Grundlagen der Bioinformatik
Assignment 3: Alignment

SS 2016

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Vorstellung Lösungen Übung 2

Overview – Assignment 2 (20P)

(1) Analyse transcription factor *GATA2* (4P)

Vorstellung durch eine Gruppe

(2) Substring search (10P)

Vorstellung durch zwei Gruppen

(3) Properties of Boyer Moore Algorithm (6P)

Je eine Person für (a) und (b)

Assignment 3
Alignment

Overview – Assignment 3 (20P)

(1) Local Alignment (10P)

(2) Global Alignment (5P)

(3) Aligning real sequences (5P)

(1) Local Alignment (10P)

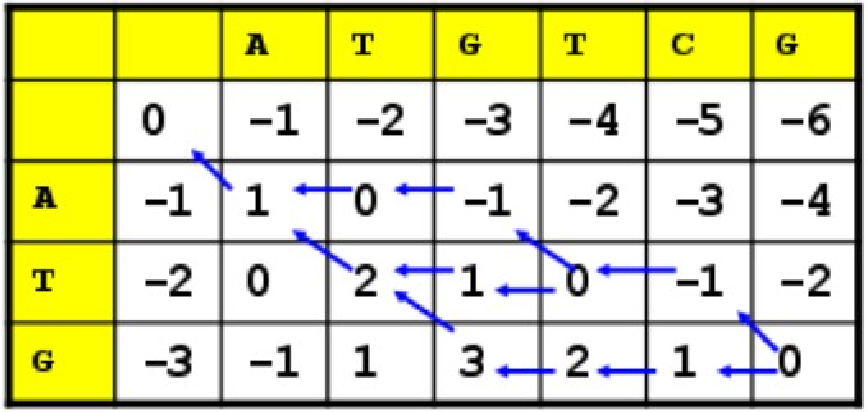
- Write a program to compute the local similarity of two DNA sequences using Smith Waterman
  - Sequences must be read from a FASTA file (pair.fasta) (1P)
  - Use replacement costs provided in matrix file (matrix.txt) (2P)
    - Deletion/Insertion cost is 8
  - Print length of best local alignment, score, number of matches, replacements and deletions+insertions (3P)
  - Print alignment (4P)
  - Programmaufruf:

```
AAATT_GCC
|. || |.|
AC_TTTGGC
```

```
java -jar Assignment3_GrXY.jar pairs.fasta matrix.txt
```

(1) Global/Local Alignment (10P)

Global alignment



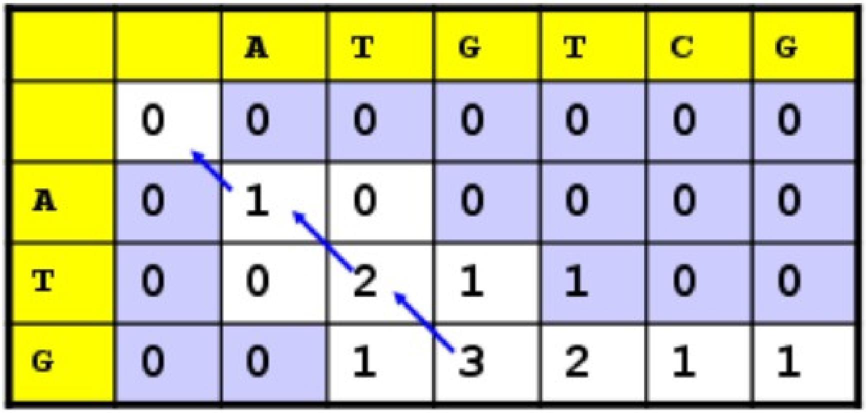
		A	T	G	T	C	G
	0	−1	−2	−3	−4	−5	−6
A	−1	1	0	−1	−2	−3	−4
T	−2	0	2	1	0	−1	−2
G	−3	−1	1	3	2	1	0

ATGTCG
ATG___

ATGTCG
AT___G

ATGTCG
A__T_G

Local alignment



		A	T	G	T	C	G
	0	0	0	0	0	0	0
A	0	1	0	0	0	0	0
T	0	0	2	1	1	0	0
G	0	0	1	3	2	1	1

ATG
ATG

(1) Local Alignment (10P)

pair.fasta:

```
>seq1
CCCAGCAGCAGAAGTTATCACTGGCTATCAACGATTGAACTCCCAATGTGGCGAGCAACGGA
CGGCACAGCAGGCAGCCTTACTCCATGTTGTTCGACAATACTCAGTTCTACAGTCCAG
>seq2
CTGAGCACCGCTTTTGCACTACAAGGATTCGAACCCCATTGTGCGAACAACGGACGCACAGC
ATTACACCTGTTTGCCGATATTCACCCTGATGTGGG
```

matrix.txt:

```
#
# DNA scoring matrix
#
# Lowest score = -4, Highest
score = 5
#
    A   T   G   C
A   5  -3  -4  -4
T  -3   5  -4  -4
G  -4  -4   5  -2
C  -4  -4  -2   5
```

deletion/insertion cost is 8
→ score = -8

(2) Global Alignment (5P)

Derive a formula which calculates how many optimal alignments exist between a string of length *n* and a string of length *m*, if both strings are defined over the same one-element alphabet.

Explain how you derived this formula.

