

Algorithms in Genome Research
Winter 2020/2021

Exercises

Number 2, Discussion: 2020 November 20

1. For “reads” 1-8 from the following list, build the overlap graphs (a) with a minimum overlap of 2, and (b) with a minimum overlap of 3.

1 ATCCA
2 AGAGC
3 AAGAT
4 GAGCA
5 CCATA
6 GCAAG
7 AGATC
8 TAGAG
9 AGAGC
10 GAGCA

2. Find a shortest common superstring (error-free) for “reads” 1-10 of Exercise 1 above. Is the coverage uniform? If not, find a layout with a more uniform coverage.
3. Discuss the main experimental problems that make sequence assembly difficult in practice.
4. What are mate pairs and paired-end reads? What can be done with long reads that can not be done with paired-end reads?
5. What are the main differences between “traditional” (*de-novo*) genome assembly and comparative assembly?
6. What are the major steps in the comparative assembly strategy?
7. Let the following DNA sequence be a “reference genome”:

AATGAGGTCATCCTTGCTGGACTCTAGCAC

The following three sets of “reads” (a), (b), (c) originate from three “target” genomes that are closely related to the reference.

Consider the following conditions:

- There are no sequencing errors.
- Each target genome differs from the reference by a single structural variation (rearrangement).
- A read may come from any of the two complementary DNA strands.

Reconstruct the three target genomes by mapping the reads to the reference and identify the rearrangements.

(a)

- 1 AATGAGGTCA
- 2 AGGTCATCGAC
- 3 AGTCGATGAC
- 4 CATCGACTCT
- 5 CTAGAGTCGAT
- 6 GTGCTAGAGT

(b)

- 1 ACTCTAGCAC
- 2 AGTCCTGTACAG
- 3 CCTTGCTGTA
- 4 GCTGTACAGGAC
- 5 GGTCATCCTT
- 6 TGACCTCATT

(c)

- 1 AATGACAAGG
- 2 ACCCTGGACTCT
- 3 GGATGACCCTG
- 4 GTCATCCTTG
- 5 GTGCTAGAGT
- 6 TCCAGGGTCA