

The Genomics Toolkit

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Chapter 1

Introduction

Recent advances in DNA sequencing have revolutionized the field of genomics, making it possible for research groups to generate large amounts of sequenced data, very rapidly and at substantially lower cost [?]. The storage of genomic data is being addressed using specific file formats, such as FASTQ and FASTA. Therefore, its analysis and manipulation is crucial [?]. Many frameworks for analysis and manipulation emerged, namely GALAXY [?], GATK [?], HTSeq [?], MEGA [?], among others. Several of these frameworks require licenses, while others do not provide a low level access to the information, since they are commonly approached by scripting or programming laguages not efficient for the purpose. Moreover, several lack on variety, namely the ability to perform multiple tasks using only one toolkit.

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

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

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

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

1.1 Installation

For GTO installation, run:

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

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Chapter 2

FASTQ tools

Current available tools for FASTQ format analysis and manipulation include:

- 1. gto_fastq_to_fasta: it converts a FASTQ file format to a pseudo FASTA file.
- 2. gto_fastq_to_mfasta: it converts a FASTQ file format to a pseudo Multi-FASTA file.
- 3. gto_fastq_exclude_n: it discards the FASTQ reads with the minimum number of "N" symbols.
- 4. gto_fastq_extract_quality_scores: it extracts all the quality-scores from FASTQ reads.
- 5. gto_fastq_info: it analyses the basic information of FASTQ file format.
- 6. gto_fastq_maximum_read_size: it filters the FASTQ reads with the length higher than the value defined.
- 7. gto_fastq_minimum_quality_score: it discards reads with average quality-score below of the defined.
- 8. gto_fastq_minimum_read_size: it filters the FASTQ reads with the length smaller than the value defined.
- 9. gto_fastq_rand_extra_chars: it substitues in the FASTQ files, the DNA sequence the outside ACGT chars by random ACGT symbols.
- 10. gto_fastq_from_seq: it converts a genomic sequence to pseudo FASTQ file format.
- 11. gto_fastq_mutate: it creates a synthetic mutation of a FASTQ file given specific rates of mutations, deletions and additions.
- 12. gto_fastq_split: it splits Paired End files according to the direction of the strand ('/1' or '/2').
- 13. gto_fastq_pack: it packages each FASTQ read in a single line.
- 14. gto_fastq_unpack: it unpacks the FASTQ reads packaged using the gto_fastq_pack tool.

- 15. gto_fastq_quality_score_info: it analyses the quality-scores of a FASTQ file.
- 16. gto_fastq_quality_score_min: it analyses the minimal quality-scores of a FASTQ file.
- 17. gto_fastq_quality_score_max: it analyses the maximal quality-scores of a FASTQ file.
- 18. gto_fastq_cut: it cuts read sequences in a FASTQ file.
- 19. gto_fastq_minimum_local_quality_score_forward: it filters the reads considering the quality score average of a defined window size of bases.
- 20. gto_fastq_minimum_local_quality_score_reverse: it filters the reverse reads, considering the average window size score defined by the bases.
- 21. gto_xs: it is a skilled FASTQ read simulation tool, flexible, portable and tunable in terms of sequence complexity.

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 paramters.

Input parameters

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

The attribution is given according to:

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

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

It does NOT align the sequence.

It extracts the sequence and adds a pseudo header.

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

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

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

The output of the gto_fastq_to_fasta program a FASTA file.

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

```
> Computed with Fastq2Fasta

GGGTGATGGCCGCTGCCGATGGCGTCAAATCCCACCAAGTTACCCTTAACAACTTAAGGGTTTTCAAATAGA

GTTCAGGGATACGACGTTTGTATTTTAAGAATCTGAAGCAGAAGTCGATGATAATACGCGTCGTTTTATCAT
```

$2.2 \quad Program\ gto_fastq_to_mfasta$

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

For help type:

```
./gto_fastq_to_mfasta -h
```

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

Input parameters

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

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

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

It does NOT align the sequence.

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

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

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

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

Output

The output of the gto_fastq_to_mfasta program a Multi-FASTA file.

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

```
>SRR001666.1 071112_SLXA-EAS1_s_7:5:1:817:345 length=72
GGGTGATGGCCGCTGCCGATGGCGTCAAATCCCACCAAGTTACCCTTAACAACTTAAGGGTTTTCAAATAGA
>SRR001666.2 071112_SLXA-EAS1_s_7:5:1:801:338 length=72
GTTCAGGGATACGACGTTTGTATTTTAAGAATCTGAAGCAGAAGTCGATGATAATACGCGTCGTTTTATCAT
```

2.3 Program gto fastq exclude n

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

For help type:

```
./gto_fastq_exclude_n -h
```

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

Input parameters

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

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

It discards the FASTQ reads with the minimum number of "N" symbols.

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

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

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

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

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

Output

The output of the gto_fastq_exclude_n program is a set of all the filtered FASTQ reads, followed by the execution report. The execution report only appears in the console.

Using the input above with the max value as 5, an output example for this is the following:

2.4 Program gto fastq extract quality scores

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

```
./gto_fastq_extract_quality_scores -h
```

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

Input parameters

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

The attribution is given according to:

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

An example of such an input file is:

Output

The output of the gto_fastq_extract_quality_scores program is a set of all the quality scores from the FASTQ reads, followed by the execution report. The execution report only appears in the console. Using the input above, an output example for this is the following:

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</pre>
                         Input FASTQ file format (stdin)
   > output
                         Output read information (stdout)
Example: ./gto_fastq_info < input.fastq > output
Output example:
Total reads : value
Max read length : value
Min read length : value
Min QS value : value
Max QS value : value
QS range
              : value
```

An example of such an input file is:

Output

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

```
Total reads : 2
Max read length : 72
Min read length : 72
```

```
Min QS value : 41
Max QS value : 73
QS range : 33
```

2.6 Program gto fastq maximum read size

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

```
./gto_fastq_maximum_read_size -h
```

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

Input parameters

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

The attribution is given according to:

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

The output of the gto_fastq_maximum_read_size program is a set of all the filtered FASTQ reads, followed by the execution report. The execution report only appears in the console.

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

2.7 Program gto fastq minimum quality score

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

