UNIVERSITY
OF SKÖVDE

School of Bioscience

WRITTEN EXAMINATION

Course: Bioinformatics – Concepts and Methods

Examination: Module 5

Course code: BI760A

Credits for written examination: 1.5

Date: 28 October 2025

Examination time: 2 hours

Examination responsible: Zelmina Lubovac

Aid at the exam/appendices: None

Other

Instructions	☐	Take a new sheet of paper for each teacher.
	☒	Take a new sheet of paper when starting a new question.
	☒	Write only on one side of the paper.
	☒	Write your name and personal ID No. on all pages you hand in.
	☒	Use page numbering.
	☒	Don´t use a red pen.
	☒	Mark answered questions with a cross on the cover sheet.

Grade points: 0-15 = F; 16-18 = E; 19-21 = D; 22-24 = C; 25-27 = B; 28-30 = A

Examination results should be made public within 18 working days

Good luck!

Total number of pages: 6

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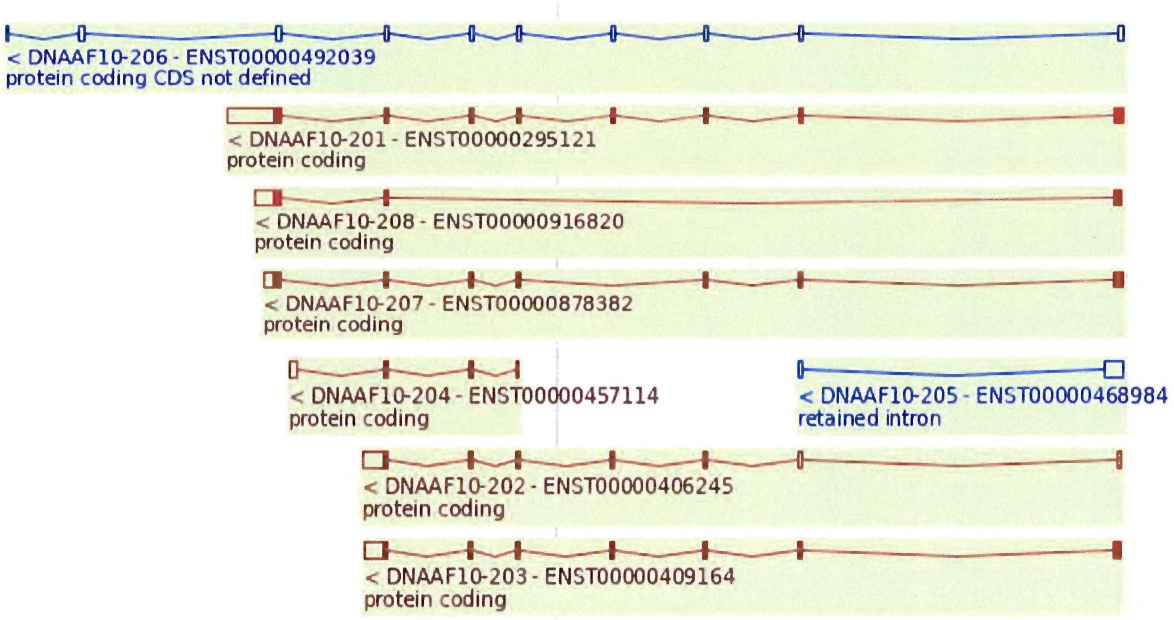
Question 1 (6p)

The figure below shows a region overview from Ensembl. It was created by searching for the DNAAF10 gene (dynein axonemal assembly factor 10) in the human genome.

1a) How many transcripts (splice variants) does DNAAF10 gene have, and how many of those transcripts are protein-coding? (**2p**)

1b) Describe what we can learn about the intron-exon structure of a transcript from the visualization in the region overview. You should pick one of the protein-coding DNAAF10 transcripts as an example to comment on in your description. Make sure to include the accession number of the chosen transcript in your answer. (**2p**)

1c) Now compare the transcript from question 1b with another DNAAF10 transcript from the region overview. Explain how they differ from each other. For the two chosen transcripts, you should mention, for example, which one has the largest number of exons and which one will produce the longest amino acid sequence. (**2p**)



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Question 2 (6p)

For each of the following claims about Ensembl, state if the claim is true or false. You do not need to give any motivations in your answer, just writing (for each claim) "true" or "false" is sufficient.

2a) Ensembl contains data only for the human genome. **(1p)**

2b) The Ensembl Variant Effect Predictor (VEP) helps in determining the potential effects of genetic variants on gene function, including for species beyond humans. **(1p)**

2c) You can find data about single-nucleotide polymorphisms (SNPs) in Ensembl. **(1p)**

2d) Ensembl's genome browser allows for accessing genomic data, while BioMart is used only for displaying results from queries. **(1p)**

2e) The Ensembl genome browser allows users to view the chromosomal location of genes. **(1p)**

2f) Ensembl does not support data export. **(1p)**

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Question 3 (6p)

For each of the following claims related to gene prediction, state if the claim is true or false. You do not need to give any motivations in your answer, just writing (for each claim) "true" or "false" is sufficient.

3a) During ORF Finder-based ORF detection, any intermediate start codons are disregarded when searching for the largest possible open reading frames. (**1p**)

3b) The structure of tRNA is highly variable, with no strict requirements for stem or loop length. (**1p**)

3c) *Ab initio* gene prediction relies only on the genome sequence without any additional information. (**1p**)

3d) In ORF prediction by ORF Finder tool, intermediate start codons are ignored while searching for the longest open reading frames. (**1p**)

3e) Homology-based gene prediction involves identifying genes in a newly sequenced genome by comparing it to related genomes. (**1p**)

3f) ORF Finder does not distinguish between actual genes and potential coding sequences, requiring further validation. (**1p**)

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Question 4 (6p)

The tRNAscan algorithm predicts tRNA genes using a rule-based approach.

4a) What features of the tRNA structure does tRNAscan roughly use as rules to predict tRNA genes, and in which way are these rules applied? (You don't need to give the exact rules, just the general idea) (**3p**)

4b) Gene prediction in prokaryote genomes is generally easier than gene prediction in eukaryote genomes. Why? (**3p**)

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Question 5 (6p)

5a) What is the key assumption behind the GeneMark tool for predicting protein-coding genes in prokaryotes? (**2p**)

5b) How does GeneMark use a Markov model in gene prediction? (**2p**)

5c) Give an example of another gene prediction tool that uses Markov models. (**2p**)