801:338 length=72
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII>IIIII-I)8I
```

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:

```
Minimum QS      : 33
@SRR001666.1 071112_SLXA-EAS1_s_7:5:1:817:345 length=72
GGGTGATGGCCGCTGCCGATGGCGTCAAATCCCACCAAGTTACCCTTAACAACCTTAAGGGTTTTCAAATAGA
+
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII9IG9ICIIIIIIIIIIIIIIIIIIIIIIIDIIIIIII>IIIIII/
@SRR001666.2 071112_SLXA-EAS1_s_7:5:1:801:338 length=72
GTTTCAGGGATACGACGTTTGTATTTTAAGAATCTGAAGCAGAAGTCGATGATAATACGCGTCGTTT
+
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIBIIIIIIIIIIIIIIIIIIIIIIIGII>IIII
Total reads     : 2
Trimmed reads   : 1
```

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 parameters.

Input parameters

The `gto_fastq_minimum_local_quality_score_reverse` 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)
-m, --minqs=<int>       The minimum value of the quality score (default 33)
< input.fastq          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:

```

@SRR001666.1 071112_SLXA-EAS1_s_7:5:1:817:345 length=72
GGGTGATGGCCGCTGCCGATGGCGTCAAATCCCACCAAGTTACCCTTAACAACCTTAAGGGTTTTCAAATAGA
+SRR001666.1 071112_SLXA-EAS1_s_7:5:1:817:345 length=72
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII>IIIIII/
@SRR001666.2 071112_SLXA-EAS1_s_7:5:1:801:338 length=72
GTTTCAGGGATACGACGTTTGTATTTTAAGAATCTGAAGCAGAAGTCGATGATAATACGCGTCGTTTTATCAT
+SRR001666.2 071112_SLXA-EAS1_s_7:5:1:801:338 length=72
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII>IIIII-I)8I

```

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:

```

Minimum QS: 33
@SRR001666.1 071112_SLXA-EAS1_s_7:5:1:817:345 length=72
GGGTGATGGCCGCTGCCGATGGCGTCAAATCCCACCAAGTTACCCTTAACAACCTTAAGGGTTTTCAAATAGA
+
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII>IIIIII/
Total reads      : 2
Trimmed reads    : 1

```

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 a FASTQ file to compute.

The attribution is given according to:

```
Usage: XS [OPTION]... [FILE]

System options:
  -h                give this help
  -v                verbose mode

Main FASTQ options:
  -t <sequencingType>    type: 1=Roche-454, 2=Illumina, 3=ABI SOLiD, 4=Ion Torrent
  -hf <headerFormat>     header format: 1=Length appendix, 2=Pair End
  -i n=<instrumentName>   the unique instrument name (use n= before name)
  -o                  use the same header in third line of the read
  -ls <lineSize>         static line (bases/quality scores) size
  -ld <minSize>:<maxSize> dynamic line (bases/quality scores) size
  -n <numberOfReads>     number of reads per file

DNA options:
  -f <A>,<C>,<G>,<T>,<N> symbols frequency
  -rn <numberOfRepeats>   repeats: number (default: 0)
  -ri <repeatsMinSize>    repeats: minimum size
  -ra <repeatsMaxSize>    repeats: maximum size
  -rm <mutationRate>     repeats: mutation frequency
  -rr                  repeats: use reverse complement repeats

Quality scores options:
  -qt <assignmentType>    quality scores distribution: 1=uniform, 2=gaussian
  -qf <statsFile>         load file: mean, standard deviation (when: -qt 2)
  -qc <template>         custom template ascii alphabet

Filtering options:
  -eh                excludes the use of headers from output
  -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
  -es                excludes the use of quality scores from output

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
```

An example of such an input file is:

```
@SRR001666.1 071112_SLXA-EAS1_s_7:5:1:817:345 length=60
GGGTGATGGCCGCTGCCGATGGCGTCAAATCCCACCAAGTTACCTTAACAACCTTAAGGG
+
```

```

IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII9IG9ICIIIIIIIIIIIIIIIIIIIIIDIII
@SRR001666.2 071112_SLXA-EAS1_s_7:5:1:801:338 length=72
GTTCAGGGATACGACGTTTGTATTTTAAAGATCTGAAGCAGAAGTCGATGATAATACGCGTCGTTTATCAT
+
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII6IBIIIIIIIIIIIIIIIIIIIIIIIGII>IIIII-I)8I

```

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 LQGQLWHO1D5WVZ length=62
TTCNTNCCAGGTAAAGAGAACATNCCGNCGCACTACTCGTAAGACTTGCTGGNCGAGAAAGG
+
)(+!*$')($((+'))$$()'!)$!!$*+)+''('!)))+!)(+!*$'$*)****!
@output.fastq.1510 LQGQLWHO1A7LJI length=57
CTAGACTACTCGAGCACTAGGCTCGCGTNTACCANGGGNCTGCGNGTTGGCNCGGT
+
)+(*($*+!*)!'!!(!(!(*'$!+!((('$'!+*+!)))*!')****!$+'
@output.fastq.2153 LQGQLWHO1CHBQJ length=33
ACTTTTGTCTCAAGCAGGGTTGCCTAGCAANAC
+
*)++!+$''')*)**!+)$(*((*)$!'!+!*
@output.fastq.3251 LQGQLWHO1C80Y4 length=75
TCTTTCCTTCNCGNCCNAATTCCCCATAANAACCTAAATCNCNNGCTGCGCGTGATCAACAATATTAATACTCC
+
!*''+*'''+!!*!'!+(+)*(*($!*($(')$*!$(!'!'+)$+*!$*!***'())$!*'+*'+!!+')(
@output.fastq.3934 LQGQLWHO1AQDXM length=36
GGTAACNNGGAATTCTTCCAATTANCCNTGTCCGGC
+
$+)'!'!+)++!'**$*$*!')!+)!)*()!))$

```

2.22 Program `gto_fastq_clust_reads`

The `gto_fastq_clust_reads` agroups reads and creates an index file. It cluster reads in terms 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 accoring to the lexicographic order of the genomic sequences.

The attribution is given according to:

```

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

```

An example of such an input file is:

```

@SRR001661.1 071112_SLXA-EAS1_s_7:5:1:817:345
GGGTGATGGCCGCTGCCGATGGCGTCAAATCCCACCAAGTTACCTTAACAACCTTAAGGG
+
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII9IG9ICIIIIIIIIIIIIIIIIIIIIIDIII
@SRR001661.2 071112_SLXA-EAS1_s_7:5:1:801:338
GTTTCAGGGATACGACGTTTGTATTTTAAAGATCTGAAGCAGAAGTCGATGATAATACGCG
+
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII6IBIIIIIIIIIIIIIIIIIIIIIIIGI
@SRR001661.3 071112_SLXA-EAS1_s_7:5:1:821:328
AACGCGTATTTCGGAGCTTCTTCGTTGGGTACGTGCGCCTATTATGCGGCGCGATTGCTAT
+
IIIIIII6BBB6BBBBBBBBBBBBBBBBBDDDDDDDDDDDDDDDDDDDDDDDDDDDDDDDDDD
@SRR001661.4 071112_SLXA-EAS1_s_7:5:1:943:128
ATCGCGCATTTCGACTGGTACGTGTACGTGTAGTCGTAGCGTATGTTCCGGTCGTATGCGTG
+
II77777LPMMMPMMMMIIIIIIIIIIII77777777BBBBBBBDDDDDDIIIIII

```

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:

```

@SRR001661.3 071112_SLXA-EAS1_s_7:5:1:821:328
AACGCGTATTTCGGAGCTTCTTCGTTGGGTACGTGCGCCTATTATGCGGCGCGATTGCTAT
+
IIIIIII6BBB6BBBBBBBBBBBBBBBBBDDDDDDDDDDDDDDDDDDDDDDDDDDDDDDDDDD
@SRR001661.4 071112_SLXA-EAS1_s_7:5:1:943:128
ATCGCGCATTTCGACTGGTACGTGTACGTGTAGTCGTAGCGTATGTTCCGGTCGTATGCGTG
+
II77777LPMMMPMMMMIIIIIIIIIIII77777777BBBBBBBDDDDDDIIIIII
@SRR001661.1 071112_SLXA-EAS1_s_7:5:1:817:345
GGGTGATGGCCGCTGCCGATGGCGTCAAATCCCACCAAGTTACCTTAACAACCTTAAGGG
+
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII9IG9ICIIIIIIIIIIIIIIIIIIIIIDIII

```

```
@SRR001661.2 071112_SLXA-EAS1_s_7:5:1:801:338
GTTCCAGGGATACGACGTTTGTATTTTAAAGATCTGAAGCAGAAGTCGATGATAATACGCG
+
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII6IBIIIIIIIIIIIIIIIIIIIIIGI
```

2.23 Program gto_fastq_complement

The `gto_fastq_complement` replaces the ACGT bases with their complements in a FASTQ file format. For help type:

```
./gto_fastq_complement -h
```

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

Input parameters

The `gto_fastq_complement` 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_complement [options] [--] args]
or: ./gto_fastq_complement [options]

It replaces the ACGT bases with their complements in a FASTQ file format.