```
./gto_fastq_minimum_quality_score -h
```

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

Input parameters

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

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>
            : value
Total reads
Filtered reads : value
```

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

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

$2.8 \quad Program\ gto_fastq_minimum_read_size$

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

```
./gto_fastq_minimum_read_size -h
```

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

Input parameters

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

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

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

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

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

Basic options
```

Output

The output of the gto_fastq_minimum_read_size program is a set of all the filtered FASTQ reads, followed by the execution report. The execution report only appears in the console.

Using the input above with the size values as 65, an output example for this is the following:

2.9 Program gto fastq rand extra chars

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

For help type:

```
./gto_fastq_rand_extra_chars -h
```

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

Input parameters

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

The attribution is given according to:

An example of such an input file is:

Output

The output of the gto_fastq_rand_extra_chars program is a FASTQ file. Using the input above, an output example for this is the following:

2.10 Program gto fastq from seq

The gto_fastq_from_seq converts a genomic sequence to pseudo FASTQ file format. For help type:

```
./gto_fastq_from_seq -h
```

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

Input parameters

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

The attribution is given according to:

An example of such an input file is:

Output

The output of the gto_fastq_from_seq program is a pseudo FASTQ file.

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

```
@SeqToFastq4
\tt GCGAATCCGCGCGCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCACCCCCCCAGC
@SeqToFastq5
@SeqToFastq7
@SeqToFastq8
@SeqToFastq9
\tt AGAATGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCTCCTGCAAATAAAACCTCACCCATGAATGCTCACGCAAGTT
@SeqToFastq10
TAATTACAGACCTGAA
FFFFFFFFFFFFFF
```

2.11 Program gto fastq mutate

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

```
./gto_fastq_mutate -h
```

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

Input parameters

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

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

Output

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

2.12 Program gto fastq split

The gto_fastq_split splits Paired End files according to the direction of the strand ('/1' or '/2'). It writes by default singleton reads as forward stands.

For help type:

```
./gto_fastq_split -h
```

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

Input parameters

The gto_fastq_split program needs a stream for the computation, namely the input standard. The input stream is a FASTQ file.

The attribution is given according to:

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

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

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

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

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

2.13 Program gto fastq pack

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

For help type:

```
./gto_fastq_pack -h
```

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

Input parameters

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

```
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
```

Output

The output of the gto_fastq_pack program is a packaged FASTQ file.

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

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

$2.14 \quad Program\ gto_fastq_unpack$

The gto_fastq_unpack unpacks the FASTQ reads packaged using the gto_fastq_pack tool. For help type:

```
./gto_fastq_unpack -h
```

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

Input parameters

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

Output

The output of the gto_fastq_unpack program is a FASTQ file.

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

2.15 Program gto fastq quality score info

The gto_fastq_quality_score_info analyses the quality-scores of a FASTQ file. For help type:

```
./gto_fastq_quality_score_info -h
```

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

Input parameters

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

The attribution is given according to:

```
Usage: ./gto_fastq_quality_score_info [options] [[--] args]
   or: ./gto_fastq_quality_score_info [options]
It analyses the quality-scores of a FASTQ file.
   -h, --help
                         show this help message and exit
Basic options
   < input.fastq</pre>
                       Input FASTQ file format (stdin)
                         Output read information (stdout)
   > output
Optional
   -m, --max=<int>
                        The lenght of the maximum window
Example: ./gto_fastq_quality_score_info -m <max> < input.fastq > output
Output example :
Total reads : value
Max read length : value
Min read length : value
Min QS value : value
Max QS value : value
              : value
QS range
```

An example of such an input file is:

Output

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

```
Total reads : 2

Max read length : 72

Min read length : 72

Min QS value : 41

Max QS value : 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 paramters.

Input parameters

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

The attribution is given according to:

The output of the gto_fastq_quality_score_max program is a set of information related to the file read, considering the maximal quality scores.

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

2.17 Program gto fastq quality score min

The gto_fastq_quality_score_min analyses the minimal quality-scores of a FASTQ file. For help type:

```
./gto_fastq_quality_score_min -h
```

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

Input parameters

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

The attribution is given according to:

The output of the gto_fastq_quality_score_min program is a set of information related to the file read, considering the minimum quality scores.

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

2.18 Program gto fastq cut

The gto_fastq_cut cuts read sequences in a FASTQ file. It requires that the initial and end positions for the cut.

For help type:

```
./gto_fastq_cut -h
```

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

Input parameters

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

The attribution is given according to:

The output of the gto_fastq_cut program is a FASTQ file cut.

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

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

2.19 Program gto fastq minimum local quality score forward

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

For help type:

```
./gto_fastq_minimum_local_quality_score_forward -h
```

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

Input parameters

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

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

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

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

```
Basic options
   -k, --windowsize=<int>
                            The window size of bases (default 5)
   -w, --minavg=<int> The minimum average of quality score (default 25)
   -m, --minqs=<int>
                           The minimum value of the quality score (default 33)
   < input.fastq</pre>
                             Input FASTQ file format (stdin)
   > output.fastq
                             Output FASTQ file format (stdout)
Example: ./gto_fastq_minimum_local_quality_score_forward -k <windowsize> -w <minavg>
-m <minqs> < input.fastq > output.fastq
Console output example:
Minimum QS : value
<FASTQ output>
Total reads
             : value
Trimmed reads : value
```

Output

The output of the gto_fastq_minimum_local_quality_score_forward program is a FASTQ file with the reads filtered following a quality score average of a defined window of bases. The execution report only appears in the console.

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

2.20 Program gto fastq minimum local quality score reverse

The gto_fastq_minimum_local_quality_score_reverse filters the reverse reads, considering the quality score average of a defined window size of bases.

For help type:

```
./gto_fastq_minimum_local_quality_score_reverse -h
```

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

Input parameters

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

The attribution is given according to:

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

An example of such an input file is:

Output

The output of the gto_fastq_minimum_local_quality_score_reverse program is a FASTQ file with the reads filtered following a quality score average of a defined window of bases. The execution report only

appears in the console.

