

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 [1]. The storage of genomic data is being addressed using specific file formats, such as FASTQ and FASTA. Therefore, its analysis and manipulation is crucial [2]. Many frameworks for analysis and manipulation emerged, namely GALAXY [3], GATK [4], HTSeq [5], MEGA [6], among others. Several of these frameworks require licenses, while others do not provide a low level access to the information, since they are commonly approached by scripting or programming laguages not efficient for the purpose. Moreover, several lack on variety, namely the ability to perform multiple tasks using only one toolkit.

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

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

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

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

1.1 Installation

For GTO installation, run:

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

1.2 License

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

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Chapter 2

Amino acid sequence tools

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

- 1. gto_amino_acid_to_group: it converts an amino acid sequence to a group sequence.
- 2. gto_amino_acid_to_pseudo_dna: it converts an amino acid (protein) sequence to a pseudo DNA sequence.

2.1 Program gto amino acid to group

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

```
./gto_amino_acid_to_group -h
```

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

Input parameters

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

```
R.
        Р
           Amino acids with electric charged side chains: POSITIVE
        Р
D
        N
           Amino acids with electric charged side chains: NEGATIVE
S
        U
T
        U
N
        U
           Amino acids with electric UNCHARGED side chains
        U
С
        S
U
        S
G
        S
           Special cases
Р
        S
        Н
        Н
Ι
        Н
        Н
M
        H Amino acids with hydrophobic side chains
F
        Η
Y
        Н
        Н
           Others
           Unknown
```

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

```
IPFLLKKQFALADKLVLSKLRQLLGGRIKMMPCGGAKLEPAIGLFFHAIGINIKLGYGMTETTATVSCWHDFQFNPNSIG
TLMPKAEVKIGENNEILVRGGMVMKGYYKKPEETAQAFTEDGFLKTGDAGEFDEQGNLFITDRIKELMKTSNGKYIAPQY
IESKIGKDKFIEQIAIIADAKKYVSALIVPCFDSLEEYAKQLNIKYHDRLELLKNSDILKMFE
```

Output

The output of the gto_amino_acid_to_group program is a group sequence.

An example, for the input, is:

HSHHHPPUHHHHNPHHHUPHPUHHSSPHPHHSSSSHPHNSHHSHHHPHHSHUHPHSHSHUNUUHUHUSHPNHUHUSUUHS UHHSPHNHPHSNUUNHHHPSSHHHPSHHPPSNNUHUHHUNNSHHPUSNHSNHNNUSUHHHUNPHPNHHPUUUSPHHHSUH HNUPHSPNPHHNUHHHHHNHPPHHUHHHHSSHNUHNNHHPUHUHPHPNPHNHHPUUNHHPHHN

2.2 Program gto amino acid to pseudo dna

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

For help type:

```
./gto_amino_acid_to_pseudo_dna -h
```

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

Input parameters

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

```
Usage: ./gto_amino_acid_to_pseudo_dna [options] [[--] args]
   or: ./gto_amino_acid_to_pseudo_dna [options]
It converts a protein sequence to a pseudo DNA sequence.
    -h, --help
                         show this help message and exit
Basic options
    < input.prot
                         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
         {\tt DNA}
Α
         GCA
С
         TGC
D
         GAC
Ε
         GAG
F
         TTT
G
         {\tt GGC}
Н
         CAT
Ι
         ATC
K
         A\,A\,A
L
         CTG
         ATG
N
         {\tt AAC}
Р
         CCG
Q
         CAG
         CGT
R
S
         TCT
T
         {\tt ACG}
V
         {\tt GTA}
W
         TGG
Y
         TAC
         {\tt TAG}
Х
         GGG
```

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

IPFLLKKQFALADKLVLSKLRQLLGGRIKMMPCGGAKLEPAIGLFFHAIGINIKLGYGMTETTATVSCWHDFQFNPNSIG TLMPKAEVKIGENNEILVRGGMVMKGYYKKPEETAQAFTEDGFLKTGDAGEFDEQGNLFITDRIKELMKTSNGKYIAPQY IESKIGKDKFIEQIAIIADAKKYVSALIVPCFDSLEEYAKQLNIKYHDRLELLKNSDILKMFE

Output

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

Chapter 3

FASTQ tools

Current available tools for FASTQ format analysis and manipulation include:

- 1. gto_fastq_to_fasta: it converts a FASTQ file format to a pseudo FASTA file.
- 2. gto_fastq_to_mfasta: it converts a FASTQ file format to a pseudo Multi-FASTA file.
- 3. gto_fastq_exclude_n: it discards the FASTQ reads with the minimum number of "N" symbols.
- 4. gto_fastq_extract_quality_scores: it extracts all the quality-scores from FASTQ reads.
- 5. gto_fastq_info: it analyses the basic informations 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.