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

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

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

An example of such an input file is:

[illegible]

Output

The output of the `gto_fastq_complement` program is the FASTQ file with the ACGT base complements. 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
CCCACTACCGGCGACGGCTACCGCAGTTAGGGTGGTTCAATGGGAATTGTTGAATCCCAAAAGTTTATCT
+SRR001666.1 071112_SLXA-EAS1_s_7:5:1:817:345 length=72
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII>IIIIII/
@SRR001666.2 071112_SLXA-EAS1_s_7:5:1:801:338 length=72
CAAGTCCCTATGCTGCAAACATAAAATTCTTAGACTTCGTCTTCAGCTACTATTATGCGCAGCAAAATAGTA
+SRR001666.2 071112_SLXA-EAS1_s_7:5:1:801:338 length=72
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII>IIIII-I)8I
```

2.24 Program gto_fastq_reverse

The `gto_fastq_reverse` reverses the ACGT bases order for each read in a FASTQ file format. For help type:

```
./gto_fastq_reverse -h
```

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

Input parameters

The `gto_fastq_reverse` 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_reverse [options] [--] args]
or: ./gto_fastq_reverse [options]

It reverses the ACGT bases order for each read in a FASTQ file.

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

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

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

An example of such an input file is:

```
@SRR001666.1 071112_SLXA-EAS1_s_7:5:1:817:345 length=72
GGGTGATGGCCGCTGCCGATGGCGTCAAATCCCACCAAGTTACCCTTAACAACCTTAAGGGTTTTCAAATAGA
+SRR001666.1 071112_SLXA-EAS1_s_7:5:1:817:345 length=72
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII>IIIIII/
@SRR001666.2 071112_SLXA-EAS1_s_7:5:1:801:338 length=72
GTTTCAGGGATACGACGTTTGTATTTTAAAGAATCTGAAGCAGAAGTCGATGATAATACGCGTCGTTTTATCAT
+SRR001666.2 071112_SLXA-EAS1_s_7:5:1:801:338 length=72
IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII>IIIII-I)8I
```

Output

The output of the `gto_fastq_reverse` program is the FASTQ file complement with the flag "(Reversed)" added in the header.

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 (Reversed)
AGATAAACTTTTGGGAATTCAACAATTCCCATTGAACCACCCTAAACTGCGGTAGCCGTCGCCGGTAGTG
+
/IIIII>IIIIIDIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII
@SRR001666.2 071112_SLXA-EAS1_s_7:5:1:801:338 length=72 (Reversed)
TACTATTTTGCTGCGCATAATAGTAGCTGAAGACGAAGTCTAAGAATTTTATGTTGCAGCATAGGGACTTG
+
I8)I-IIII>IIGIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII
```


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.
7. `gto_fasta_rand_extra_chars`: it substitues in the DNA sequence the outside ACGT chars by random ACGT symbols.
8. `gto_fasta_extract_read_by_pattern`: it extracts reads from a Multi-FASTA file format given a pattern in the header.
9. `gto_fasta_find_n_pos`: it reports the "N" regions in a sequence or FASTA (seq) file.
10. `gto_fasta_split_reads`: it splits a Multi-FASTA file to multiple FASTA files.
11. `gto_fasta_rename_human_headers`: it changes the headers of FASTA or Multi-FASTA file to simple chrX by order, where X is the number.

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.
13. `gto_fasta_complement`: it replaces the ACGT bases with their complements in FASTA or Multi-FASTA file format.
14. `gto_fasta_reverse`: it reverses the order of a FASTA or Multi-FASTA file format.

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 parameters.

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:

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

It converts a FASTA or Multi-FASTA file format to a seq.

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

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

Example: ./gto_fasta_to_seq < input.mfasta > output.seq
```

An example of such an input file is:

```
>AB000264 |acc=AB000264|descr=Homo sapiens mRNA
ACAAGACGGCCTCCTGCTGCTGCTGCTCTCCGGGGCCACGGCCCTGGAGGTCCACCGCTGCCCTGCTGCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGGCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGTGCC
GCGAATCCGCGCGCCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACCTTCTTCTGGAAGACCTTCTCCACCCCCCAGC
TAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
>AB000263 |acc=AB000263|descr=Homo sapiens mRNA
ACAAGATGCCATTGTCCCCGGGCCTCCTGCTGCTGCTGCTCTCCGGGGCCACGGCCACCGCTGCCCTGCCCTGGAGGGT
GGCCCCACCGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAATAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTG
GTGGTTTGAGTGGACCTCCAGGCCAGTGCCGGGGCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAG
GCGCACCCCCCAGCAATCCGCGCGCCGGGACAGAATGCCTGCAGGAACCTTCTTCTGGAAGACCTTCTCCTCTGCAAA
```

```
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:

```
ACAAGACGGCCTCCTGCTGCTGCTGCTCCTCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGCCCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGTGCC
GCGAATCCGCGCGCCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACCTTCTTCTGGAAGACCTTCTCCACCCCCCAGC
TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAAACAAGATGCCATTGTCCCCCGGCCTCCTGCTG
CTGCTGCTCCTCGGGGCCACGGCCACCGCTGCCCTGCCCTGGAGGGTGGCCCCACGGCCGAGACAGCGAGCATATGCA
GGAAGCGGCAGGAATAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGTGGTTTGAGTGGACCTCCAGGCCAGTGCCG
GGCCCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGGCGCACCCCCCAGCAATCCGCGCGCCGGGAC
AGAATGCCCTGCAGGAACCTTCTTCTGGAAGACCTTCTCCTCTGCAAATAAAAACCTCACCCATGAATGCTCACGCAAGTT
TAATTACAGACCTGAA
```

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 parameters.

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:

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

It converts a genomic sequence to pseudo FASTA file format.

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

Basic options

```
< input.seq        Input sequence file (stdin)
> output.fasta     Output FASTA file format (stdout)
```

Optional options

```
-n, --name=<str>    The read's header
-l, --lineSize=<int> The maximum of chars for line
```

```
Example: ./gto_fasta_from_seq -l <lineSize> -n <name> < input.seq > output.fasta
```

An example of such an input file is:

```
ACAAGACGGCCTCCTGCTGCTGCTGCTCTCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGTAGTGGACCTCCGGGGCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGTGCC
GCGAATCCGCGCGCCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCACCCCCCAGC
TAAACCTCACCCTGAATGCTCAGCAAGTTTAATTACAGACCTGAAACAAGATGCCATTGTCCCCCGGCCTCCTGCTG
CTGCTGCTCTCCGGGGCCACGGCCACCGCTGCCCTGCCCTGGAGGGTGGCCCCACCGCCGAGACAGCGAGCATATGCA
GGAAGCGGCAGGAATAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGTGGTTTGAGTGGACCTCCAGGCCAGTGCCG
GGCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGGCGCACCCCCCAGCAATCCGCGCGCCGGGAC
AGAATGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCTCCTGCAAATAAAACCTCACCCATGAATGCTCAGCAAGTT
TAATTACAGACCTGAA
```

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:

```
>SeqToFasta
ACAAGACGGCCTCCTGCTGCTGCTGCTCTCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGTAGTGGACCTCCGGGGCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGTGCC
GCGAATCCGCGCGCCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCACCCCCCAGC
TAAACCTCACCCTGAATGCTCAGCAAGTTTAATTACAGACCTGAAACAAGATGCCATTGTCCCCCGGCCTCCTGCTG
CTGCTGCTCTCCGGGGCCACGGCCACCGCTGCCCTGCCCTGGAGGGTGGCCCCACCGCCGAGACAGCGAGCATATGCA
GGAAGCGGCAGGAATAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGTGGTTTGAGTGGACCTCCAGGCCAGTGCCG
GGCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGGCGCACCCCCCAGCAATCCGCGCGCCGGGAC
AGAATGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCTCCTGCAAATAAAACCTCACCCATGAATGCTCAGCAAGTT
TAATTACAGACCTGAA
```

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 parameters.

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`

An example of such an input file is:

```
>AB000264 |acc=AB000264|descr=Homo sapiens mRNA
ACAAGACGGCCTCCTGCTGCTGCTCTCCGGGGCCACGGCCCTGGAGGTCCACCGCTGCCCTGCTGCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGGCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGTGCC
GCGAATCCGCGCGCCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAAGTCTTCTGGAAGACCTTCTCCACCCCCCAGC
TAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
```

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:

```
ACAAGACGGCCTCCTGCTGCTGCTGCTCTCCGGGGCCACGGCCCTGGAGG
```

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 parameters.

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
```

An example of such an input file is:

```
>AB000264 |acc=AB000264|descr=Homo sapiens mRNA
ACAAGACGGCCTCCTGCTGCTGCTGCTCTCCGGGGCCACGGCCCTGGAGGTCCACCGCTGCCCTGCTGCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCGGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGGCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGTGCC
GCGAATCCGCGCGCCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACCTTCTTCTGGAAGACCTTCTCCACCCCCCAGC
TAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
>AB000263 |acc=AB000263|descr=Homo sapiens mRNA
ACAAGATGCCATTGTCCCCGGGCCTCCTGCTGCTGCTGCTCTCCGGGGCCACGGCCACCGCTGCCCTGCCCTGGAGGGT
GGCCCCACCGCGCGGAGACAGCGAGCATATGCAGGAAGCGGCAGGAATAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTG
GTGGTTTGAGTGGACCTCCAGGCCAGTGCCGGGGCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAG
GCGCACCCCCCAGCAATCCGCGCGCCGGGACAGAATGCCTGCAGGAACCTTCTTCTGGAAGACCTTCTCCTCCTGCAAA
TAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
```

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
ACAAGACGGCCTCCTGCTGCTGCTGCTCTCCGGGGCCACGGCCCTGGAGG
>AB000263 |acc=AB000263|descr=Homo sapiens mRNA
ACAAGATGCCATTGTCCCCGGGCCTCCTGCTGCTGCTGCTCTCCGGGGCC
```

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 parameters.

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             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              : value
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:

```
>AB000264 |acc=AB000264|descr=Homo sapiens mRNA
ACAAGACGGCCTCCTGCTGCTGCTGCTCCTCGGGGCCACGGCCCTGGAGGTTCCACCGCTGCCCTGCTGCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGCCCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGTGCC
GCGAATCCGCGCGCCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAAGTTCTTCTGGAAGACCTTCTCCACCCCCCAGC
TAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
>AB000263 |acc=AB000263|descr=Homo sapiens mRNA
ACAAGATGCCATTGTCCCCCGGCCTCCTGCTGCTGCTGCTCCTCGGGGCCACGGCCACCGCTGCCCTGCCCTGGAGGGT
GGCCCCACCGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAATAAGGAAAAGCAGCCTCCTGACTTTCTCGCTTG
GTGGTTTGAGTGGACCTCCAGGCCAGTGCCGGGCCCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAG
GCGCACCCCCCAGCAATCCGCGCGCCGGGACAGAATGCCTGCAGGAAGTTCTTCTGGAAGACCTTCTCCTCCTGCAAA
TAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
```

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
AVG 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 they are optional.

