4/26/24, 10:58 PM Homework #5



## 100 Points Possible





## **Unlimited Attempts Allowed**

4/22/2024 to 5/10/2024

∨ Details

Homework #5

Due May 5<sup>th</sup>, 11:59pm

Each homework submission must include:

- An archive (.zip or .gz) file of the source code containing:
  - The makefile used to compile the code on Monsoon (5pts)
  - All .cpp and .h files (5pts)
- A full write-up (.pdf of .doc) file containing answers to homework's questions (5pts), including
  the exact command line needed to execute every subproblem of the homework

The source code must follow the following guidelines:

- No external libraries that implement data structures discussed in class are allowed, unless specifically stated as part of the problem definition. Standard input/output and utilities libraries (e.g. math.h) are ok.
- All external data sources (e.g. input data) must be passed in as a command line argument (no hardcoded paths within the source code (5pts).
- Solutions to sub-problems must be executable separately from each other. For example, via a special flag passed as command line argument (5pts)

For this homework, you will use the *query dataset* located on Monsoon:

/common/contrib/classroom/inf503/human\_reads\_version2.fa

For this homework, you will also need to use the <u>subject dataset</u> (human genome assembly that you used in HW#1). Recall that it is located at:

/common/contrib/classroom/inf503/genomes/human.txt

- This file contains multiple scaffolds that comprise the human genome
- The genome is in FASTA format (see insert)
  - The headers are unique and always begin with the ">" character. These can be discarded for this homework. Each line of genome file is exactly 80 characters long (plus carriage return character)

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• The genomic sequences consist of the following alphabet {A, C, G, T, N}- please replace all instances of 'N' with 'A'. I want to make sure you are dealing with a 4 character alphabet for this homework assignment (makes things much easier).

## Problem #1 (of 1): Prefix Trie

Create a class called *Prefix\_Trie*. The purpose of the class will be to contain a dataset of genomic sequences (queries) and all of the functions needed to operate on this set. Use the <u>prefix trie</u> datastructure to store the genomic fragments of a given size. Here you will be performing fuzzy matching, tolerating up to 1 mismatch.

At minimum, the class must contain(15pts):

- A default constructor
- At least one custom constructor to build a trie from a set of queries (of size n)
- A function to traverse (search) the trie using a genome of size G. Note that you can assume that G >> n. You will need to implement a fuzzy search tolerating up to 1 mismatch (substitutions only). Hint: use a stack to keep track of branches in the tree that need to be explored.
- A destructor

A. (20pts) Basic prefix trie: Pick a random 50K long segment from the human genome assembly. Generate 5K, 50K, 100K, and 1M random 36-mers this segment and store them in the prefix trie. Hint: generate a random starting position somewhere in the segment and read 36 characters starting from that position.

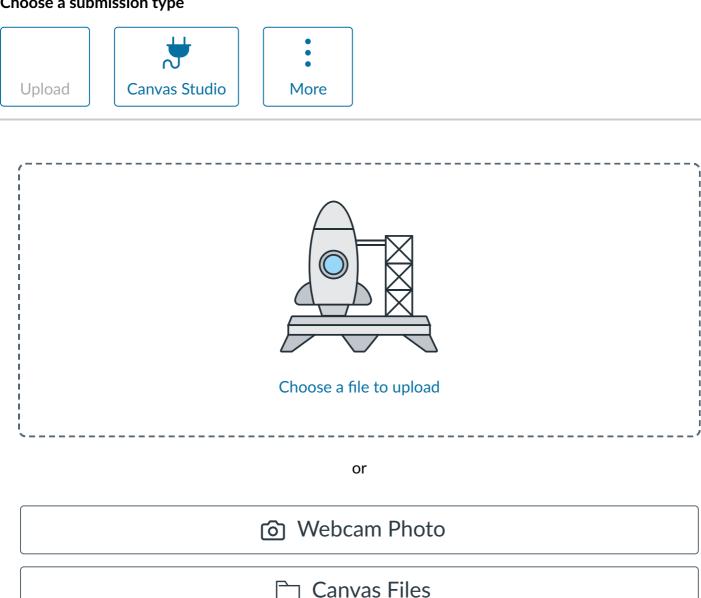
- For each of the 36-mer datasets, what are the sizes of the trie (# of nodes)? Explain the pattern that you observed.
- Iterate through all possible 36-mers in the segment, using each to search / traverse the prefix trie with up to 1 mismatch. How many of your 36-mers had a match? Does it make sense? Explain why.
- **B.** (20pts) Impact of error rate on trie structure: Use the same random 50K long segment from the human genome assembly that you used in part A. Generate 5K, 50K, 100K, and 1M random 36-mers from this segment with 5% per-base error rate and store them in the prefix trie. Hint: repeat the process from part A, except each base of 36-mer has a 5% chance of mutation/error.
  - For each of the 36-mer datasets, what are the sizes of the trie (# of nodes)? Explain differences (if any) between the trie sizes in partA and part B.
  - Iterate through all possible 36-mers in segment, using each to search / traverse the prefix trie with up to 1 mismatch. How many of your 36-mers had a match? Does it make sense? Explain why.

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C. (20pts) Full prefix trie experience: Load the entire query dataset into the prefix trie. Generate a random segment of the human genome of size 100K, 1M, and 100M characters and use the 32mers in this this segment to search against the prefix trie. For this problem, only look for perfect matches (no need for fuzzy matching).

- How long did it take you to find all 32-mers of 100K, 1M, and 100M character segments within the prefix trie? Estimate how long it would take to search the entire human genome.
- How many 'hits' did you find for 100K, 1M, and 100M segments? Estimate how many you would find in the full genome.

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