3.1 Program gto fastq to fasta

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

For help type:

```
./gto_fastq_to_fasta -h
```

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

Input parameters

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

The attribution is given according to:

An example on such an input file is:

Output

The output of the gto_fastq_to_fasta program a FASTA file.

An example, for the input, is:

```
GGGTGATGGCCGCTGCCGATGGCGTCAAATCCCACCAAGTTACCCTTAACAACTTAAGGGTTTTCAAATAGA
GTTCAGGGATACGACGTTTGTATTTTAAGAATCTGAAGCAGAAGTCGATGATAATACGCGTCGTTTTATCAT
```

3.2 Program gto fastq to mfasta

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

For help type:

```
./gto_fastq_to_mfasta -h
```

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

Input parameters

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

The attribution is given according to:

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

It converts a FASTQ file format to a pseudo Multi-FASTA file.
It does NOT align the sequence.

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

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

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

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

An example on such an input file is:

Output

The output of the ${\tt gto_fastq_to_mfasta}$ program a Multi-FASTA file.

An example, for the input, is:

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

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

GTTCAGGGATACGACGTTTGTATTTTAAGAATCTGAAGCAGAAGTCGATGATAATACGCGTCGTTTTATCAT
```

3.3 Program gto fastq exclude n

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

For help type:

```
./gto_fastq_exclude_n -h
```

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

Input parameters

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

The attribution is given according to:

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

An example on such an input file is:

Output

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

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

3.4 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:

An example on 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.

An example, for the input, is:

3.5 Program gto fastq info

The gto_fastq_info analyses the basic informations 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.

```
Usage: ./gto_fastq_info [options] [[--] args]
   or: ./gto_fastq_info [options]
It analyses the basic informations 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
```

Output

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

```
Total reads : 2

Max read length : 72

Min read length : 72

Min QS value : 41

Max QS value : 73

QS range : 33
```

3.6 Program gto fastq maximum read size

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

```
./gto_fastq_maximum_read_size -h
```

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

Input parameters

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

```
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
```

Output

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

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

3.7 Program gto_fastq_minimum_quality_score

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

```
./gto_fastq_minimum_quality_score -h
```

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

Input parameters

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

```
Usage: ./gto_fastq_minimum_quality_score [options] [[--] args]
   or: ./gto_fastq_minimum_quality_score [options]
It discards reads with average quality-score below value.
                          show this help message and exit
    -h, --help
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 read information (stdout)
    > output
Example: ./gto_fastq_minimum_quality_score < input.fastq > output
Output example :
<FASTQ non-filtered reads>
Total reads : value
Filtered reads : value
```

Output

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

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

$3.8 \quad {\bf Program~gto_fastq_minimum_read_size}$

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.

The attribution is given according to:

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

An example on such an input file is:

Output

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

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

3.9 Program gto fastq rand extra chars

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

For help type:

```
./gto_fastq_rand_extra_chars -h
```

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

Input parameters

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

The attribution is given according to:

An example on such an input file is:

Output

The output of the gto_fastq_rand_extra_chars program is a FASTQ file. An example, for the input, is:

3.10 Program gto fastq from seq

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