For help type:

```
./gto_fasta_mutate -h
```

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

Input parameters

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

The attribution is given according to:

```

Usage: ./gto_fasta_mutate [options] [--] args]
       or: ./gto_fasta_mutate [options]

Creates a synthetic mutation of a fasta file given specific rates of editions,
deletions and additions

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

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

Optional
    -s, --seed=<int>          Starting point to the random generator
    -e, --edit-rate=<dbl>     Defines the edition rate (default 0.0)
    -d, --deletion-rate=<dbl> Defines the deletion rate (default 0.0)
    -i, --insertion-rate=<dbl> Defines the insertion rate (default 0.0)
    -a, --ACGTN-alphabet      When active, the application uses the ACGTN alphabet

Example: ./gto_fasta_mutate -s <seed> -e <edit rate> -d <deletion rate> -i
<insertion rate> -a < input.mfasta > output.fasta

```

An example of such an input file is:


```
>AB000264 |acc=AB000264|descr=Homo sapiens mRNA
ACAAGACGGCCTCCTGCTGCTGCTGCTCCTCGGGGCCACGGCCCTGGAGGTTCCACCGCTGCCCTGCTGCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGGCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGTGCC
GCGAATCCGCGCGCCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACCTTCTTCTGGAAGACCTTCTCCACCCCCCAGC
TAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
>AB000263 |acc=AB000263|descr=Homo sapiens mRNA
ACAAGATGCCATTGTCCCCCGGCCTCCTGCTGCTGCTGCTCCTCGGGGCCACGGCCACCGCTGCCCTGCCCTGGAGGGT
GGCCCCACCGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAATAAGGAAAAGCAGCCTCCTGACTTTCTCGCTTG
GTGGTTTGAGTGGACCTCCAGGCCAGTGCCGGGGCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAG
GCGCACCCCCCAGCAATCCGCGCGCCGGGACAGAATGCCCTGCAGGAACCTTCTTCTGGAAGACCTTCTCCTCCTGCAAA
TAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
```

Output

The output of the `gto_fasta_mutate` program is a FASTA or Multi-FASTA file with 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:

```
>AB000264 |acc=AB000264|descr=Homo sapiens mRNA
ACGCAACGNATTCTGCTGATCATANTGTNCCGCNCCCCNGCGACGGGGNCTCNCNNGCACACATNGTACCATTGTCCAC
NCTTNCANGTNANCGCTAGCAGGCTACNGTTTTNTCCTCNCCTANNCCAANCNGGCGTNNNTACTGGCACGTGCAGGCA
TNGGTCGGCNGGNCTCCGNAACGGCACGGAGACGAAGCTCGGNGGNTATACAGGTGTCANGAAACATCCCCGCGNC
GNGTGNCNNGAANCCANAGAGTATCTCACTCACAACCTGCGTGACANTCTAGAGNANGACCTTACNCAACNTCCCNTT
NNGTACCACACCAATGAACGCTGCAGAAAGTCTGTTTNNAGGNGNGCA
>AB000263 |acc=AB000263|descr=Homo sapiens mRNA
ATTTGAAGGCAANCGGNCCAGNAATNCGGNGGGTGCGCTCNTGTNGGCTACGGNCATCGCGGCCCTGCTNTANTAAGCN
TGAACCACCGNTCGNNGCACTTAGCAATNGCGNAANCCGTCGGCACGGCGGAGACNAANCCGCTANTNNTTTCCCGCTNA
ATGGNTGTACAAGACCNACTANACCANCTCCGTCAACCACTGGAGCGCANGATGNNCGCTGNCTAGNAGCNNTGAG
GCGCTCCNTCCTANAAANCCGTGGNCGAGCNCCCTATGGNAGNGTGGGGGTTTTACCGGAAGACNTCGNGCCCTATGGG
AGCAATCANAANCTAGAAAGCTTACNGATGGTGANGAANTAGACTANG
```

3.7 Program `gto_fasta_rand_extra_chars`

The `gto_fasta_rand_extra_chars` substitutes 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 parameters.

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]
```

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

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

Basic options

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

Example: ./gto_fasta_rand_extra_chars < input.mfasta > output.mfasta

An example of such an input file is:

```
>AB000264 |acc=AB000264|descr=Homo sapiens mRNA
ANAAGACGGCCTCCTGCTGCTGCTCTCCGGGGCCACGNCCCTGGAGGGTCCNCCGCTGCCCTGCTGCCATTGNCNCC
NGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCTCGCTTGGGCCGAGACAGCGAGCATATGCNGGAAGCGGCAGGAA
GNGGTTTGAGTGGACCTCCNGGGCCCCTCATAGGAGAGGAAGCNGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGTGNC
GCGAATCCGNGCGCCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACCTTCTTCTGGAAGACCTTCTCCACCCCCCENN
TAAANNNTACCCATGAATGCTCACGCAANTTTAATTACAGACCTGAA
>AB000263 |acc=AB000263|descr=Homo sapiens mRNA
GCGAATCCGNGCGCCGGGACAGAATCTCCTTCTCCACCCCCCENNNTGCAAAGCCCTGCAGGAACCTTCTTCTGGAAGACC
NGCCCCACCTAAGGAAAAGCAGCCTCCAGGAACCTGACTTTCTCGCTTGGGCCGAGACAGCGAGCATATGCNGGAAGCGG
ANAAGACGGCCTCCTGCTGCTGCTGCTCTCCGGGGCCACGNCCCTGGCNCAGGGTCCNCCGCTGCCCTGCTGCCATTGN
GAGGAAGCNGGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGTGNCNGGTTTGAGTGGACCTCCNGGGCCCCTCATAGGA
TCACGCAANTTTAATTACAGACCTGAATAAANNNTACCCATGAATGC
```

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:

```
>AB000264 |acc=AB000264|descr=Homo sapiens mRNA
ATAAGACGGCCTCCTGCTGCTGCTCTCCGGGGCCACGGCCCTGGAGGGTCCCCCGCTGCCCTGCTGCCATTGTCCCC
TGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCTCGCTTGGGCCGAGACAGCGAGCATATGCGGGAAGCGGCAGGAA
GAGGTTTGAGTGGACCTCCCGGGCCCCTCATAGGAGAGGAAGCCGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGTGTC
GCGAATCCGGGCGCCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACCTTCTTCTGGAAGACCTTCTCCACCCCCCTTG
TAAAAGATACCCATGAATGCTCACGCAANTTTAATTACAGACCTGAA
>AB000263 |acc=AB000263|descr=Homo sapiens mRNA
GCGAATCCGTGCGCCGGGACAGAATCTCCTTCTCCACCCCCCATCTGCAAAGCCCTGCAGGAACCTTCTTCTGGAAGACC
GGCCCCACCTAAGGAAAAGCAGCCTCCAGGAACCTGACTTTCTCGCTTGGGCCGAGACAGCGAGCATATGCGGGAAGCGG
AGAAGACGGCCTCCTGCTGCTGCTGCTCTCCGGGGCCACGTCCCTGGCTCCAGGGTCCCTCGCTGCCCTGCTGCCATTGC
GAGGAAGCGGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGTGCGCGGTTTGAGTGGACCTCCTGGCCCCCTCATAGGA
TCACGCAACTTTAATTACAGACCTGAATAAATGTACCCATGAATGC
```

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 parameters.

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:

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

It extracts reads from a Multi-FASTA file format given a pattern in the header (ID).

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

Basic options
-p, --pattern=<str>       Pattern to search in the file header
< input.fasta             Input Multi-FASTA file format (stdin)
> output.fasta            Output Multi-FASTA file format (stdout)

Example: ./gto_fasta_extract_read_by_pattern -p <pattern> < input.mfasta > output.fasta
```

An example of such an input file is:

```
>AB000264 |acc=AB000264|descr=Homo sapiens mRNA
ACAAGACGGCCTCCTGCTGCTGCTGCTCTCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGCCCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGTGCC
GCGAATCCGCGCGCCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACCTTCTTCTGGAAGACCTTCTCCACCCCCCAGC
TAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
>AB000263 |acc=AB000263|descr=Homo sapiens mRNA
ACAAGATGCCATTGTCCCCGGCCTCCTGCTGCTGCTGCTCTCCGGGGCCACGGCCACCGCTGCCCTGCCCTGGAGGGT
GGCCCCACCGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAATAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTG
GTGGTTTGAGTGGACCTCCAGGCCAGTGCCGGGCCCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAG
GCGCACCCCCCAGCAATCCGCGCGCCGGGACAGAATGCCTGCAGGAACCTTCTTCTGGAAGACCTTCTCCTCCTGCAAA
TAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
```

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:

```
>AB000264 |acc=AB000264|descr=Homo sapiens mRNA
ACAAGACGGCCTCCTGCTGCTGCTGCTCTCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAAGTGGACCTCCGGGGCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGTGCC
GCGAATCCGCGCGCCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACCTTCTTCTGGAAGACCTTCTCCACCCCCCAGC
TAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
```

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 parameters.

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:

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

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

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

Basic options
  < input.fasta      Input FASTQ file format or a sequence (stdin)
  > output           Output report of 'N' positions (stdout)

Example: ./gto_fasta_find_n_pos < input.fasta > output

The output obeys the following structure:
Begin   End Positions
<value> <value> <value>
```

An example of such an input file is:

```
>AB000264 |acc=AB000264|descr=Homo sapiens mRNA
NCNNNACGGCCTCCTGCTGCTGCTGCTCTCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC
GNCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTNGTTTGAAGTGGACCTCCGGGGCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGTGCC
GCGAATCCGCGCGCCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACNTCTTCTGGAAGACCTTCTCCACCCCCCAGC
TAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAN
```

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
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 parameters.

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:

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

It splits a Multi-FASTA file to multiple FASTA files.

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

Basic options
    < input.fasta             Input Multi-FASTA file format (stdin)

Optional options
    -l, --location=<str>     Location to store the files

Example: ./gto_fasta_split_reads < input.mfasta
```

An example of such an input file is:

```
>AB000264 |acc=AB000264|descr=Homo sapiens mRNA
ACAAGACGGCCTCCTGCTGCTGCTGCTCCTCGGGGCCACGGCCCTGGAGGTCACCGCTGCCCTGCTGCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
```

```

GTGGTTTGAGTGGACCTCCGGGGCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGTGCC
GCGAATCCGCGCGCCGGGACAGAATCTCTGCAAAGCCCTGCAGGAACCTTCTTCTGGAAGACCTTCTCCACCCCCCAGC
TAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
>AB000263 |acc=AB000263|descr=Homo sapiens mRNA
ACAAGATGCCATTGTCCCCGGCCTCCTGTGCTGCTGCTCTCCGGGGCCACGGCCACCGCTGCCCTGCCCTGGAGGGT
GGCCCCACCGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAATAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTG
GTGGTTTGAGTGGACCTCCCAGGCCAGTGCCGGGGCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAG
GCGCACCCCCCAGCAATCCGCGCGCCGGGACAGAATGCCCTGCAGGAACCTTCTTCTGGAAGACCTTCTCCTCCTGCAAA
TAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA

```

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 parameters.