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

2.21 Program gto xs

The gto_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_xs program needs program needs a FASTQ file to compute.

```
[OPTION]... [FILE]
Usage: XS
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)
                          use the same header in third line of the read
 -0
 -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:
                          excludes the use of headers from output
                         excludes the use of optional headers (+) from output
 -eo
                         excludes the use of DNA bases from output
 -ed
 -edb
                          excludes \' \' when DNA bases line size is reached
                          excludes the use of quality scores from output
 -es
Stochastic options:
-s <seed>
                         generation seed
<genFile>
                         simulated output file
Common usage:
 ./XS -v -t 1 -i n=MySeq -ld 30:80 -n 20000 -qt=1 -qc 33,36,39:43 File
 ./XS -v -ls 100 -n 10000 -eh -eo -es -edb -f 0.3,0.2,0.2,0.3,0.0 -rn 50 -ri 300 -ra 3000 -rm 0.1 File
```

Output

The output of the gto_xs program is a FASTQ file

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

```
@output.fastq.598 LQGQLWH01D5WVZ length=62
TTCNTNCCAGGTAAAGAGAACATNCCGNCGCACTACTCGTAAGACTTGCTGGNCGAGAAAGG
+
)(+!*!$')($(()+'))$$()'!)!$!!$*+)+''('!)))!+!)(!+!*$!'$*)**++!
@output.fastq.1510 LQGQLWH01A7LJI length=57
CTAGACTACTCGAGCACTAGGCTCGCGTNTACCANGGGGNCTGCGNGTTGGCNCGGT
+
)+(*(+$*)+!*)!'!!(!(!!(*'$!+!(()$'!!+*+!!))!*!')***+!$+''
```

```
@output.fastq.2153 LQGQLWH01CHBQJ length=33
ACTTTTTGCTCAAGCAGGGTTGCCTAGCAANAC
+
*)++!+$''')*)**!+)$(*((*)$!'!+!!*
@output.fastq.3251 LQGQLWH01C80Y4 length=75
TCTTTCCTTCNCGNCCNAATTCCCCATAANAACTTAAAATCNCNNGCTGCGCGTGATCAACAATATTAATACTCC
+
!*''+*'!''!+!!!*!'!+(++)*(*($!!*((')$*!$(!'!!'+)$+*!$*!**!'()$!*'+'*'+!!+'(
@output.fastq.3934 LQGQLWH01AQDXM length=36
GGTAACNNGGAATTCTTCCAATTANCCNTGTCCGGC
+
$+)'!'!!)+)+!''**$$*!!')!+)!)*()!))$
```

Chapter 3

FASTA tools

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

- 1. gto_fasta_to_seq: it converts a FASTA or Multi-FASTA file format to a seq.
- 2. gto_fasta_from_seq: it converts a genomic sequence to pseudo FASTA file format.
- 3. gto_fasta_extract: it extracts sequences from a FASTA file, which the range is defined by the user in the parameters.
- 4. gto_fasta_extract_by_read: it extracts sequences from each read in a Multi-FASTA file (splited by \n), which the range is defined by the user in the parameters.
- 5. gto_fasta_info: it shows the readed information of a FASTA or Multi-FASTA file format.
- 6. gto_fasta_mutate: it reates a synthetic mutation of a fasta file given specific rates of editions, deletions and additions.
- 7. gto_fasta_rand_extra_chars: it substitues in the DNA sequence the outside ACGT chars by random ACGT symbols.
- 8. gto_fasta_extract_read_by_pattern: it extracts reads from a Multi-FASTA file format given a pattern in the header.
- 9. gto_fasta_find_n_pos: it reports the "N" regions in a sequence or FASTA (seq) file.
- 10. gto_fasta_split_reads: it splits a Multi-FASTA file to multiple FASTA files.
- 11. gto_fasta_rename_human_headers: it changes the headers of FASTA or Multi-FASTA file to simple chrX by order, where X is the number.
- 12. gto_fasta_extract_pattern_coords: it extracts the header and coordinates from a Multi-FASTA file format given a pattern/motif in the sequence.

3.1 Program gto fasta to seq

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

```
./gto_fasta_to_seq -h
```

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

Input parameters

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

The attribution is given according to:

An example of such an input file is:

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

ACAAGACGGCCTCCTGCTGCTGCTCTCCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC

GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA

GTGGTTTGAGTGGACCTCCGGGCCCCTCATAGGAAGAGCACCGGGAGGTGGCCAGGCGAGGAAGCAGGCCAGTGCC

GCGAATCCGCGCGCGCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCACCCCCCCAGC

TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA

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

ACAAGATGCCATTGTCCCCCGGGCTCCTGCTGCTGCTGCTCCTCCGGGGCCACCGCCACCGCTGCCCTTGCCCCTGGAGGGT

GGCCCCACCGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAATAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTG

GTGGTTTGAGTGGACCTCCCAGGCCAGTGCCGGGCCCCTCATAGGAGAGCTCCGGGAGGTGGCCAGGCGGCAGGAAG

GCGCACCCCCCCAGCAATCCGCGCGCCCGGGACAGAATGCCCTGCAGGAACTTCTTCTCGGAAGACCTTCTCCTCCTCCTGCAAA

TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
```

Output

The output of the gto_fasta_to_seq program is a group sequence.

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

ACAAGACGGCCTCCTGCTGCTGCTGCTCCCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGCCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGAGAAGCAGGCCAGTGCC
GCGAATCCGCGCGCGCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCCACCCCCCCAGC
TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAAACAAGATGCCATTGTCCCCCGGCCTCCTGCTG
CTGCTGCTCTCCGGGGCCACCGCCACCGCTGCCCCTGGAGGGTGGCCCCACCGGCCGAGACAGCGAGCATATGCA
GGAAGCGGCAGGAATAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGTGGTTTGAGTGGACCTCCCAGGCCAGTGCCG
GGCCCCTCATAGGAAGAGCTCCGGGAGGTGGCCAGGCGGCAGAATCCGCCCCCCAGGACATCCGCGCGGGAC
AGAATGCCCTGCAGGAACTTCTTCTTGGAAGACCTTCCTCCTCCTGCAAATAAAACCTCACCCATGAATGCTCACGCAAGTT
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 paramters.

Input parameters

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

The attribution is given according to:

An example of such an input file is:

Output

The output of the gto_fasta_from_seq program is a pseudo FASTA file.

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

3.3 Program gto fasta extract

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

For help type:

```
./gto_fasta_extract -h
```

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

Input parameters

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

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

It extracts sequences from a FASTA file.

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

```
Basic options

-i, --init=<int> The first position to start the extraction (default 0)

-e, --end=<int> The last extract position (default 100)

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

> output.seq Output sequence file (stdout)

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

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

Output

The output of the gto_fasta_extract program is a group sequence.