```
./gto_fastq_from_seq -h
```

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

Input parameters

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

The attribution is given according to:

An example on such an input file is:

Output

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

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

SeqToFastq1 CAAGACGGCCTCCTGCTGCTGCTGCTCTCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTC FFFFFFFFFFFFFFFFFFFFFFFFFFF	FFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFF	
CAAGACGGCCTCCTGCTGCTGCTGCTCCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTC FFFFFFFFFFFFFFFFFFFFFFFFFFF	FFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFF	
FFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFF	FFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFF	
SeqToFastq2 GCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCA FFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFF	CAGGAAGCGGCAGGAA CFFFFFFFFFFFFFFFFFFFFF	
SeqToFastq2 GCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCA FFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFF	CAGGAAGCGGCAGGAA CFFFFFFFFFFFFFFFFFFFFF	
GCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCA FFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFF	FFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFF	
FFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFF	FFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFF	
FFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFF	AAGCAGGCCAGTGCC FFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFF	
SeqToFastq3 TGGTTTGAGTGGACCTCCGGGCCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGA FFFFFFFFFFFFFFFFFFFFFFFF	AAGCAGGCCAGTGCC FFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFF	
TGGTTTGAGTGGACCTCCGGGCCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAGGTTGGCTAGGAGAGATTTGAGGAGAGGTTGAGGAGAGAGA	CTCCACCCCCCAGC CFFFFFFFFFFFFFFFFFFFFFFFFFF	
FFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFF	CTCCACCCCCCAGC CFFFFFFFFFFFFFFFFFFFFFFFFFF	
FFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFF	CTCCACCCCCCAGC FFFFFFFFFFFFFFFFFFFFFFFFFFF	
SeqToFastq4 CGAATCCGCGCGCGCGGACAGAATCTCCTGCAAAGCCCTGCAGGAACTTCTTCTGGAAGACCTTC FFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFF	CTCCACCCCCCAGC FFFFFFFFFFFFFFFFFFFFFFFFFFF	
CGAATCCGCGCGCCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACTTCTTCTGGAAGACCTTC FFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFF	CCCGGCCTCCTGCTG	
FFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFF	CCCGGCCTCCTGCTG	
FFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFF	CCCGGCCTCCTGCTG	
SeqToFastq5 AAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAAACAAGATGCCATTGTCC FFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFF	CCCGGCCTCCTGCTG	
AAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAAACAAGATGCCATTGTCCC		
FFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFF		
	FFFFFFFFFFFF	
	FFFFFFFFFFFF	
SoctoFoctof		
Seq10rastq0		
TGCTGCTCTCCGGGGCCACGGCCACCGCTGCCCTGCAGGGTGGCCCCACCGGCCGAGA	CAGCGAGCATATGCA	
FFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFF	FFFFFFFFFFFF	
SeqToFastq7		
GAAGCGGCAGGAATAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGTGGTTTGAGTGGACCT	CCCAGGCCAGTGCCG	
FFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFF	FFFFFFFFFFFF	
SeqToFastq8		
GCCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGGGGGGAAGGCGCACCCCCCAGCAA	TCCGCGCGCGGGAC	
	FFFFFFFFFFFF	
SeqToFastq9		
GAATGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCTCCTGCAAATAAAACCTCACCCATGAA	ATGCTCACGCAAGTT	
FFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFFF		

```
@SeqToFastq10
TAATTACAGACCTGAA
+
FFFFFFFFFFFFFFF
```

3.11 Program gto fastq mutate

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

```
./gto_fastq_mutate -h
```

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

Input parameters

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

The attribution is given according to:

```
Usage: ./gto_fastq_mutate [options] [[--] args]
  or: ./gto_fastq_mutate [options]
Creates a synthetic mutation of a FASTQ file given specific rates of mutations,
deletions and additions
   -h, --help
                              show this help message and exit
Basic options
                              Input FASTQ file format (stdin)
   < input.fasta
   > 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
<insertion rate> -a < input.fastq > output.fastq
```

An example on such an input file is:

Output

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

Chapter 4

FASTA tools

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

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

4.1 Program gto_fasta_to_seq

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

```
./gto_fasta_to_seq -h
```

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

Input parameters

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

The attribution is given according to:

An example on such an input file is:

Output

The output of the gto_fasta_to_seq program is a group sequence.

An example, for the input, is:

GGCCCCTCATAGGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGGCGCACCCCCCAGCAATCCGCGCGCCGGGACAGAATGCCCTGCAGGAACTTCTTCTTGGAAGACCTTCTCCTCCTGCAAATAAAACCTCACCCATGAATGCTCACGCAAGTT

4.2 Program gto_fasta_from_seq

The gto_fasta_from_seq converts a genomic sequence to pseudo FASTA file format. For help type:

```
./gto_fasta_from_seq -h
```

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

Input parameters

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

The attribution is given according to:

An example on such an input file is:

Output

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

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

4.3 Program gto fasta extract

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

For help type:

```
./gto_fasta_extract -h
```

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

Input parameters

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

```
>ABO00264 |acc=ABO00264|descr=Homo sapiens mRNA
ACAAGACGGCCTCCTGCTGCTGCTCCCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGCCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGTGCC
GCGAATCCGCGCGCGCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCACCCCCCAGC
TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
```

Output

The output of the gto_fasta_extract program is a group sequence.

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

ACAAGACGGCCTCCTGCTGCTGCTCCCGGGGCCACGGCCCTGGAGG

4.4 Program gto_fasta_extract_by_read

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

For help type:

```
./gto_fasta_extract_by_read -h
```

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

Input parameters

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

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

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

ACAAGACGCCTCCTGCTGCTGCTCCTCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGCCCCTCATAGGAAGAGCAGCCTGGGAGGTGGCCAGGCGGCAGGAAGCAGCCAGTGCC
GCGAATCCGCGCGCCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCACCCCCCCAGC
TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
>AB000263 | acc=AB000263 | descr=Homo sapiens mRNA
ACAAGATGCCATTGTCCCCCGGCCTCCTGCTGCTGCTCTCCCGGGGCCACCGCTGCCCTGCCCCTGGAGGGT
GGCCCCACCGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAATAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTG
GTGGTTTGAGTGGACCTCCCAGGCCAGTGCCGGGCCCCTCATAGGAGAAGCTCCGGGAGGTGGCCAGGCGGCAGGAAG
GCGCACCCCCCCAGCAATCCGCGCGCCCGGGACAGAATGCCCTGCAGGAACTTCTTCTCTGGAAGACCTTCTCCTCCTGCAAA
TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
```

Output

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

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

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

4.5 Program gto_fasta_info

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

```
./gto_fasta_info -h
```

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

Input parameters

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

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

It shows read information of a FASTA or Multi-FASTA file format.
```

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

```
>ABOOO264 |acc=ABOOO264|descr=Homo sapiens mRNA
ACAAGACGGCCTCCTGCTGCTGCTCTCCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGCCCCTCATAGGAGAGAGCACCCGGGAGCCAGGCCAGGAAGCAGGCCAGTGCC
GCGAATCCGCGCGCGCGGGACAGATCTCCTGCAAAGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCACCCCCCCAGC
TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
>ABOOO263 |acc=ABOOO263|descr=Homo sapiens mRNA
ACAAGATGCCATTGTCCCCCGGGCCTCCTGCTGCTGCTGCTCCTCCGGGGCCACCGCCACCGCTGCCCTTGCCCCTGGAGGGT
GGCCCCACCGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAATAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTG
GTGGTTTGAGTGGACCTCCCAGGCCAGTGCCGGGCCCCTCTAAGGAGAGCTCCGGGAGGTGGCCAGGCGCAGGAAG
GCGCACCCCCCCAGCAATCCGCGGGCCCGGGACAGAATGCCCTGCAGGAACTTCTTCTCGCAAA
TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCCTGAA
```

Output

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

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

4.6 Program gto fasta mutate

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

```
./gto_fasta_mutate -h
```

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

Input parameters

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

The attribution is given according to:

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

An example on such an input file is:

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

ACAAGACGGCCTCCTGCTGCTGCTCTCCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC

GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA

GTGGTTTGAGTGGACCTCCGGGCCCCTCATAGGAGAGAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGTGCC

GCGAATCCGCGCGCGCGGGACAGATCTCCTGCAAAGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCACCCCCCCAGC

TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA

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

ACAAGATGCCATTGTCCCCCGGGCCTCCTGCTGCTGCTGCTCCCGGGGCCACCGCTGCCCTGCCCCTGGAGGGT

GGCCCCACCGGCCGAGACAGCCATATGCAGGAAGCGGCAGGAATAAGGAAAAAGCAGCCTCCTGACTTTCCTCGCTTG

GTGGTTTGAGTGGACCTCCCAGGCCAGTGCCGGGCCCCTCATAGGAGAGAGCTCCGGGAGGTGGCCAGGCGCAGGAAG

GCGCACCCCCCCAGGAATCCGCGGGCCGGGACAGAATGCCCTGCAGGAACTTCTTCTCGGAAGACCTTCTCCTCCTCCTGCAAA

TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
```