(3) Aligning real Sequences (5P)

- *KRAS* is a *RAS* family member and an important oncogene. Mutation status is used to estimate drug response for colorectal cancer
- Download the DNA sequences for human (NM_004985.3) and mouse (NM_021284.6): www.ncbi.nlm.nih.gov/nuccore
- Calculate local alignment score and alignment using your program (1P)
- Calculate local alignment score using EMBOSS (2P)
- Are the results the same? Discuss if not. Explain the required steps to get the same results (2P)

(3) Aligning real Sequences (5P)

EMBOSS

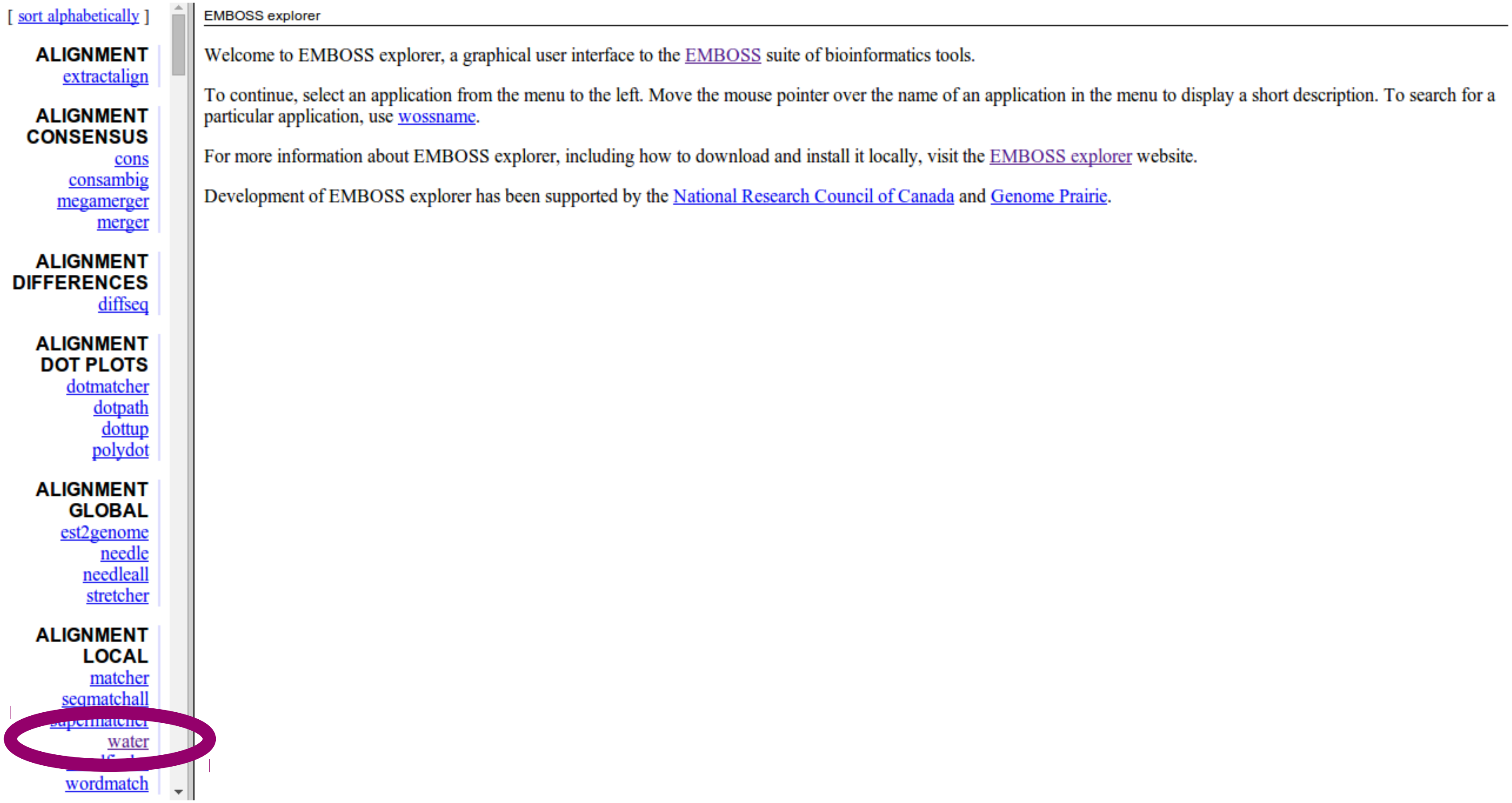
- **E**uropean **M**olecular **B**iology **O**pen **S**oftware **S**uite

- Framework for many tasks
  - Sequence retrieval
  - Alignment
  - Folding
  - Motif finding
  - ...

- Can be used online or locally
  - http://emboss.sourceforge.net/
    http://emboss.bioinformatics.nl/

(3) Aligning real Sequences (5P)

EMBOSS http://emboss.bioinformatics.nl/



Welcome to EMBOSS explorer, a graphical user interface to the EMBOSS suite of bioinformatics tools.

To continue, select an application from the menu to the left. Move the mouse pointer over the name of an application in the menu to display a short description. To search for a particular application, use wossname.

For more information about EMBOSS explorer, including how to download and install it locally, visit the EMBOSS explorer website.

Development of EMBOSS explorer has been supported by the National Research Council of Canada and Genome Prairie.

Abgabe

- Abgabe bis MIwttwoch den 01.06.2016 um 23:59 Uhr

- Abgabe per Email an: yvonne.lichtblau@informatik.hu-berlin.de (gerne auch Fragen zur Übung per Email)
    - PDF mit
        - Task 1: Output eures Programms
        - Task 2: Antwort
        - Task 3: Output Eures Programms, Emboss Score, Antwort zu Task 3
    - Code als Jar Datei wie beschrieben (Übung 1)
    - Sourcecode

- .jar auf gruenau2 testen!

- **Tipp: Score für Task 1 ist zwischen 150 and 170!**