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

```
ACAAGACGGCCTCCTGCTGCTCCCGGGGCCACGGCCCTGGAGG
```

3.4 Program gto_fasta_extract_by_read

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

For help type:

```
./gto_fasta_extract_by_read -h
```

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

Input parameters

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

```
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
```

```
>ABO00264 |acc=ABO00264|descr=Homo sapiens mRNA
ACAAGACGGCCTCCTGCTGCTGCTCCTCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGCCCCTCATAGGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGCCAGTGCC
GCGAATCCGCGCGCGCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCACCCCCCCAGC
TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
>ABO00263 |acc=AB000263|descr=Homo sapiens mRNA
ACAAGATGCCATTGTCCCCCGGCCTCCTGCTGCTGCTCTCCCGGGGCCACCGCTGCCCTGCCCCTGGAGGGT
GGCCCCACCGGCCGAGACAGCGAGCATATGCAGGAAGCGCCAGGAAAAAGCAGCCTCCTGACTTTCCTCGCTTG
GTGGTTTGAGTGGACCTCCCAGGCCAGTGCCGGGCCCCTCATAGGAGAGGAACTTCTTCTTGGGAAGACCTTCTCCTCCTGCAAA
TAAAACCTCACCCCATGAATGCTCACGCAGGAATGCCCTGCAGGAACTTCTTCTTGGAAGACCTTCTCCTCCTGCAAA
```

Output

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

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

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

3.5 Program gto_fasta_info

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

```
./gto_fasta_info -h
```

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

Input parameters

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

The attribution is given according to:

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

An example of such an input file is:

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

ACAAGACGGCCTCCTGCTGCTGCTCTCCCGGGGCCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC

GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA

GTGGTTTGAGTGGACCTCCGGGCCCCTCATAGGAAGAGCCCTGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGTGCC

GCGAATCCGCGCGCGCGGGACAGATCTCCTGCAAAGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCACCCCCCCAGC

TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA

>ABO00263 | acc=AB000263 | descr=Homo sapiens mRNA

ACAAGATGCCATTGTCCCCCGGGCCTCCTGCTGCTGCTGCTCCCCGGGGCCACCGCTGCCCTGCCCCTGGAGGGT

GGCCCCACCGGCCGAGACAGCGAACATTGCAGGAAGCGGCAGGAATAAGGAAAAAGCAGCCTCCTGACTTTCCTCGCTTG

GTGGTTTGAGTGGACCTCCCAGGCCAGTGCCGGGCCCCTCATAGGAGAGAGCTCCGGGAGGTGGCCAGGCGCAGGAAG

GCGCACCCCCCCAGCAATCCGCGGCCCGGGACAGAATGCCCTGCAGGAACTTCTTCTCTGGAAGACCTTCTCCTCCTGCAAA

TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
```

Output

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

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

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

3.6 Program gto fasta mutate

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

For help type:

```
./gto_fasta_mutate -h
```

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

Input parameters

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

The attribution is given according to:

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

An example of such an input file is:

```
>AB000264 |acc=AB000264|descr=Homo sapiens mRNA
ACAAGACGGCCTCCTGCTGCTGCTCTCCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGCCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGTGCC
```

```
GCGAATCCGCGCGCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCACCCCCCAGC
TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
>ABOOO263 |acc=ABOOO263|descr=Homo sapiens mRNA
ACAAGATGCCATTGTCCCCCGGCCTCCTGCTGCTGCTCTCCCGGGGCCACCGCCACCGCTGCCCTTGCCCTTGAGGGT
GGCCCCACCGGCCGAGACAGCGAATATGCAGGAAGCGGCAGGAATAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTG
GTGGTTTGAGTGGACCTCCCAGGCCAGTGCCGGGCCCCTCATAGGAGAGAGCTCGGGAGGTGGCCAGGGGGAAG
GCGCACCCCCCCAGCAATCCGCGCGCCGGGACAGAATGCCCTGCAGGAACTTCTTCTTGGAAGACCTTCTCCTCCTGCAAA
```

Output

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

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

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

ACGCAACGNATTCCTGCTGATCATANTGTNCCGCNCCCNGCGACGGGGNCTCNCNNGCACACATNGTACCATTGTCCAC

NCTTNCANGTNANCGCTAGCAGGCTACNGTTTNTCCTCNCCTANNCCAANCNGGCGTNNNTACACTGGCACGTGCAGGCA

TNGGTCGGCNGGNNCCTCCGGNAACGGCACCGGAGACCGAGAGCTCGGNGGNTATACAGGTGTCANGAAACATCCCCGCGNC

GNGTGNCCNNGAANCCANAGAGTATCTCACTCACAACCCTGCGTGCACNTCTAGAGNANGACCTTACNCACCNTCCCNTT

NNGTACCACACCAATGAACGCTGCAGAAAGTCTGTTTNNAGGNGNGCA

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

ATTTGAAGGCAANCGGNCCAGNAATNCGGNGGGTGCNGCTCNTGTNGGCTACGGNCATCGCGGCCCTGCTNTANTAAGCN

TGAACCACCGNTCGNNGCACTTAGCAATNGCGNAANCCGTCGGCACGGCGGAGACNAANCCGCTANTNNTTTCCCGCTNA

ATGGNTGTACAAGACCNACTANACCANCCTCCGTCACCACACTGGAGCGCANGATGGNNCGCTGNCTAGNAGNCNNTGAG

GCGCTCCNTCCTANAAANCCGTGGNCGAGCNCCCTATGGNAGNGTGGGGGTTTTACCGGAAGACCNTCGNGCCCTATGGG

AGCAATCANAANCTAGAAAGCTTACNGATGGTGANGAANTAGACTANG
```