Output

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

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

```
> ABOOO264 | acc = ABOOO264 | descr = Homo sapiens mrna
ACGCAACGNATTCCTGCTGATCATANTGTNCCGCNCCCCNGCGACGGGGNCTCNCNNGCACACATNGTACCATTGTCCAC
NCTTNCANGTNANCGCTAGCAGGCTACNGTTTNTCCTCNCCTANNCCAANCNGGCGTNNNTACACTGGCACGTGCAGGCA
TNGGTCGGCNGGNNCCTCCGGNAACGGCACCGGAGACGACGTCGGNGGNTATACAGGTGTCANGAAACATCCCCGCGNC
GNGTGNCCNNGAANCCANAGAGTATCTCACTCACAACCCTGCGTGCACNTCTAGAGNANGACCTTACNCACCNTCCCNTT
NNGTACCACACCAATGAACGCTGCAGAAAGTCTGTTTNNAGGNGNGCA
> ABOOO263 | acc = ABOOO263 | descr = Homo sapiens mrna
ATTTGAAGGCAANCGGNCCAGNAATNCGGNGGGTGCNGCTCNTGTNGGCTACGGNCATCGCGGCCCTGCTNTANTAAGCN
TGAACCACCGNTCGNNGCACTTAGCAATNGCGNAANCCGTCGGCACGGCGGAGACNAANCCGCTANTNNTTTCCCGCTNA
ATGGNTGTACAAGACCNACTANACCANCCTCCGTCACCACACTGGGGCGCANGATGGNNCGCTGNCTAGNAGNCNNTGAG
GCGCTCCNTCCTANAAANCCGTGGNCGAGCNCCCTATGGNAGNGTGGGGGTTTTACCGGAAGACCNTCGNGCCCTATGGG
AGCAATCANAANCTAGAAAGCTTACNGATGGTGANGAANTAGACTANG
```

4.7 Program gto fasta rand extra chars

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

For help type:

```
./gto_fasta_rand_extra_chars -h
```

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

Input parameters

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

Output

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

4.8 Program gto_fasta_extract_read_by_pattern

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

For help type:

```
./gto_fasta_extract_read_by_pattern -h
```

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

Input parameters

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

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

ACAAGACGGCCTCCTGCTGCTGCTCTCCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCCATTGTCCCC

GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA

GTGGTTTGAGTGGACCTCCGGGCCCCTCATAGGAGAGAGCTCGGGAGGTGGCCAGGCGAGGCAGGAAGCAGGCCAGTGCC

GCGAATCCGCGCGCGCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCACCCCCCCAGC

TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA

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

ACAAGATGCCATTGTCCCCCGGCCTCCTGCTGCTGCTCCTCCGGGGCCACCGCTGCCCTGCCCTTGCAGGGT

GGCCCCACCGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAATAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTG

GTGGTTTGAGTGGACCTCCCAGGCCAGTGCCGGGCCCCTCATAGGAAGAGCTCGGGAGGTGGCCAGGCGGCAGGAAG

TAAAACCTCACCCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
```

Output

The output of the gto_fasta_extract_read_by_pattern program is a Multi-FASTA file. An example, using the pattern "264", for the provided input, is:

```
>ABO00264 | acc=AB000264 | descr=Homo sapiens mRNA
ACAAGACGCCTCCTGCTGCTGCTCCTCCGGGGCCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGCCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGCCCCCCCAGC
GCGAATCCGCGCGCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCCACCCCCCCAGC
TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
```

4.9 Program gto_fasta_find_n_pos

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

```
./gto_fasta_find_n_pos -h
```

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

Input parameters

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

The attribution is given according to:

An example on such an input file is:

```
>ABOOO264 |acc=ABOOO264|descr=Homo sapiens mRNA
NCNNNACGGCCTCCTGCTGCTGCTCCTCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCCGTTGCCATTGTCCCC
GNCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTNGTTTGAGTGGACCTCCGGGCCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGTGCC
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.