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:

```
> AB000264 | acc = AB000264 | descr = Homo sapiens mRNA
ACAAGACGGCCTCCTGCTGCTGCTCTCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGGCCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGTGCC
GCGAATCCGCGCGCCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAAGTCTTCTGGAAGACCTTCTCCACCCCCCAGC
TAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
> AB000263 | acc = AB000263 | descr = Homo sapiens mRNA
ACAAGATGCCATTGTCCCCCGGCCTCCTGCTGCTGCTGCTCTCCGGGGCCACGGCCACCGCTGCCCTGCCCCTGGAGGGT
GGCCCCACCGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAATAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTG
GTGGTTTGAGTGGACCTCCCAGGCCAGTGCCGGGGCCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAG
GCGCACCCCCCAGCAATCCGCGCGCCGGGACAGAATGCCTGCAGGAAGTCTTCTGGAAGACCTTCTCCTCCTGCAAA
TAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
```

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:

```
>chr1
ACAAGACGGCCTCCTGCTGCTGCTGCTCTCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGGCCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGTGCC
GCGAATCCGCGCGCCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAAGTCTTCTGGAAGACCTTCTCCACCCCCCAGC
TAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
>chr2
ACAAGATGCCATTGTCCCCCGGCCTCCTGCTGCTGCTGCTCTCCGGGGCCACGGCCACCGCTGCCCTGCCCCTGGAGGGT
GGCCCCACCGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAATAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTG
GTGGTTTGAGTGGACCTCCCAGGCCAGTGCCGGGGCCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAG
GCGCACCCCCCAGCAATCCGCGCGCCGGGACAGAATGCCTGCAGGAAGTCTTCTGGAAGACCTTCTCCTCCTGCAAA
TAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
```

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 parameters.

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
```

Basic options

```
-p, --pattern=<str>  Pattern to search in the file header
< input.fasta        Input Multi-FASTA file format (stdin)
> output.coords       Output coordinates (stdout)
```

```
Example: ./gto_fasta_extract_pattern_coords -p <pattern> < input.fasta > output.coords
```

An example of such an input file is:

```
>AB000264 |acc=AB000264|descr=Homo sapiens mRNA
ACAAGACGGCCTCTGCTGCTGCTCTCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGGCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGTGCC
GCGAATCCGCGCGCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACCTTCTTCTGGAAGACCTTCTCCACCCCCCAGC
TAAACCTCACCCATGAATGCTCGCAACACGCAAGTTTAATTCGCAAGTTAGACCTGAACGGGAGGTGGCCACGCAAGTT
```

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
```

3.13 Program `gto_fasta_complement`

The `gto_fasta_complement` replaces the ACGT bases with their complements in FASTA or Multi-FASTA file format.

For help type:

```
./gto_fasta_complement -h
```

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

Input parameters

The `gto_fasta_complement` 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_complement [options] [--] args]
       or: ./gto_fasta_complement [options]

It replaces the ACGT bases with their complements in FASTA or Multi-FASTA file format.

    -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_complement < input.mfasta > output.mfasta
```

An example of such an input file is:

```
>AB000264 |acc=AB000264|descr=Homo sapiens mRNA
ACAAGACGGCTCCTGCTGCTGCTCCTCGGGGCCACGGCCCTGGAGGTCCACCGCTGCCCTGCTGCCATTGTCCC
CGGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCGGAGACAGCGAGCATATGCAGGAAGCGGCAGG
AAGTGGTTTGAGTGGACCTCCGGGCCCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGT
GCCGCGAATCCGCGCGCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACCTTCTTCTGGAAGACCTTCTCCACCCCCC
CAGCTAAAACCTCACCCATGAATGCTCAGCAAGTTTAATTACAGACCTGAA
>AB000263 |acc=AB000263|descr=Homo sapiens mRNA
ACAAGATGCCATTGTCCCCGGCCTCCTGCTGCTGCTGCTCCTCGGGGCCACGGCCACCGCTGCCCTGCCCTGGAGGG
TGGCCCCACCGCGCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAATAAGGAAAAGCAGCCTCCTGACTTTCCTCGCT
TGGTGGTTTGAGTGGACCTCCAGGCCAGTGCCGGGCCCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGG
AAGGCGCACCCCCCAGCAATCCGCGCGCGGGACAGAATGCCCTGCAGGAACCTTCTTCTGGAAGACCTTCTCCTCCTG
CAAAATAAACCTCACCCATGAATGCTCAGCAAGTTTAATTACAGACCTGAA
```

Output

The output of the `gto_fasta_complement` program is FASTA or Multi-FASTA file with the ACGT base complements.

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

```
>AB000264 |acc=AB000264|descr=Homo sapiens mRNA
TGTTCTGCCGGAGGACGACGACGACGAGAGGCCCGGTGCCGGACCTCCCAGGTGGCGACGGGACGAGGTAACAGGG
GCCGGGGTGGATTCTTTTCGTCGGAGGACTGAAAGGAGCGAACCGGCTCTGTCGCTCGTATACGTCCTTCGCCGTCC
TTCACCAAACCTCACCTGGAGGCCCGGGAGTATCCTCTCCTTCGAGCCCTCCACCGGTCCGCCGTCCTTCGTCCGGTCA
CGGCGCTTAGGCGCGCGGCCCTGTCTTAGAGGACGTTTCGGGACGTCCTTGAAGAAGACCTTCTGGAAGAGGTGGGGG
GTCGATTTTGGAGTGGGTACTTACGAGTGCGTTCAAATTAATGTCTGGACTT
>AB000263 |acc=AB000263|descr=Homo sapiens mRNA
TGTTCTACGGTAACAGGGGGCCGGAGGACGACGACGACGAGAGGCCCGGTGCCGGTGGCGACGGGACGGGGACCTCCC
ACCGGGGTGGCCGGCTCTGTCGCTCGTATACGTCCTTCGCCGTCCTTATTCCTTTTCGTCGGAGGACTGAAAGGAGCGA
ACCACCAAACCTCACCTGGAGGGTCCGGTCACGGCCCGGGAGTATCCTCTCCTTCGAGCCCTCCACCGGTCCGCCGTCC
TTCCGCGTGGGGGGGTCTTAGGCGCGCGGCCCTGTCTTACGGGACGTCCTTGAAGAAGACCTTCTGGAAGAGGAGGAC
GTTTATTTTGGAGTGGGTACTTACGAGTGCGTTCAAATTAATGTCTGGACTT
```

3.14 Program gto_fasta_reverse

The `gto_fasta_reverse` reverses the ACGT bases order for each read in a FASTA or Multi-FASTA file format.

For help type:

```
./gto_fasta_reverse -h
```

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

Input parameters

The `gto_fasta_reverse` 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_reverse [options] [--] args]
       or: ./gto_fasta_reverse [options]

It reverses the ACGT bases order for each read in a FASTA or Multi-FASTA file.

    -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_reverse < input.mfasta > output.mfasta
```

An example of such an input file is:

```
>AB000264 |acc=AB000264|descr=Homo sapiens mRNA
ACAAGACGGCCTCTGCTGCTGCTCTCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCC
CGGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCGGAGACAGCGAGCATATGCAGGAAGCGGCAGG
AAGTGGTTTGAGTGGACCTCCGGGGCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGT
GCCGCGAATCCGCGCGCCGGGACAGAATCTCTGCAAAGCCCTGCAGGAAGTCTTCTGGAAGACCTTCTCCACCCCCC
CAGCTAAAACCTCACCCATGAATGCTCAGCAAGTTTAATTACAGACCTGAA
>AB000263 |acc=AB000263|descr=Homo sapiens mRNA
ACAAGATGCCATTGTCCCCGGGCCTCTGCTGCTGCTCTCCGGGGCCACGGCCACCGCTGCCCTGCCCTGGAGGG
TGGCCCCACCGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAATAAGGAAAAGCAGCCTCCTGACTTTCCTCGCT
TGGTGGTTTGAGTGGACCTCCAGGCCAGTCCGGGGCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGG
AAGGCGCACCCCCCAGCAATCCGCGCGCCGGGACAGAATGCCCTGCAGGAAGTCTTCTGGAAGACCTTCTCCTCCTG
CAATAAAAACCTCACCCATGAATGCTCAGCAAGTTTAATTACAGACCTGAA
```

Output

The output of the `gto_fasta_reverse` program is FASTA or Multi-FASTA file with the bases reversed and the flag "(Reversed)" added in the header.

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

```
>AB000264 |acc=AB000264|descr=Homo sapiens mRNA (Reversed)
AAGTCCAGACATTAATTTGAACGCACTCGTAAGTACCCACTCCAAAATCGACCCCCCACCTCTTCCAGAAGGTCTTCT
TCAAGGACGTCCCGAAACGTCCTCTAAGACAGGGCCGCGCGCCTAAGCGCCGTGACCGGACGAAGGACGGCGGACCGGT
GGAGGGCTCGAAGGAGAGGATACTCCCCGGGCCTCCAGGTGAGTTTGGTGAAGGACGGCGAAGGACGTATACGAGCGAC
AGAGCCGGGTTCGCTCCTTTTCAGTCCTCCGACGAAAAGGAATCCACCCGGCCCCCTGTTACCGTCGTCCCGTCGCCACC
TGGGAGGTCCCGGCACCGGGGCCTCTCGTCGTCGTCGTCCTCCGGCAGAACA
>AB000263 |acc=AB000263|descr=Homo sapiens mRNA (Reversed)
AAAGTCCAGACATTAATTTGAACGCACTCGTAAGTACCCACTCCAAAATAAACGTCCTCCTCTTCCAGAAGGTCTTCTT
CAAGGACGTCCCGTAAGACAGGGCCGCGCGCCTAACGACCCCCCACGCGGAAGGACGGCGGACCGGTGGAGGGCTCGA
AGGAGAGGATACTCCCCGGGCCGTGACCGGACCCTCCAGGTGAGTTTGGTGGTTCGCTCCTTTTCAGTCCTCCGACGAAA
AGGAATAAGGACGGCGAAGGACGTATACGAGCGACAGAGCCGGCCACCCGGTGGGAGGTCCCCGTCCCGTCGCCACCG
GCACCGGGGCCTCTCGTCGTCGTCGTCCTCCGGCCCCCTGTTACCGTAGAACA
```

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 substitutes 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.
7. `gto_genomic_compressor`: it compress and decompress genomic sequences for storage purposes (also under the alias `gto_geco`).
8. `gto_genomic_complement`: it replaces the ACGT bases with their complements in a DNA sequence.
9. `gto_genomic_reverse`: it reverses the ACGT bases order for each read in a sequence file (also under the alias `gto_reverse`).

4.1 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 parameters.