3.7 Program gto fasta rand extra chars

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

For help type:

```
./gto_fasta_rand_extra_chars -h
```

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

Input parameters

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

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

Output

The output of the gto_fasta_rand_extra_chars program is a FASTA or Multi-FASTA file. Using the input above, an output example for this is the following:

3.8 Program gto_fasta_extract_read_by_pattern

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

For help type:

```
./gto_fasta_extract_read_by_pattern -h
```

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

Input parameters

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

The attribution is given according to:

An example of such an input file is:

Output

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

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

3.9 Program gto fasta find n pos

The gto_fasta_find_n_pos reports the "N" regions in a sequence or FASTA (seq) file. For help type:

```
./gto_fasta_find_n_pos -h
```

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

Input parameters

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

The attribution is given according to:

An example of such an input file is:

```
>ABO00264 |acc=ABO00264|descr=Homo sapiens mRNA
NCNNNACGGCCTCCTGCTGCTGCTGCTCCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC
GNCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTNGTTTGAGTGGACCTCCGGGCCCCTCATAGGAGAGGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGTGCC
GCGAATCCGCGCGCGCGGGACAGATCTCCTGCAAAGCCCTGCAGGAACNTCTTCTGGAAGACCTTCTCCACCCCCCCAGC
TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAN
```

Output

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

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

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

3.10 Program gto fasta split reads

The gto_fasta_split_reads splits a Multi-FASTA file to multiple FASTA files. For help type:

```
./gto_fasta_split_reads -h
```

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

Input parameters

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

The attribution is given according to:

An example of such an input file is:

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

ACAAGACGGCCTCCTGCTGCTGCTCCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC

GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA

GTGGTTTGAGTGGACCTCCGGGCCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGCAGGAAGCAGCCAGTGCC

GCGAATCCGGCGCCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACCTTCTTCTGGAAGACCTTCTCCACCCCCCCAGC

TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA

>ABO00263 |acc=AB000263|descr=Homo sapiens mRNA

ACAAGATGCCATTGTCCCCCGGGCCTCCTGCTGCTGCTCCTCCGGGGCCACCGCCTGCCCTGCCCCTGGAGGGT

GGCCCCACCGGCCGAGACAGCGAAGCATATGCAGGAAAGCGGCAGGAATAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTG

GTGGTTTGAGTGGACCTCCCAGGCCAGTGCCGGGCCCCTCATAGGAGAGACCTTCTTCTGGAAGACCTTCTCCTCCTGCAAA

GCGCACCCCCCCAGCAATCCGCGCGCCGGGACAGAATGCCCTTGCAGGAACCTTCTTCTTCTGGAAGACCTTCTCCTCCTGCAAA
```

Output

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

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

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

3.11 Program gto fasta rename human headers

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

For help type:

```
./gto_fasta_rename_human_headers -h
```

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

Input parameters

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

The attribution is given according to:

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

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

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

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

Example: ./gto_fasta_rename_human_headers < input.mfasta > output.mfasta
```

An example of such an input file is:

```
> ABO00264 | acc = ABO00264 | descr = Homo sapiens mRNA
ACAAGACGGCCTCCTGCTGCTGCTCTCCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCCATTGTCCCC
GGCCCCACCTAAGGAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGCCCCTCATAGGAGAGGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGCCAGTGCC
GCGAATCCGCGCGCGCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCACCCCCCCAGC
TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
```

```
> ABOOO263 | acc = ABOOO263 | descr = Homo sapiens mRNA
ACAAGATGCCATTGTCCCCCGGCCTCCTGCTGCTGCTCCCGGGGCCACCGCCACCGCTGCCCTGCCCCTGGAGGGT
GGCCCCACCGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAATAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTG
GTGGTTTGAGTGGACCTCCCAGGCCAGTGCCGGGCCCCTCATAGGAGAGAGCTCGGGAGGTGGCCAGGCGGCAGGAAG
GCGCACCCCCCCAGCAATCCGCGCGCGGGACAGAATGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCTCCTGCAAA
```

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
ACAAGACGGCCTCCTGCTGCTGCTCCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGCCCCTCATAGGAAGAGCAGGCCGAGGAGCCAGGCGAGCAGGAAGCAGGCCAGTGCC
GCGAATCCGCGCGCGCGCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACCTTCTCTGGAAGACCTTCTCCACCCCCCCAGC
TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
>chr2
ACAAGATGCCATTGTCCCCCGGCCTCCTGCTGCTGCTCCTCCGGGGCCACGGCCACCGCTGCCCTGCCCCTGGAGGGT
GGCCCCACCGGCCGAGACATTGCAGGAAGCGGCAAGAATAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTG
GTGGTTTGAGTGGACCTCCCAGGCCAGTGCCGGGCCCCTCCATAGGAGAGAAGCTCCTGGGAAGCGCCACGCCACGCCAGCCGCCAGGCAGAAG
GCGCACCCCCCCAGCAATCCGCGCGCCCGGGACAGAATGCCCTGCAGGAACTTCTTCTTCGGAAGACCTTCTCCTCCTCCTCCAAA
```

3.12 Program gto fasta extract pattern coords

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

For help type:

```
./gto_fasta_extract_pattern_coords -h
```

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

Input parameters

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

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

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

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

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

Output

The output of the gto_fasta_extract_pattern_coords program is a Multi-FASTA file. Using the input above, with the pattern ACA, an output example for this is the following:

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

Chapter 4

Genomic sequence tools

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

- 1. gto_genomic_gen_random_dna: it generates a synthetic DNA.
- 2. gto_genomic_rand_seq_extra_chars: it substitues in the DNA sequence the outside ACGT chars by random ACGT symbols.
- 3. gto_genomic_dna_mutate: it creates a synthetic mutation of a sequence file given specific rates of mutations, deletions and additions.
- 4. gto_genomic_extract: it extracts sequences from a sequence file, which the range is defined by the user in the parameters.
- 5. gto_genomic_period: it calculates the best order depth of a sequence, using FCMs.
- 6. gto_genomic_count_bases: it counts the number of bases in sequence, FASTA or FASTQ files.

4.1 Program gto genomic gen random dna

The gto_genomic_gen_random_dna generates a synthetic DNA.

For help type:

```
./gto_genomic_gen_random_dna -h
```

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

Input parameters

The gto_genomic_gen_random_dna program needs one stream for the computation, namely the output standard.

