GOOSE

A toolkit for DNA sequence analysis and manipulation

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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. Its storage have been made using specific file formats, such as FASTQ and FASTA. Therefore, its analysis and manipulation is crucial [?]. Several frameworks for analysis and manipulation emerged, namely GALAXY [?], GATK [?], HTSeq [?], MEGA [?], among others. In the majority, these frameworks require licenses and do not provide a low level access to the information, since they are commonly approached by scripting or interfaces.

We present GOOSE, a (free) novel toolkit for analyzing and manipulating SEQ ("ACGTN"), FASTA or FASTQ formats, with many complementary tools, for Linux-based systems, built for fast processing. GOOSE supports pipes for easy integration. It includes tools for information display, randomizing, edition, conversion, extraction, searching, calculation and visualization. GOOSE is prepared to deal high very large datasets, typically GB and TB sizes. For this dimension of data fast access and flexibility is crucial, which are some of the qualities of GOOSE.

The toolkit is a command line version, using the prefix "goose-" followed by the suffix with the respective name of the program. GOOSE is implemented in C language and it is freely available, under GPLv3, at `https://pratas.github.io/goose`.

FASTQ tools

- goose-fastq2fasta

- goose-fastq2mfasta

- goose-fastqclustreads

- goose-fastqexcluden

- goose-FastqExcludeN

- goose-fastqmaximumreadsize

- goose-FastqMaximumReadSize

- goose-FastqMinimumLocalQualityScoreForward

- goose-FastqMinimumLocalQualityScoreReverse

- goose-fastqminimumlocalqualityscoresingleend

- goose-fastqminimumqualityscore

- goose-FastqMinimumQualityScore

- goose-fastqminimumreadsize

- goose-FastqMinimumReadSize

- goose-fastqpack

- goose-fastqsimulation

- goose-fastqtrimm

- goose-FastqTrimm

- goose-fastqunpack

- goose-mutatefastq

- goose-randfastqextrachars

- goose-seq2fastq

FASTA tools

SEQ tools

General purpose tools

Note 11: Software availability & characteristics

All source code for the FALCON is available at:

- **https://github.com/pratas/falcon**.

FALCON includes the following programs (See Note 2 for a common pipeline using the programs):

1. **FALCON** - infers metagenomic composition;

2. **FALCON-FILTER** - filters local similarity of inferred sequences;

3. **FALCON-EYE** - displays the output from FALCON and FALCON-FILTER in a compact map;

FALCON program

The FALCON program enables to identify and quantify the similarity between present-days reference sequences and the FASTQ reads from ancient genomes. In the following subsections, we explain the input and output paramters.

Input parameters

The FALCON program needs two files for the computation. The FILE1, with the reads provided from the NGS platform, and the FILE2, with the multi-FASTA cointaining the sequences of the genomes and respective headers, are mandatory arguments. The rest of the parameters are non mandatory, namely:

```
Usage: FALCON [OPTION]... [FILE1] [FILE2]
A compression-based method to infer metagenomic sample composition.

Non-mandatory arguments:

  -h                      give this help,
  -F                      force mode (overwrites top file),
  -V                      display version number,
  -v                      verbose mode (more information),
  -Z                      database local similarity,
  -s                      show compression levels,
```

```
  -l <level>                compression level [1;44],
  -p <sample>               subsampling (default: 1),
  -t <top>                  top of similarity (default: 20),
  -n <nThreads>             number of threads (default: 2),
  -x <FILE>                 similarity top filename,
  -y <FILE>                 local similarities filename,


 Mandatory arguments:


  [FILE1]                   metagenomic filename (FASTA or FASTQ),
  [FILE2]                   database filename (FASTA or Multi-FASTA).
```

There are two hidden parameters. One for setting the cache size:

```
  -c <cache>                maximum collisions for hash cache. Memory
                            values are higly dependent of the parameter
                            specification.
```

This is a parameter that affects the precision and computational resources needed for the computation. Lower values use less memory. For a complete description see [**?**, **?**]. The previous parameter is only needed when the context of the Markov model is higher than 13. For setting specifically the models through the command line use the following explanation (more information can be found at [**?**, **?**]):

Output

The output of the FALCON program are two files. One file contains the top-n values with the highest similarity relatively to the reads. The following output shows an example of a top-5:

```
1    9472     92.597   548558394    Human endogenous retrovirus K113
2    4668621  42.184   1069460419   Escherichia coli strain 210221272
3    4558287  42.016   1008930592   Shigella sp. PAMC 28760
4    4574246  36.661   844762407    Shigella boydii strain ATCC 9210
5    4878853  36.531   992379426    Shigella sonnei strain FDAARGOS 90
```

These column values stand, respectively, for the ranking of higher similarity, the sequence size, the percentage of similarity, the global identifyer (GI) and the name of the FASTA read (genome name).

The other file contains the relative local similarities and it is only available when FALCON runs with the "-Z" flag. This file is the input of the FALCON-FILTER program that we describe next. This file is packed in a compact format for not increasing the substantially the storage.

FALCON-FILTER program

The FALCON-FILTER program enables to identify and quantify where the similarity between present-days reference sequences and the FASTQ reads from ancient genomes, below a certain threshold, occurs. In the following subsections, we explain the input and output paramters.

Input parameters

The FALCON-FILTER program needs only one file for the computation. The file is provided from the output of the FALCON program (when the flag "-Z" is set on the FALCON program). The non-mandatory arguments are mostly filters and the threshold.

```
Usage: FALCON-FILTER [OPTION]... [FILE]
Filter and segment FALCON output.

Non-mandatory arguments:

  -h                       give this help,
  -F                       force mode (overwrites top file),
  -V                       display version number,
  -v                       verbose mode (more information),
  -s  <size>               filter window size,
  -w  <type>               filter window type,
  -x  <sampling>           filter window sampling,
  -sl <lower>              similarity lower bound,
  -su <upper>              similarity upper bound,
  -dl <lower>              size lower bound,
  -du <upper>              size upper bound,
  -t  <threshold>          threshold [0;2.0],
  -o  <FILE>               output filename,

Mandatory arguments:

  [FILE]                   profile filename (from FALCON).
```

Output

The output of the file contains the coordinates of the relative similar regions with the respective self-similarity classification.