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)
    -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 <nsymbols> -f <fa,fc,fg,ft> > output.seq
```

Output

The output of the `gto_genomic_gen_random_dna` program is a sequence group file with 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:

```
TCTTTACTCGCGCGTTGGAGAAATACAATAGTGCGGCTCTGTCTCCTTATGAAGTCAACAATTTGCTGGGACTTGCGGC
TCTTTACTCGCGCGTTGGAGAAATACAATAGTGCGGCTCTGTCTCCTTATGAAGTCAACAATTTGCTGGGACTTGCGGC
GACTTCATCGTGGTCTCTGTCTCATTATGCGCTCCAACGCATAACTTTGCGCCAGAAGATAGATAGAATGGTGTAAAGAACT
GTAATATATATAATGAACTTCGGCGAGTCTGTGGAGTTTTTGTGTCATTAGAGAGCCAAGAGGTCGGACGTCCTCACGTA
GCCCCAGACGGGCAGGGCGATGGCGACTGAACGGGCTCCATATCACTTTGAGCTTTTATGCTTTTCTGACTCCTCCAGGAGC
TGAACAACCTTGTTCCTCCGGCAAAGCCCACTGCGTCATGGAGCTCACGGTCTACATTCATGACTGACTAACCCTAACTGC
```

4.2 Program `gto_genomic_rand_seq_extra_chars`

The `gto_genomic_rand_seq_extra_chars` substitutes 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 parameters.

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:

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

```
It substitutes in the DNA sequence the outside ACGT chars by random ACGT symbols.
It works in sequence file formats
```

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

Basic options

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

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

An example of such an input file is:

```
ANAAGACGNNTTCCTGCTGCTGCTGCTCCTCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC
NNCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGCCCCNNNNNGGAGAGGAAGCTCGGGAGNGTNNNGGCCAGGCGGCAGNNNNCCAGTGCC
GCGAATCCGCGCGCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACCTTCTTCTGGAAGACCTTCTCCACCCCCCAGC
TANNNNTCACCCATGAATGCTCAGCAAGTTTAATTACAGACCTGAAACAAGATGCCATTGTCCCCGGCCCTCCTGCTG
CTGCTGCTCCTCGGGGCCACGGCCACCGCTGCCCTGCCCTGGAGGGTGGCCCCACGGCCGAGACAGCGAGCATATGCA
GGAAGCGGCAGGAATAAGNNNAAGCAGCCTCCTGACTTTCCTCGCTTGNNNNTTGTAGTGGACCTCCAGGCCAGTGCCG
GGCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGGCGCACCCCCCAGCAATCCGCGCGCGGGGAC
AGAATGCCCTGCAGGAACCTTCTTCTGGAAGACCTTCTCCTCCTGCAAATAAAACCTCACCCATGAATGCTCAGCAAGTT
NNATTACNNNCCTGNN
```

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:

```
ATAAGACGGCTTCCTGCTGCTGCTGCTCCTCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC
CTCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGCCCCGACGGGAGAGGAAGCTCGGGAGTGTGTTGGCCAGGCGGCAGGAGACCAGTGCC
GCGAATCCGCGCGCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACCTTCTTCTGGAAGACCTTCTCCACCCCCCAGC
TAATATCTCACCCATGAATGCTCAGCAAGTTTAATTACAGACCTGAAACAAGATGCCATTGTCCCCGGCCCTCCTGCTG
CTGCTGCTCCTCGGGGCCACGGCCACCGCTGCCCTGCCCTGGAGGGTGGCCCCACGGCCGAGACAGCGAGCATATGCA
GGAAGCGGCAGGAATAAGCGGAAGCAGCCTCCTGACTTTCCTCGCTTGGTTTTTTGAGTGGACCTCCAGGCCAGTGCCG
```

```

GGCCCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGGCGCACCCCCCAGCAATCCGCGCGCCGGGAC
AGAATGCCCTGCAGGAACCTTCTTCTGGAAGACCTTCTCCTCCTGCAAATAAAACCTCACCCATGAATGCTCACGCAAGTT
CGATTACGGCCCTGTC

```

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 parameters.

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
    -s, --seed=<int>         Starting point to the random generator
    -m, --mutation-rate=<dbl> Defines the mutation rate (default 0.0)
    -d, --deletion-rate=<dbl> Defines the deletion rate (default 0.0)
    -i, --insertion-rate=<dbl> Defines the insertion rate (default 0.0)
    -a, --ACGTN-alphabet     When active, the application uses the ACGTN alphabet

Example: ./gto_genomic_dna_mutate -s <seed> -m <mutation rate> -d <deletion rate> -i
<insertion rate> -a < input.seq > output.seq

```

An example of such an input file is:

```

TCTTTACTCGCGCGTTGGAGAAATACAATAGTGGCGCTCTGTCTCCTTATGAAGTCAACAATTTGCTGGGACTTGCGGC
TCTTTACTCGCGCGTTGGAGAAATACAATAGTGGCGCTCTGTCTCCTTATGAAGTCAACAATTTGCTGGGACTTGCGGC
GACTTCATCGTGGTCTCTGTCTATTATGCGCTCCAACGCATAACTTTGCGCCAGAAGATAGATAGAATGGTGTAAGAAACT

```

```
GTAATATATATAATGAACTTCGGGCGAGTCTGTGGAGTTTTTGTGTCATTAGAGAGCCAAGAGGTCGGACGTCCTCACGTA
CCCCGAGACGGGCGAGGGCGATGGCGACTGAACGGGCTCCATATCACTTTGAGCTTTTATGCTTTTCGACTCCTCCAGGAGC
TGAACAACCTTGTTCCCGGCAAAAGCCCACTGCGTCATGGAGCTCACGGTCTACATTCATGACTGACTAACCGTAAACTGC
```

Output

The output of the `gto_genomic_dna_mutate` program is a sequence file which 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:

```
TCACGACTGTGCGTTGGCACACCAGATAGGTGCTTCTACGTTTTGTATCTAATTTACAATTCTCGCTGGGAGTTCATTC
GCTATTGATGGGACTAGAAACCCATCCGTAGCTTGCCGCGTTTAAAGAATAAACACTCCACTTGCACCGAGACGTAGCGC
AACCAAGGCTATGTTCTTTGACCTTATGCGGTCCAACGCAGGAGTAGACCCCGTAGTTAGGTACTATCGCAGAATAGGC
TTAAGCAGCCGTGCTGAACGCTGGAGGGTCTGTTTAATTACTGAGTGAATGGAGAGCTAAGAGTTCGGAGCACCGCACGA
GGCTCAAGAGCGGAAGGGCGTCAGCCTGGCGACCACCTGCCTACCGCTCGAGTCTGTCTTCACTACAGTCCGTGGAGGAC
CCCCAACGACCTAGTATCCTACAAAGCCGCATACGACTTACAGAACAGGCTGTATCGTCAGGAGTGTGTACACGAAGAGT
A
```

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 parameters.

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:

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

It extracts sequences from a sequence 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.seq        Input sequence file (stdin)
```



```
> output.seq      Output sequence file (stdout)
```

```
Example: ./gto_genomic_extract -i <init> -e <end> < input.seq > output.seq
```

An example of such an input file is:

```
TCTTTACTCGCGCGTTGGAGAAAATACAATAGTGGCGCTCTGTCTCCTTATGAAGTCAACAATTCGCTGGGACTTGCGGC
TCTTTACTCGCGCGTTGGAGAAAATACAATAGTGGCGCTCTGTCTCCTTATGAAGTCAACAATTCGCTGGGACTTGCGGC
GACTTCATCGTGGTCTCTGTCATTATGCGCTCCAACGCATAACTTTGCGCCAGAAGATAGATAGAATGGTGTAAGAACT
GTAATATATATAATGAACTTCGGCGAGTCTGTGGAGTTTTTGTTCATTAGAGAGCCAAGAGGTCGGACGTCCTCACGTA
GCCCCGAGACGGGCAGGGCGATGGCGACTGAACGGGCTCCATATCACTTTGAGCTTTTATGCTTTCGACTCCTCCAGGAGC
TGAACAACCTTGTTCCCGGCAAGCCCACTGCGTCATGGAGCTCACGGTCTACATTGACTGACTAACCGTAAACTGC
```

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:

```
TCTTTACTCGCGCGTTGGAGAAAATACAATAGTGGCGCTCTGTCTCCTTAT
```

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 parameters.

Input parameters

The `gto_genomic_period` 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         Output is given by log2(4)*K(x)/|x| (stdout)

Example: ./gto_genomic_period < input.seq > output

```

An example of such an input file is:

```

TCTTTACTCGCGCGTTGGAGAAATACAATAGTGCGGCTCTGTCTCCTTATGAAGTCAACAATTCGCTGGGACTTGCGGC
TCTTTACTCGCGCGTTGGAGAAATACAATAGTGCGGCTCTGTCTCCTTATGAAGTCAACAATTCGCTGGGACTTGCGGC
GACTTCATCGTGGTCTCTGTTCATTATGCGCTCCAACGCATAACTTTGCGCCAGAAGATAGATAGAATGGTGTAAAGAACT
GTAATATATATAATGAACCTTCGGCGAGTCTGTGGAGTTTTTGTTCATTAGAGAGCCAAGAGGTCGGACGTCCTCACGTA
GCCCCGAGACGGGCAGGGCGATGGCGACTGAACGGGCTCCATATCACTTTGAGCTTTTATGCTTTTCTCCAGGAGC
TGAACAACCTTGTTCCCGGCAAAGCCCACTGCGTCATGGAGCTCACGGTCTACATTTCATGACTGACTAACCGTAAACTGC