```
Usage: ./gto_genomic_gen_random_dna [options] [[--] args]
   or: ./gto_genomic_gen_random_dna [options]
It generates a synthetic DNA.
   -h, --help
                              show this help message and exit
Basic options
   > output.seq
                              Output synthetic DNA sequence (stdout)
Optional
   -s, --seed=<int>
                              Starting point to the random generator (Default 0)
   -n, --nSymbols=<int>
                            Number of symbols generated (Default 100)
   -f, --frequency=<str>
                             The frequency of each base. It should be represented
                              in the following format: <fa,fc,fg,ft>.
Example: ./gto_genomic_gen_random_dna -s <seed> -n <nsybomls> -f <fa,fc,fg,ft> > output.seq
```

Output

The output of the gto_genomic_gen_random_dna program is a sequence group file whith the synthetic DNA.

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

4.2 Program gto genomic rand seq extra chars

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

For help type:

```
./gto_genomic_rand_seq_extra_chars -h
```

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

Input parameters

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

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

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

It works in sequence file formats

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

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

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

Output

The output of the gto_genomic_rand_seq_extra_chars program is a sequence file. Using the input above, an output example for this is the following:

4.3 Program gto genomic dna mutate

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

```
./gto_genomic_dna_mutate -h
```

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

Input parameters

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

The attribution is given according to:

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

Output

The output of the gto_genomic_dna_mutate program is a sequence file whith the synthetic mutation of input file.

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

4.4 Program gto genomic extract

The gto_genomic_extract extracts sequences from a sequence file, which the range is defined by the user in the parameters.

For help type:

```
./gto_genomic_extract -h
```

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

Input parameters

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

The attribution is given according to:

An example of such an input file is:

Output

The output of the gto_genomic_extract program is a group sequence.

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

TCTTTACTCGCGCGTTGGAGAAATACAATAGTGCGGCTCTGTCTCCTTAT

4.5 Program gto_genomic_period

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

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

For help type:

```
./gto_genomic_period -h
```

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

Input parameters

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

The attribution is given according to:

An example of such an input file is:

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
```

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

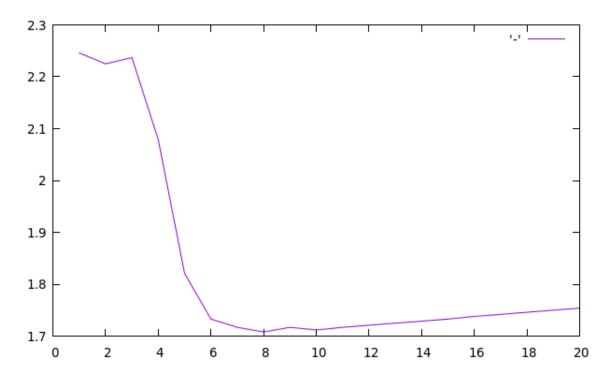


Figure 4.1: gto_genomic_period execution plot.

4.6 Program gto_genomic_count_bases

The gto_genomic_count_bases counts the number of bases in sequence, FASTA or FASTQ files. For help type:

```
./gto_genomic_count_bases -h
```

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

Input parameters

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

```
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
```

Output

The output of the gto_genomic_count_bases program is report which describes the number of each base in the file, and the file type.

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

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

Chapter 5

Amino acid sequence tools

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_ac: it is a new lossless compressor to compress efficiently amino acid sequences (proteins).

5.1 Program gto amino acid to group

The gto_amino_acid_to_group converts an amino acid sequence to a group sequence. For help type:

```
./gto_amino_acid_to_group -h
```

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

Input parameters

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

```
Example: ./gto_amino_acid_to_group < input.prot > output.group
Table:
Prot
       Group
        Р
        P Amino acids with electric charged side chains: POSITIVE
K
D
        N
Ε
       N Amino acids with electric charged side chains: NEGATIVE
S
        U
Т
N
        U Amino acids with electric UNCHARGED side chains
        IJ
Q
С
        S
U
        S
G
       S Special cases
Р
        S
Α
        Н
V
        Η
Ι
        Н
L
        Н
М
        H Amino acids with hydrophobic side chains
F
        Н
Y
        Н
        Н
        * Others
Х
        X Unknown
```

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

```
IPFLLKKQFALADKLVLSKLRQLLGGRIKMMPCGGAKLEPAIGLFFHAIGINIKLGYGMTETTATVSCWHDFQFNPNSIG
TLMPKAEVKIGENNEILVRGGMVMKGYYKKPEETAQAFTEDGFLKTGDAGEFDEQGNLFITDRIKELMKTSNGKYIAPQY
IESKIGKDKFIEQIAIIADAKKYVSALIVPCFDSLEEYAKQLNIKYHDRLELLKNSDILKMFE
```

Output

The output of the gto_amino_acid_to_group program is a group sequence. Using the input above, an output example for this is the following:

HSHHHPPUHHHHNPHHHUPHPUHHSSPHPHHSSSSHPHNSHHSHHHPHHSHUHPHSHSHUNUUHUHUSHPNHUHUSUUHS UHHSPHNHPHSNUUNHHHPSSHHHPSHHPPSNNUHUHHUNNSHHPUSNHSNHNNUSUHHHUNPHPNHHPUUUSPHHHSUH HNUPHSPNPHHNUHHHHHNHPPHHUHHHHSSHNUHNNHHPUHUHPHPNPHNHHPUUNHHPHHN

5.2 Program gto amino acid to pseudo dna

The gto_amino_acid_to_pseudo_dna converts an amino acid (protein) sequence to a pseudo DNA sequence.