An example, for the input, is:

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

4.10 Program gto_fasta_split_reads

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

```
./gto_fasta_split_reads -h
```

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

Input parameters

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

The attribution is given according to:

An example on such an input file is:

```
>ABOOO264 |acc=ABOOO264|descr=Homo sapiens mRNA
ACAAGACGGCCTCCTGCTGCTGCTCTCCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGCCCCTCATAGGAAGAGCACCTCGGGAGGTGGCCAGGCGAGGAAGCAGGCCAGTGCC
GCGAATCCGCGCGCGCGGGACAGATCTCCTGCAAAGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCACCCCCCCAGC
TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
>ABOOO263 |acc=ABOOO263|descr=Homo sapiens mRNA
ACAAGATGCCATTGTCCCCCGGGCTCCTGCTGCTGCTGCTCCTCCGGGGCCACCGCCACCGCTGCCCTTGCCCCTGGAGGGT
GGCCCCACCGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAATAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTG
GTGGTTTGAGTGGACCTCCCAGGCCAGTGCCGGGCCCCTCTAAGGAGAGCTCCGGGAGGTGGCCAGGCGGCAGGAAG
GCGCACCCCCCCAGCAATCCGCGCGCCGGGACAGAATGCCCTGCAGGAACTTCTTCTCGGAAGACCTTCTCCTCCTCCCAAA
TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
```

Output

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

An example, for the input, is:

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

4.11 Program gto fasta rename human headers

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

For help type:

```
./gto_fasta_rename_human_headers -h
```

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

Input parameters

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

The attribution is given according to:

An example on such an input file is:

```
> ABOOO264 | acc = ABOOO264 | descr = Homo sapiens mRNA
ACAAGACGGCCTCCTGCTGCTGCTGCTCCCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGCCCCTCATAGGAGAGGGAAGCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGTGCC
GCGAATCCGCGCGCGCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACTTCTTCTTGGAAGACCTTCTCCACCCCCCCAGC
TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA
> ABOOO263 | acc = ABOOO263 | descr = Homo sapiens mRNA
ACAAGATGCCATTGTCCCCCGGGCCTCCTGCTGCTGCTCCTCCCGGGGCCACCGCTGCCCTGCCCTTGCAGGGT
GGCCCCACCGGCCGAGACAGCATATGCAGGAAGCGGCAGGAATAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTG
GTGGTTTGAGTGGACCTCCCAGGCCAGTGCCCGGGCCCCTCCATAGGAGAGACCTCCTTCTCGGAAGACCTTCTCCTCCTCCAAA
TAAAACCTCACCCCTGAATGCTCACGCAAGTTTAATTACAGACCTGAA
```

Output

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

>chr1

ACAAGACGCCTCCTGCTGCTGCTGCTCTCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCTGCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGCCCCTCATAGGAGAGGAAGCTCGGGAGGTGGCCAGGCGAGGCAGGAAGCAGGCCAGTGCC
GCGAATCCGCGCGCGCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCACCCCCCCAGC
TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA

>chr2

ACAAGATGCCATTGTCCCCCGGCCTCCTGCTGCTGCTGCTCTCCCGGGGCCACGGCCACCGCTGCCCTGCCCTGGAGGGT
GGCCCCACCGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAATAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTG
GTGGTTTGAGTGGACCTCCCAGGCCAGTGCCGGGCCCCTCATAGGAGAGGCTCCTGGGAGGTGGCCAGGCGGCAGGAAG
GCGCACCCCCCCAGCAATCCGCGCGCCGGGACAGATGCCCTGCAGGAACTTCTTCTTGGAAGACCTTCTCCTCCTGCAAA
TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAA

Chapter 5

Genomic sequence tools

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

- 1. gto_genomic_gen_random_dna: it generates a synthetic DNA.
- 2. gto_genomic_rand_seq_extra_chars: it substitues in the DNA sequence the outside ACGT chars by random ACGT symbols.

5.1 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.

The attribution is given according to:

```
in the following format: <fa,fc,fg,ft>.
Example: ./gto_genomic_gen_random_dna > output.seq
```

Output

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

Using the seed value as 1 and the number of symbols as 400, an example of an execution, is:

5.2 Program gto genomic rand seq extra chars

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

For help type:

```
./gto_genomic_rand_seq_extra_chars -h
```

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

Input parameters

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

The attribution is given according to:

An example on such an input file is:

Output

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

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.