```

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
15  1.733
16  1.738
17  1.742
18  1.746
19  1.75
20  1.754

```

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

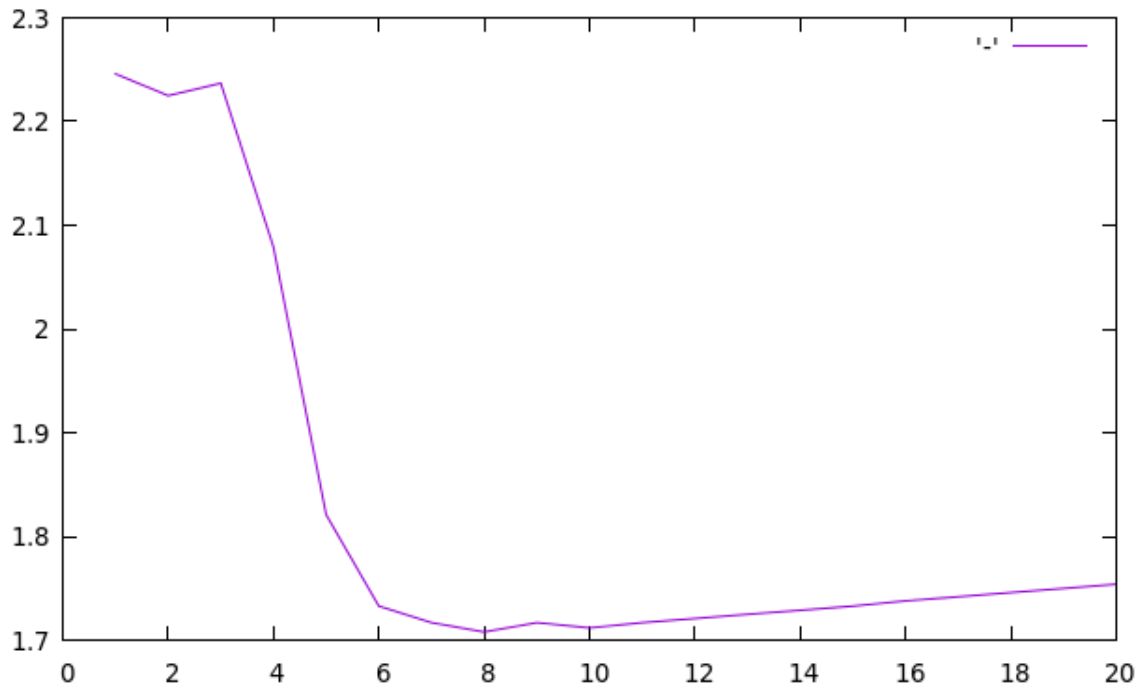


Figure 4.1: `gto_genomic_period` execution plot.

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 parameters.

Input parameters

The `gto_genomic_count_bases` 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

```

An example of such an input file is:

```

TCTTTACTCGCGCGTTGGAGAAAATACAATAGTGGGCTCTGTCTCCTTATGAAGTCAACAATTTGCTGGGACTTGCGGC
TCTTTACTCGCGCGTTGGAGAAAATACAATAGTGGGCTCTGTCTCCTTATGAAGTCAACAATTTGCTGGGACTTGCGGC
GACTTTCATCGTGGTCTCTGTCTCATTATGCGCTCCAAACGCATAACTTTGCGCCAGAAGATAGATAGAATGGTGTAAGAACT
GTAATATATATAATGAACCTTCGGCGAGTCTGTGGAGTTTTTGTGTCATTAGAGAGCCAAGAGGTCGGACGTCCTCACGTA
GCCCAGACGGGCGAGGCGATGGCGACTGAACGGGCTCCATATCACTTTGAGCTTTTATGCTTTGACTCCTCCAGGAGC
TGAACAACCTTGTTCCCGGCAAAGCCCACTGCGTCATGGAGCTCACGGTCTACATTCATGACTGACTAACCGTAAACTGC

```

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

```

4.7 Program gto_genomic_compressor

The `gto_genomic_compressor` 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_genomic_compressor -h
```

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

Input parameters

The `gto_genomic_compressor` program needs a sequence to compress.

The attribution is given according to:

SYNOPSIS

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

SAMPLE

```
Run Compression      : ./gto_genomic_compressor -v -l 3 sequence.txt
Run Decompression    : ./gto_genomic_decompressor -v sequence.txt.co
Run Information Profile : ./gto_genomic_compressor -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
    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.

-l [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 confident 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 0 does not contemplate 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_H]: (integer [1;254]) size of the cache-hash for deeper context models, namely for [NB_C] > 14. When the [NB_C] ≤ 14 use, for example, 1 as a default. The RAM is highly dependent of this value (higher value stand for higher 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 than [NB_C], while it is turned on when a complete match of size [NB_C] is seen again. This is probabilistic-algorithmic model very useful 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 analogous 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 trained 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_genomic_compressor -v -l 2 BuEb
../../bin/gto_genomic_decompressor -v BuEb.co
cmp BuEb BuEb.de -l

```

4.8 Program gto_genomic_complement

The `gto_genomic_complement` replaces the ACGT bases with their complements in a DNA sequence. It works in sequence file formats.

For help type:

```
./gto_genomic_complement -h
```

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

Input parameters

The `gto_genomic_complement` program needs two parameters, namely the input and output standard. The input stream is a sequence file.

The attribution is given according to:

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

It replaces the ACGT bases with their complements in a DNA sequence.
It works in sequence file formats

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

Basic options

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

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

An example of such an input file is:

```
TCTTTACTCGCGCGTTGGAGAAATACAATAGTGGGCTCTGTCTCCTTATGAAGTCAACAATTTGCTGGGACTTGCGG
CTCTTTACTCGCGCGTTGGAGAAATACAATAGTGGGCTCTGTCTCCTTATGAAGTCAACAATTTGCTGGGACTTGCG
GCGACTTCATCGTGGTCTCTGTCTCATTATGCGCTCCAACGCATAACTTTGCGCCAGAAGATAGATAGAATGGTGTAAAGAA
ACTGTAATATATATAATGAACCTCGGCGAGTCTGTGGAGTTTTTGTGTCATTAGAGAGCCAAGAGGTCGGACGTCCTCA
CGTAGCCCGAGACGGGCAGGGCGATGGCGACTGAACGGGCTCCATATCACTTTGAGCTTTTATGCTTTGACTCCTCCA
GGAGCTGAACAACCTTGTTCCCGGCAAAGCCCACTGCGTCATGGAGCTCACGGTCTACATTCATGACTGACTAACCGTA
AACTGC
```

Output

The output of the `gto_genomic_complement` program is a group sequence with the ACGT base complements.

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

```
AGAAATGAGCGCGCAACCTCTTTATGTTATCACGCCGAGACAGAGGAATACTTCAGTTGTTAAAGCGACCCTGAACGCC
GAGAAATGAGCGCGCAACCTCTTTATGTTATCACGCCGAGACAGAGGAATACTTCAGTTGTTAAAGCGACCCTGAACGCC
CGCTGAAGTAGCACCAGAGACAGTAATACGCGAGGTTGCGTATTGAAACGCGGTCTTCTATCTATCTTACCACATTCTT
TGACATTATATATATTACTTGAAGCCGCTCAGACACCTCAAAAACAACGTAATCTCTCGGTTCTCCAGCCTGCAGGAGT
GCATCGGGCTCTGCCGCTCCCGCTACCGCTGACTTGCCCGAGGTATAGTGAAACTCGAAAATACGAAAGCTGAGGAGGT
CCTCGACTTGTTGGAACAAGGGCCGTTTCGGGTGACGCAGTACCTCGAGTGCCAGATGTAAGTACTGACTGATTGGCAT
TTGACG
```

4.9 Program `gto_genomic_reverse`

The `gto_genomic_reverse` reverses the ACGT bases order for each read in a sequence file.

For help type:

```
./gto_genomic_reverse -h
```

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

Input parameters

The `gto_genomic_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_genomic_reverse [options] [--] args]
       or: ./gto_genomic_reverse [options]

It reverses the ACGT bases order for each read in a sequence file.

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

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

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

An example of such an input file is:

```
ACAAGACGGCCTCCTGCTGCTGCTGCTCTCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAAGTGGACCTCCGGGCCCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGTGCC
GCGAATCCGCGCGCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACCTTCTTCTGGAAGACCTTCTCCACCCCCCAGC
TAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAAACAAGATGCCATTGTCCCCCGGCCTCCTGCTG
CTGCTGCTCTCCGGGGCCACGGCCACCGCTGCCCTGCCCCTGGAGGGTGGCCCCACCGCCGAGACAGCGAGCATATGCA
GGAAGCGGCAGGAATAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGTGGTTTGAAGTGGACCTCCAGGCCAGTGCCG
GGCCCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGGCGCACCCCCCAGCAATCCGCGCGCGGGAC
AGAATGCCCTGCAGGAACCTTCTTCTGGAAGACCTTCTCCTCCTGCAAATAAACCTCACCCATGAATGCTCACGCAAGTT
TAATTACAGACCTGAA
```

Output

The output of the `gto_genomic_reverse` program is a group sequence.

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

```
AAGTCCAGACATTAATTTGAACGCACTCGTAAGTACCCACTCCAAAATAAACGTCCTCCTCTTCCAGAAGGTCTTCTTCA
AGGACGTCCCGTAAGACAGGGCCGCGCGCTAACGACCCCCCACGCGGAAGGACGGCGGACCGGTGGAGGGCTCGAAGG
AGAGGATACTCCCCGGGCCGTGACCGGACCCTCCAGGTGAGTTTGGTGGTTCTGCTCCTTTCAGTCCTCCGACGAAAAGGA
ATAAGGACGGCGAAGGACGTATACGAGCGACAGAGCCGGCCACCCCGTGGGAGGTCCCGTCCCGTCCGACCGGCACC
GGGGCCTCTCGTCGTCGTCCTCCGGCCCCCTGTTACCGTAGAACAAAGTCCAGACATTAATTTGAACGCACTCGTAA
GTACCCACTCCAAAATCGACCCCCCACCTCTTCCAGAAGGTCTTCTTCAAGGACGTCCCGAAACGTCCTCTAAGACAGG
GCCGCGCGCCTAAGCGCGTGACCGGACGAAGGACGGCGGACCGGTGGAGGGCTCGAAGGAGAGGATACTCCCCGGGCCT
CCAGGTGAGTTTGGTGAAGGACGGCGAAGGACGTATACGAGCGACAGAGCCGGGTTCTGCTCCTTTTCAGTCCTCCGACGAA
AAGGAATCCACCCGGGCCCTGTTACCGTCGTCCTCGCCACCTGGGAGGTCCCGGCACCGGGGCCTCTCGTCGTCGTC
GTCCTCCGGCAGAACA
```

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 parameters.