For help type:

```
./gto_amino_acid_to_pseudo_dna -h
```

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

Input parameters

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

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

```
X GGG
```

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

IPFLLKKQFALADKLVLSKLRQLLGGRIKMMPCGGAKLEPAIGLFFHAIGINIKLGYGMTETTATVSCWHDFQFNPNSIG TLMPKAEVKIGENNEILVRGGMVMKGYYKKPEETAQAFTEDGFLKTGDAGEFDEQGNLFITDRIKELMKTSNGKYIAPQY IESKIGKDKFIEQIAIIADAKKYVSALIVPCFDSLEEYAKQLNIKYHDRLELLKNSDILKMFE

Output

The output of the gto_amino_acid_to_pseudo_dna program is a DNA sequence. Using the input above, an output example for this is the following:

5.3 Program gto ac

The gto_ac 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_ac -h
```

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

Input parameters

The gto_ac program needs a file with amino acid sequences to compress.

```
Usage: AC [OPTION]... -r [FILE] [FILE]:[...]
Compression of amino acid sequences.

Non-mandatory arguments:

-h give this help,
```

```
show AC compression levels,
 - s
  – v
                       verbose mode (more information),
 – V
                       display version number,
 -f
                       force overwrite of output,
 -1 <level>
                       level of compression [1;7] (lazy -tm setup),
                       threshold frequency to discard from alphabet,
 -t <threshold>
                       it creates a file with the extension ".iae"
 - e
                       with the respective information content.
 -rm <c>:<d>:<g>/<m>::<e>:<a> reference model (-rm <math>5:90:0.9/1:50:0.8),
 -tm <c>:<d>:<g>/<m>:<e>:<a> target model (-tm 1:1:0.8/0:0:0),
 target and reference templates use <c> for
                       context-order size, \langle d \rangle for alpha (1/\langle d \rangle), \langle g \rangle
                       for gamma (decayment forgetting factor) [0;1),
                       <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)</a>
                        [0;1) (tolerant model),
 -r <FILE>
                       reference file ("-rm" are loaded here),
Mandatory arguments:
 <FILE>:<...>:<...>
                      file to compress (last argument). For more
                       files use splitting ":" characters.
Example:
             ./AC -v -tm 1:1:0.8/0:0:0 -tm 5:20:0.9/3:20:0.9 seq.txt
  [Decompress] ./AD -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_ac -v -1 2 HI
./gto_ad -v HI.co
cmp HI HI.de
```

Chapter 6

General purpose tools

- 1. gto_char_to_line: it splits a sequence into lines, creating an output sequence which has a char for each line.
- 2. gto_reverse: it reverses the order of a sequence.
- 3. gto_new_line_on_new_x: it splits different rows with a new empty row.
- 4. gto_upper_bound: it sets an upper bound in a file with a value per line.
- 5. gto_lower_bound: it sets an lower bound in a file with a value per line.
- 6. gto_brute_force_string: it generates all combinations, line by line, for an inputted alphabet and specific size.
- 7. gto_real_to_binary_with_threshold: it converts a sequence of real numbers into a binary sequence, given a threshold.
- 8. gto_sum: it adds decimal values in file, line by line, splitted by spaces or tabs.
- 9. gto_filter: it filters numerical sequences.
- 10. gto_word_search: it search for a word in a file.
- 11. gto_permute_by_blocks: it permutates by block sequence, FASTA and Multi-FASTA files.
- 12. gto_info: it gives the basic properties of the file, namely size, cardinality, distribution percentage of the symbols, among others.
- 13. gto_segment: it segments a filtered sequence.
- 14. gto_comparative_map: it creates a visualization for comparative maps.
- 15. gto_max: it computes the maximum value in each row between two files.
- 16. gto_min: it computes the minimum value in each row between two files.

```
17. gto_geco: it ...18. gto_gede: it ...
```

6.1 Program gto char to line

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

For help type:

```
./gto_char_to_line -h
```

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

Input parameters

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

The attribution is given according to:

An example of such an input file is:

ACAAGACGGCCTCCTGCTGCTGCTGCTCCTCCGGGGCCACGGCCCTGAAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGCCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGAGAAGCAGGCCAGTGCC
GCGAATCCGCGCGCCGGGACAGAATCTCCTGCAAAGCCCTCCAGGAACCTTCTTCTGGAAGACCTTCTCCCACCCCCCAGC
TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAAACAAGATGCCATTGTCCCCCGGCCTCCTGCTG
CTGCTGCTCTCCGGGGCCACCGCCACCGCTGCCCCTGGAGGGTGGCCCCACCGGCCGAGACAGCGAGCATATGCA
GGAAGCGGCAGGAATAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGTGGTTTGAGTGGACCTCCCAGGCCAGTGCCG
GGCCCCTCATAGGAAGAGCTCCGGGAGGTGGCCAGGCGGCAGAATCCGCCCCCCAGGACATCCGCGCGGGAC
AGAATGCCCTGCAGGAACTTCTTCTGGAAGACCTTCCTCCTCCTCCTGCAAATAAAACCTCACCCATGAATGCTCACGCAAGTT
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:

```
С
Α
G
Α
С
G
G
С
С
Т
С
С
T
G
С
Т
G
С
Т
```

6.2 Program gto_reverse

The gto_reverse reverses the order of a sequence file.

For help type:

```
./gto_reverse -h
```

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

Input parameters

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

Output

The output of the gto_reverse program is a group sequence.

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

6.3 Program gto new line on new x

The gto_new_line_on_new_x splits different rows with a new empty row.

For help type:

```
./gto_new_line_on_new_x -h
```

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

Input parameters

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

```
      1
      2
      2

      1
      2
      2

      4
      4
      1

      10
      12
      2

      15
      15
      1

      45
      47
      3

      45
      47
      3

      45
      47
      3

      55
      55
      1
```

Output

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

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

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

6.4 Program gto upper bound

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

For help type:

```
./gto_upper_bound -h
```

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

Input parameters

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

The attribution is given according to:

An example of such an input file is:

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

Output

The output of the gto_upper_bound program is a set of numbers truncated at the a defined upper bound. Using the input above, an output example for this is the following:

```
Using upper bound: 4
0.123000
3.432000
2.341000
```

```
1.323000
4.000000
4.000000
0.242000
0.654000
4.000000
```

6.5 Program gto lower bound

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

For help type:

```
./gto_lower_bound -h
```

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

Input parameters

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

The attribution is given according to:

An example of such an input file is:

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

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

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

6.6 Program gto_brute_force_string

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

For help type:

```
./gto_brute_force_string -h
```

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

Input parameters

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

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

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

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

6.7 Program gto real to binary with threshold

The gto_real_to_binary_with_threshold converts a sequence of real numbers into a binary sequence, given a threshold. The numbers below to the threshold will be 0.

For help type:

```
./gto_real_to_binary_with_threshold -h
```

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

Input parameters

The gto_real_to_binary_with_threshold program needs program needs two streams for the computation, namely the real sequence as input. These numbers should be splitted by lines.

The attribution is given according to:

An example of such an input file is:

```
12.25

1.2

5.44

5.51

7.97

2.34

8.123
```

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
1
0
```

6.8 Program gto sum

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

For help type:

```
./gto_sum -h
```

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

Input parameters

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

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

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

An example of such an input file is:

```
      0.123
      5
      5

      3.432
      2

      1.323
      7.538
      5

      4.122
      0.242

      0.654
      5.633
      10
```

Output

The output of the gto_sum program is a sum of the elements in the input file.

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

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

6.9 Program gto_filter

The gto_filter filters numerical sequences using a low-pass filter.

For help type:

```
./gto_filter -h
```

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

Input parameters

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

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

An example of such an input file is:

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

Output

The output of the gto_filter program is a numeric file, identical of the input.

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

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

6.10 Program gto word search

The gto_word_search search for a word in a file. It is case sensitive.

For help type:

```
./gto_word_search -h
```

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

Input parameters

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

The attribution is given according to:

An example of such an input file is:

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

Output

The output of the gto_word_search program is a text file with the matching paragraphs and the location of the word found.

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

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

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

6.11 Program gto permute by blocks

The gto_permute_by_blocks permutates by block sequence, FASTA and Multi-FASTA files. For help type:

```
./gto_ -h
```

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

Input parameters

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

The attribution is given according to:

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

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

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

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

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

An example of such an input file is:

```
>ABO00264 |acc=AB000264|descr=Homo sapiens mRNA
ACAAGACGGCCTCCTGCTGCTGCTCTCCCGGGGCCACGGCCCTGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGGCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGTGCC
GCGAATCCGCGCGCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCACCCCCCCAGC
TAAAACCTCACCCATGAATGCTCGCAACACGCAAGTTTAATTCGCAAGTTAGACCTGAACGGGAGGTGGCCACGCAAGTT
```

Output

The output of the gto_permute_by_blocks program is a sequence, FASTA or Multi-FASTA file permuted following some parameters.

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

6.12 Program gto info

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

For help type:

```
./gto_info -h
```

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

Input parameters

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

The attribution is given according to:

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

An example of such an input file is:

```
>ABO00264 |acc=AB000264|descr=Homo sapiens mRNA
ACAAGACGGCCTCCTGCTGCTGCTCTCCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGCCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGCAGGAAGCAGCCAGTGCC
```

The output of the gto_info program is a set of information related to the file read.

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

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

6.13 Program gto segment

The gto_segment segments a filtered sequence.

For help type:

```
./gto_segment -h
```

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

Input parameters

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

The attribution is given according to:

An example of such an input file is:

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

Output

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

```
0:10
```

6.14 Program gto_comparative_map

The gto_comparative_map creates a visualization for comparative maps.

For help type:

```
./gto_comparative_map -h
```

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

Input parameters

The gto_comparative_map program needs an input file with the plot positions, respecting a defined structure.

The attribution is given according to:

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

An example of such an input file is:

```
#SCF 5000000 5000000 aaa 1 1000000 1 1000000 bbbb 3000000 4000000 3000000 4000000 bbb 1500000 2000000 1500000 2000000 cccc 1500000 2000000 1500000 2000000 aaa 2000000 3000000 2000000 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:

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

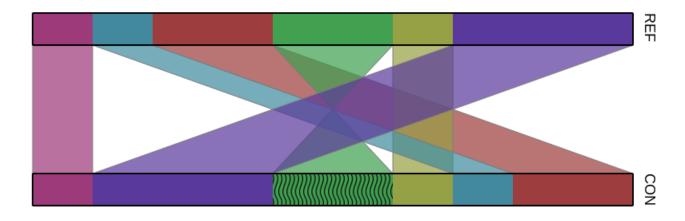


Figure 6.1: gto_comparative_map execution plot.

6.15 Program gto max

The gto_max computes the maximum value in each row between two files.

For help type:

```
./gto_max -h
```

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

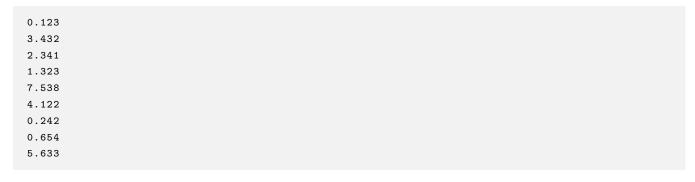
Input parameters

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

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

An example of such an input files are:

File 1:



File 2:

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

Output

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

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

```
2.123000

5.312000

2.355000

1.323000

7.538000

4.122000

0.532000

7.324000

5.633000
```

6.16 Program gto_min

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

For help type:

```
./gto_min -h
```

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

Input parameters

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

The attribution is given according to:

An example of such an input files are:

File 1:

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

File 2:

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

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

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

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

6.17 Program gto geco

The gto_geco .

For help type:

```
./gto_geco -h
```

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

Input parameters

The gto_geco program needs program needs two streams for the computation, namely the input and output standard. ...

The attribution is given according to:

```
TODO
```

An example of such an input file is:

```
TO DO
```

Output

The output of the gto_geco program is ...

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

```
TO DO
```

6.18 Program gto_gede

The ${\tt gto_gede}$.

For help type:

```
./gto_gede -h
```

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

Input parameters

The gto_gede program needs program needs two streams for the computation, namely the input and output standard. ...

The attribution is given according to:

TODO

An example of such an input file is:

TO DO

Output

The output of the gto_gede program is ...

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

TO DO