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:

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

It splits a sequence into lines, creating an output sequence which has a char for each line.
  -h, --help show this help message and exit

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

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

An example on such an input file is:

ACAAGACGGCCTCCTGCTGCTGCTGCTCCCCGGGGCCACGGCCCTGGAGGGTCCACCGCTGCCCTGCCCATTGTCCCC
GGCCCCACCTAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGGCCGAGACAGCGAGCATATGCAGGAAGCGGCAGGAA
GTGGTTTGAGTGGACCTCCGGGCCCCTCATAGGAGGAAGCCTCGGGAGGTGGCCAGGCGGCAGGAAGCAGGCCAGTGCC
GCGAATCCGCGCCGGGACAGAATCTCCTGCAAAGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCACCCCCCCAGC
TAAAACCTCACCCATGAATGCTCACGCAAGTTTAATTACAGACCTGAAACAAGATGCCATTGTCCCCCGGCCTCCTGCTG
CTGCTGCTCTCCGGGGCCACGGCCACCGCTGCCCTGGCGCTGGTGGTTTGAGTGGACCTCCCAGGCCAGTGCCG
GGAAGCGGCAGGAATAAGGAAAAGCAGCCTCCTGACTTTCCTCGCTTGGTGTTTTGAGTGGACCTCCCAGGCCAGTGCCG
GGCCCCTCATAGGAAGAAGCTCCTGGGAGGTGGCCAGGAAGGCGCACCCCCCCAGCAATCCCGCGCCGGGAC
AGAATGCCCTGCAGGAACTTCTTCTGGAAGACCTTCTCCTCCTCCTGCAAATAAAACCTCACCCATGAATGCTCACGCAAGTT
TAATTACAGACCTGAA

Output

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

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

6.2 Program gto_reverse

The gto_reverse reverses the order of a sequence file.

For help type:

```
./gto_reverse -h
```

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

Input parameters

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

The attribution is given according to:

An example on such an input file is:

Output

The output of the gto_reverse program is a group sequence.

An example, for the input, is:

6.3 Program gto new line on new x

The gto_new_line_on_new_x splits different rows with a new empty row.

For help type:

```
./gto_new_line_on_new_x -h
```

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

Input parameters

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

The attribution is given according to:

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

It splits different rows with a new empty row.
  -h, --help show this help message and exit

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

Example: ./gto_new_line_on_new_x < input > output
```

An example on such an input file is:

```
      1
      2
      2

      1
      2
      2

      4
      4
      1

      10
      12
      2

      15
      15
      1

      45
      47
      3

      45
      47
      3

      45
      47
      3

      55
      55
      1
```

Output

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

An example, for the input, is:

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

Bibliography

- [1] E. R. Mardis, "DNA sequencing technologies: 2006–2016," Nature protocols, vol. 12, no. 2, p. 213, 2017.
- [2] H. Buermans and J. Den Dunnen, "Next generation sequencing technology: advances and applications," Biochimica et Biophysica Acta (BBA)-Molecular Basis of Disease, vol. 1842, no. 10, pp. 1932–1941, 2014.
- [3] B. Giardine, C. Riemer, R. C. Hardison, R. Burhans, L. Elnitski, P. Shah, Y. Zhang, D. Blankenberg, I. Albert, J. Taylor et al., "Galaxy: a platform for interactive large-scale genome analysis," Genome research, vol. 15, no. 10, pp. 1451–1455, 2005.
- [4] M. A. DePristo, E. Banks, R. Poplin, K. V. Garimella, J. R. Maguire, C. Hartl, A. A. Philippakis, G. Del Angel, M. A. Rivas, M. Hanna et al., "A framework for variation discovery and genotyping using next-generation dna sequencing data," *Nature genetics*, vol. 43, no. 5, pp. 491–498, 2011.
- [5] S. Anders, P. T. Pyl, and W. Huber, "Htseq-a python framework to work with high-throughput sequencing data," *Bioinformatics*, p. btu638, 2014.
- [6] S. Kumar, G. Stecher, and K. Tamura, "Mega7: Molecular evolutionary genetics analysis version 7.0 for bigger datasets," *Molecular Biology and Evolution*, p. msw054, 2016.