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 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 |
|------|--|
| R | P |
| H | P Amino acids with electric charged side chains: POSITIVE |
| K | P |
| - | - |
| D | N |
| E | N Amino acids with electric charged side chains: NEGATIVE |
| - | - |
| S | U |
| T | U |
| N | U Amino acids with electric UNCHARGED side chains |
| Q | U |
| - | - |
| C | S |
| U | S |
| G | S Special cases |
| P | S |
| - | - |
| A | H |
| V | H |
| I | H |
| L | H |
| M | H Amino acids with hydrophobic side chains |
| F | H |
| Y | H |
| W | H |
| - | - |
| * | * Others |
| X | 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:

```
IPFLLKKQFALADKLVLSKLRQLLGRIKMMPCGGAKLEPAIGLFFHAIGINIKLGYGMTETTATVSCWHDFQFNPNSIG
TLMPKAEVKIGENNEILVRGGMVMKGYKKPEETAQAFTEDGFLKTGDAGEFDEQGNLFITDRIKELMKTSNGKYIAPQY
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:

```

HSHHHPPUHHHHNPHHHUPHPUHHSSPHPHSSSSHPHNSHHSHHHHPHSHUHPHSHSHUNUUHUUHUSHPNHUHUSUUHS
UHHSPHNHPHSNUUNHHHPSSHHHPSHHPPSNNUUHHUNNSHHHPUSNHSNHNNUUUHHUNPHPNHHPUUUSPHHHSUH
HNUPHSPNPHHNUHHHHHNPPPHUHHHHSSHNUHNNHHPUHUHPHPNPHNHHPUUNHHPPHN

```

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 parameters.

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        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
A       GCA
C       TGC
D       GAC
E       GAG
F       TTT
G       GGC
H       CAT
I       ATC
K       AAA
L       CTG
M       ATG
N       AAC
P       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:

```
IPFLLKKQFALADKLVLSKLRQLLGGRICKMMPCGGAKLEPAIGLFFHAIGINIKLGYGMTETTATVSCWHDFQFNPNSIG
TLMPKAEVKIGENNEILVRGGVMKGYKKPEETAQAFTEDGFLKTGDAGEFDEQGNLFITDRIKELMKTSNGKYIAPQY
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:

```
ATCCCGTTTCTGCTGAAAAAACAGTTTGCCTGGCAGACAACTGGTACTGTCTAACTGCGTCAGCTGCTGGGCGGCCG
TATCAAAATGATGCCGTGCGGCGGCGCAAACTGGAGCCGCAATCGGCCTGTTTTTTCATGCAATCGGCATCAACATCA
AACTGGGCTACGGCATGACGGAGACGACGGCAACGGTATCTTGCTGGCATGACTTTCAGTTTAACCCGAACCTATCGGC
ACGCTGATGCCGAAAGCAGAGGTAAAAATCGGCGAGAACACGAGATCCTGGTACGTGGCGGCATGGTAATGAAAGGCTA
CTACAAAAAACCGGAGGAGACGGCACAGGCATTTACGGAGGACGGCTTTCTGAAAAACGGGCGACGCAGGCGAGTTTGACG
AGCAGGGCAACCTGTTTATCACGGACCGTATCAAAGAGCTGATGAAAACGTCTAACGGCAAATACATCGCACCGCAGTAC
ATCGAGTCTAAAATCGGCAAAGACAAATTTATCGAGCAGATCGCAATCATCGCAGACGCAAAAAATACGTATCTGCACT
GATCGTACCGTGCTTTGACTCTCTGGAGGAGTACGCAAAACAGCTGAACATCAAATACCATGACCGTCTGGAGCTGCTGA
AAAACTCTGACATCCTGAAAAATGTTTGAG
```

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 parameters.

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,
-v                verbose mode (more information),
-V                display version number,
-f                force overwrite of output,
-l <level>        level of compression [1;7] (lazy -tm setup),
-t <threshold>    threshold frequency to discard from alphabet,
-e                it creates a file with the extension ".iae"
                  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>:<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),
-tm <c>:<d>:<g>/<m>:<e>:<a>  target model (-tm 7:100:0.9/2:10:0.85),
...

                  target and reference templates use <c> for
                  context-order size, <d> for alpha (1/<d>), <g>
                  for gamma (decayment forgetting factor) [0;1),
                  <m> 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)
                  [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:

```
[Compress]    ./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 -l 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_new_line_on_new_x`: it splits different rows with a new empty row.
3. `gto_upper_bound`: it sets an upper bound in a file with a value per line.
4. `gto_lower_bound`: it sets an lower bound in a file with a value per line.
5. `gto_brute_force_string`: it generates all combinations, line by line, for an inputted alphabet and specific size.
6. `gto_real_to_binary_with_threshold`: it converts a sequence of real numbers into a binary sequence, given a threshold.
7. `gto_sum`: it adds decimal values in file, line by line, splitted by spaces or tabs.
8. `gto_filter`: it filters numerical sequences.
9. `gto_word_search`: it search for a word in a file.
10. `gto_permute_by_blocks`: it permutates by block sequence, FASTA and Multi-FASTA files.
11. `gto_info`: it gives the basic properties of the file, namely size, cardinality, distribution percentage of the symbols, among others.
12. `gto_segment`: it segments a filtered sequence.

13. `gto_comparative_map`: it creates a visualization for comparative maps.
14. `gto_max`: it computes the maximum value in each row between two files.
15. `gto_min`: it computes the minimum value in each row between two files.

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 parameters.

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:

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

It splits a sequence into lines, creating an output sequence which has a char for each line.

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

Basic options

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

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

An example of such an input file is:

```
ACAAGACGGCCTCCTGCTGCTGCTGCTCTCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCTCGCTTGGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGCCCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGTGCC
GCGAATCCGCGCGCCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACCTTCTTCTGGAAGACCTTCTCCACCCCCCAGC
TAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAAACAAGATGCCATTGTCCCCCGGCCTCCTGCTG
CTGCTGCTCTCCGGGGCCACGGCCACCGCTGCCCTGCCCTGGAGGGTGGCCCCACCGCCGAGACAGCGAGCATATGCA
GGAAGCGGCAGGAATAAGGAAAAGCAGCCTCCTGACTTTCTCGCTTGGTGGTTTGAGTGGACCTCCAGGCCAGTGCCG
GGCCCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGGCGCACCCCCCAGCAATCCGCGCGCCGGGAC
AGAATGCCCTGCAGGAACCTTCTTCTGGAAGACCTTCTCCTCCTGCAAATAAACCTCACCCATGAATGCTCACGCAAGTT
TAATTACAGACCTGAA
```


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:

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

6.2 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
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

```

6.3 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 parameters.

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:

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

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

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

Basic options
    -u, --upperbound=<int>    The upper bound value
    < input.num               Input numeric file (stdin)
    > output.num              Output numeric file (stdout)

Example: ./gto_upper_bound -u <upperbound> < input.num > output.num
```

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
```

6.4 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:

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

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

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

Basic options
    -l, --lowerbound=<int>    The lower bound value
    < input.num               Input numeric file (stdin)
    > output.num              Output numeric file (stdout)

Example: ./gto_lower_bound -l <lowerbound> < input.num > output.num
```

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_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
```

6.5 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 parameters.

Input parameters

The `gto_brute_force_string` program needs some parameters for the computation, namely the alphabet and the key size.

The attribution is given according to:

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

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

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

Basic options

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

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

Output

The output of the `gto_brute_force_string` program is a set of all possible word combinations with a defined size, using the input alphabet.

Using the 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
```

6.6 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 two streams for the computation, namely the real sequence as input. These numbers should be splitted by lines.

The attribution is given according to:

```
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 `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
0
1
```

6.7 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.

The attribution is given according to:

```
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)
> output.num       Output numeric file (stdout)
```

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
2.341  3  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
```

6.8 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 parameters.

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 (default 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))
```



```
-c, --onecolumn      Read from one column
-p, --printone       Print one column
-r, --reverse        Reverse mode
```

```
Example: ./gto_filter -w <>window size> -d <drop> -t <>window type> -c -p -r <input.num> > output.num
```

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
```

6.9 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 parameters.

Input parameters

The `gto_word_search` 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:

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

Searching for a word in a text file. It is case sensitive.
```

```

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

Basic options
    -w, --word=<str>      Word to search in the file
    < input.txt           Input text file (stdin)
    > output.txt          Output text file (stdout)

Example: ./gto_word_search -w <word> < input.txt > output.txt

```

An example of such an input file is:

```

No guts, no story. Chris Brady
My life is my message. Mahatma Gandhi
Screw it, let's 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

```

6.10 Program `gto_permute_by_blocks`

The `gto_permute_by_blocks` program permutes by block sequence, FASTA and Multi-FASTA files.

For help type:

```

./gto_ -h

```

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

Input parameters

The `gto_permute_by_blocks` 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 permutes 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
ACAAGACGGCCTCCTGCTGCTGCTCTCCGGGGCCACGGCCCTGGAGGTCCACCGCTGCCCTGCTGCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGGCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGTGCC
GCGAATCCGCGCGCCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAATTCTTCTGGAAGACCTTCTCCACCCCCCAGC
TAAACCTCACCCATGAATGCTCGCAACACGCAAGTTTAATTCGCAAGTTAGACCTGAACGGGAGGTGGCCACGCAAGTT
```

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:

```
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGGCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGTGCC
GCGAATCCGCGCGCCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAATTCTTCTGGAAGACCTTCTCCACCCCCCAGC
ACAAGACGGCCTCCTGCTGCTGCTCTCCGGGGCCACGGCCCTGGAGGTCCACCGCTGCCCTGCTGCCATTGTCCCC
TAAACCTCACCCATGAATGCTCGCAACACGCAAGTTTAATTCGCAAGTTAGACCTGAACGGGAGGTGGCCACGCAAGTT
```

6.11 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 parameters.

Input parameters

The `gto_info` program needs two streams for the computation, namely the input and output standard. The input stream is a file without 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
ACAAGACGGCCTCCTGCTGCTGCTGCTCCTCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAAGTGGACCTCCGGGCCCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGTGCC
GCGAATCGGCGCGCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACCTTCTTCTGGAAGACCTTCTCCACCCCCCAGC
TAAACCTCACCCATGAATGCTCGCAACACGCAAGTTTAATTGCAAGTTAGACCTGAACGGGAGGTGGCCACGCAAGTT
```

Output

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      : 28
Alphabet           : |srponmiedcaTRNHGCBA>=6420 \n
Symbol distribution:
| : 2      0.4415011
s : 3      0.66225166
r : 1      0.22075055
```

```

p : 1      0.22075055
o : 2      0.4415011
n : 1      0.22075055
m : 2      0.4415011
i : 1      0.22075055
e : 2      0.4415011
d : 1      0.22075055
c : 3      0.66225166
a : 2      0.4415011
T : 66     14.569536
R : 1      0.22075055
N : 1      0.22075055
H : 1      0.22075055
G : 117    25.827815
C : 131    28.918322
B : 2      0.4415011
A : 89     19.646799
> : 1      0.22075055
= : 2      0.4415011
6 : 2      0.4415011
4 : 2      0.4415011
2 : 2      0.4415011
0 : 6      1.3245033
  : 4      0.88300221
\n : 5     1.1037528

```

6.12 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:

```

Usage: ./gto_segment [options] [--] args]
       or: ./gto_segment [options]

It segments a filtered sequence.

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

Basic options
    -t, --threshold=<dbl>    The segment threshold
    < input.num              Input numeric file (stdin)

```

```
> output          Output the segment file (stdout)
```

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

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 “below the threshold.

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

```
0:10
```

6.13 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 parameters.

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),
-l <link>   Link type between maps [0;4],
-w <width>  Chromosome width,
-s <space>  Space between chromosomes,
-m <mult>   Color id multiplication factor,
-b <begin>  Color id beggining,
-c <minimum> 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 3000000 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:

```

==[ PROCESSING ]=====
Printing plot ...
Found 2 regular regions.
Found 1 inverted regions.
Done!

==[ STATISTICS ]=====
Total cpu time: 0 second(s).

```

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

6.14 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.

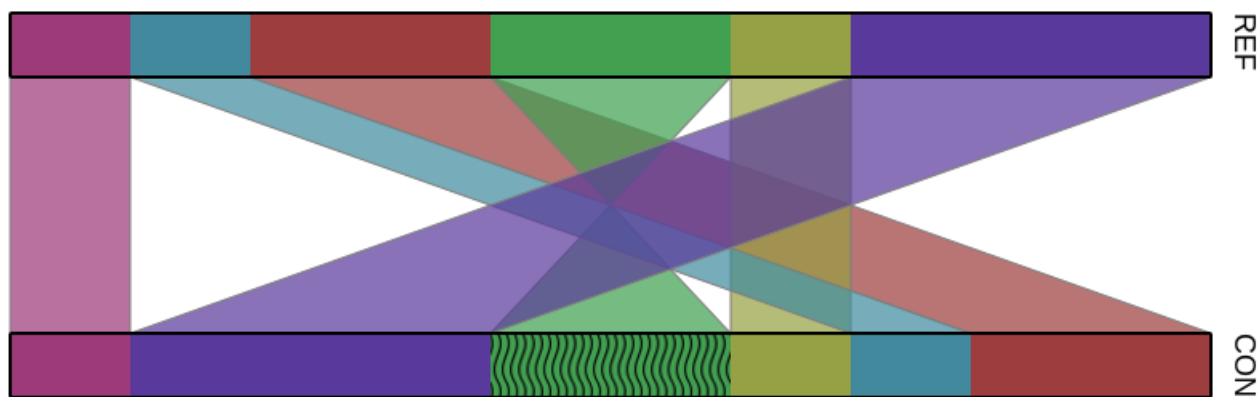


Figure 6.1: `gto_comparative_map` execution plot.

Input parameters

The `gto_max` program needs two streams for the computation, namely the input, which are two decimal files.

The attribution is given according to:

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

It computes the maximum value in each row between two files.

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

Basic options
-f, --first_file=<str>    File to compute the max
-s, --second_file=<str>   The second file to do the max computation
> output.num             Output numeric file (stdout)

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

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
```

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
```

6.15 Program `gto_min`

The `gto_min` computes the minimum value in each row between two files.

For help type:

```
./gto_min -h
```

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

Input parameters

The `gto_min` program needs two streams for the computation, namely the input, which are two decimal files.

The attribution is given according to:

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

```
It computes the minimum value in each row between two files.
```

```
-h, --help
```

```
Show this help message and exit
```

```
Basic options
-f, --first_file=<str>    File to compute the max
-s, --second_file=<str>   The second file to do the max computation
> output.num              Output numeric file (stdout)
```

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

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
```

Output

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
```

Bibliography

- [1] E. R. Mardis, “Dna sequencing technologies: 2006–2016,” *Nature protocols*, vol. 12, no. 2, p. 213, 2017.
- [2] C. Brouwer, T. D. Vu, M. Zhou, G. Cardinali, M. M. Welling, N. van de Wiele, and V. Robert, “Current opportunities and challenges of next generation sequencing (ngs) of dna; determining health and disease,” *British Biotechnology Journal*, vol. 13, no. 4, 2016.
- [3] L. Liu, Y. Li, S. Li, N. Hu, Y. He, R. Pong, D. Lin, L. Lu, and M. Law, “Comparison of next-generation sequencing systems,” *BioMed Research International*, vol. 2012, 2012.
- [4] H. Zhang, “Overview of sequence data formats,” in *Statistical Genomics*. Springer, 2016, pp. 3–17.
- [5] P. J. Cock, C. J. Fields, N. Goto, M. L. Heuer, and P. M. Rice, “The sanger fastq file format for sequences with quality scores, and the solexa/illumina fastq variants,” *Nucleic acids research*, vol. 38, no. 6, pp. 1767–1771, 2009.
- [6] A. P. Droop, “fqtools: an efficient software suite for modern fastq file manipulation,” *Bioinformatics*, vol. 32, no. 12, pp. 1883–1884, 2016.
- [7] A. Gordon, G. Hannon *et al.*, “Fastx-toolkit,” *FASTQ/A short-reads preprocessing tools (unpublished)* http://hannonlab.cshl.edu/fastx_toolkit, vol. 5, 2010.
- [8] E. Afgan, D. Baker, B. Batut, M. Van Den Beek, D. Bouvier, M. Čech, J. Chilton, D. Clements, N. Coraor, B. A. Grüning *et al.*, “The galaxy platform for accessible, reproducible and collaborative biomedical analyses: 2018 update,” *Nucleic acids research*, vol. 46, no. W1, pp. W537–W544, 2018.
- [9] M. A. DePristo, E. Banks, R. Poplin, K. V. Garimella, J. R. Maguire, C. Hartl, A. A. Philippakis, G. Del Angel, M. A. Rivas, M. Hanna *et al.*, “A framework for variation discovery and genotyping using next-generation dna sequencing data,” *Nature genetics*, vol. 43, no. 5, p. 491, 2011.
- [10] S. Kumar, G. Stecher, and K. Tamura, “Mega7: molecular evolutionary genetics analysis version 7.0 for bigger datasets,” *Molecular biology and evolution*, vol. 33, no. 7, pp. 1870–1874, 2016.
- [11] W. Shen, S. Le, Y. Li, and F. Hu, “Seqkit: a cross-platform and ultrafast toolkit for fasta/q file manipulation,” *PLoS One*, vol. 11, no. 10, p. e0163962, 2016.

- [12] J. Goecks, A. Nekrutenko, and J. Taylor, “Galaxy: a comprehensive approach for supporting accessible, reproducible, and transparent computational research in the life sciences,” *Genome biology*, vol. 11, no. 8, p. R86, 2010.
- [13] D. Blankenberg, A. Gordon, G. Von Kuster, N. Coraor, J. Taylor, A. Nekrutenko, and G. Team, “Manipulation of fastq data with galaxy,” *Bioinformatics*, vol. 26, no. 14, pp. 1783–1785, 2010.
- [14] G. A. Van der Auwera, M. O. Carneiro, C. Hartl, R. Poplin, G. Del Angel, A. Levy-Moonshine, T. Jordan, K. Shakir, D. Roazen, J. Thibault *et al.*, “From fastq data to high-confidence variant calls: the genome analysis toolkit best practices pipeline,” *Current protocols in bioinformatics*, vol. 43, no. 1, pp. 11–10, 2013.
- [15] K. Tamura, D. Peterson, N. Peterson, G. Stecher, M. Nei, and S. Kumar, “Mega5: molecular evolutionary genetics analysis using maximum likelihood, evolutionary distance, and maximum parsimony methods,” *Molecular biology and evolution*, vol. 28, no. 10, pp. 2731–2739, 2011.
- [16] M. Hosseini, D. Pratas, and A. Pinho, “A survey on data compression methods for biological sequences,” *Information*, vol. 7, no. 4, p. 56, 2016.
- [17] Y. Liu, H. Peng, L. Wong, and J. Li, “High-speed and high-ratio referential genome compression,” *Bioinformatics*, vol. 33, no. 21, pp. 3364–3372, 2017.
- [18] I. Ochoa, M. Hernaez, and T. Weissman, “idocomp: a compression scheme for assembled genomes,” *Bioinformatics*, vol. 31, no. 5, pp. 626–633, 2014.
- [19] D. Pratas, A. J. Pinho, and P. J. Ferreira, “Efficient compression of genomic sequences,” in *2016 Data Compression Conference (DCC)*. IEEE, 2016, pp. 231–240.
- [20] S. Deorowicz, A. Danek, and M. Niemiec, “Gdc 2: Compression of large collections of genomes,” *Scientific reports*, vol. 5, p. 11565, 2015.
- [21] M. Hernaez, D. Pavlichin, T. Weissman, and I. Ochoa, “Genomic data compression,” *Annual Review of Biomedical Data Science*, vol. 2, 2019.
- [22] Ö. Nalbantoglu, D. Russell, and K. Sayood, “Data compression concepts and algorithms and their applications to bioinformatics,” *Entropy*, vol. 12, no. 1, pp. 34–52, 2010.
- [23] M. Hosseini, D. Pratas, and A. J. Pinho, “Ac: A compression tool for amino acid sequences,” *Interdisciplinary Sciences: Computational Life Sciences*, pp. 1–9, 2019.
- [24] D. Pratas, M. Hosseini, and A. J. Pinho, “Compression of amino acid sequences,” in *International Conference on Practical Applications of Computational Biology & Bioinformatics*. Springer, 2018, pp. 105–113.
- [25] W. Huang, L. Li, J. R. Myers, and G. T. Marth, “Art: a next-generation sequencing read simulator,” *Bioinformatics*, vol. 28, no. 4, pp. 593–594, 2011.

- [26] A. Price and C. Gibas, “Simulome: a genome sequence and variant simulator,” *Bioinformatics*, vol. 33, no. 12, pp. 1876–1878, 2017.
- [27] G. Baruzzo, K. E. Hayer, E. J. Kim, B. Di Camillo, G. A. FitzGerald, and G. R. Grant, “Simulation-based comprehensive benchmarking of rna-seq aligners,” *Nature methods*, vol. 14, no. 2, p. 135, 2017.
- [28] M. Escalona, S. Rocha, and D. Posada, “A comparison of tools for the simulation of genomic next-generation sequencing data,” *Nature Reviews Genetics*, vol. 17, no. 8, p. 459, 2016.
- [29] D. Pratas, A. J. Pinho, and J. M. Rodrigues, “Xs: a fastq read simulator,” *BMC research notes*, vol. 7, no. 1, p. 40, 2014.