**Bioinformatics course final exam paper**

**Date: 20 April 2021**

**Question paper 3**

**The total marks for this paper will be 50 marks with 3 analytical questions. Duration is 1 h 30 min.**

**Students should write down the steps and have a rough column for alignment questions. I need to see the working out of the problems with intermediate steps to facilitate allocation of partial credit for the questions. DO NOT leave any question blank.**

**While scanning and sending your answer sheets, include the allocated question paper number for my reference.**

**Do not forget to include your name and roll number in the answer**

**sheet. All the Best!**

1a) Perform **global alignment** for **ACGTACG versus CTCGGC [15 marks]**

| **Scoring scheme:** |
|---|
| Match = 8 |
| Mismatch = -4 |
| Gap penalty = -5 |

2) Perform **local alignment** for the following 2 sequences using PAM matrix **[20 marks]**

SPEAKL
APPEAL

# PAM Substitution Matrix

| | A | R | N | D | C | Q | E | G | H | I | L | K | M | F | P | S | T | W | Y | V |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| A | 4 | -3 | -1 | -1 | -3 | -2 | 0 | 1 | -3 | -2 | -3 | -3 | -2 | -5 | 1 | 1 | 1 | -7 | -4 | 0 |
| R | -3 | 7 | -2 | -4 | -5 | 1 | -3 | -5 | 1 | -3 | -5 | 2 | -1 | -6 | -1 | -1 | -3 | 1 | -6 | -4 |
| N | -1 | -2 | 5 | 3 | -5 | -1 | 1 | -1 | 2 | -3 | -4 | 1 | -4 | -5 | -2 | 1 | 0 | -5 | -2 | -3 |
| D | -1 | -4 | 3 | 5 | -7 | 0 | 4 | -1 | -1 | -4 | -6 | -1 | -5 | -8 | -3 | -1 | -2 | -9 | -6 | -4 |
| C | -3 | -5 | -5 | -7 | 9 | -8 | -8 | -5 | -4 | -3 | -8 | -8 | -7 | -7 | -4 | -1 | -4 | -9 | -1 | -3 |
| Q | -2 | 1 | -1 | 0 | -8 | 6 | 2 | -3 | 3 | -4 | -2 | 0 | -2 | -7 | -1 | -2 | -2 | -7 | -6 | -3 |
| E | 0 | -3 | 1 | 4 | -8 | 2 | 5 | -1 | -1 | -3 | -5 | -1 | -4 | -8 | -2 | -1 | -2 | -9 | -5 | -3 |
| G | 1 | -5 | -1 | -1 | -5 | -3 | -1 | 5 | -4 | -5 | -6 | -3 | -4 | -6 | -2 | 0 | -2 | -9 | -7 | -3 |
| H | -3 | 1 | 2 | -1 | -4 | 3 | -1 | -4 | 7 | -4 | -3 | -2 | -4 | -3 | -1 | -2 | -3 | -4 | -1 | -3 |
| I | -2 | -3 | -3 | -4 | -3 | -4 | -3 | -5 | -4 | 6 | 1 | -3 | 1 | 0 | -4 | -3 | 0 | -7 | -3 | 3 |
| L | -3 | -5 | -4 | -6 | -8 | -2 | -5 | -6 | -3 | 1 | 6 | -4 | 3 | 0 | -4 | -4 | -3 | -3 | -3 | 0 |
| K | -3 | 2 | 1 | -1 | -8 | 0 | -1 | -3 | -2 | -3 | -4 | 5 | 0 | -7 | -3 | -1 | -1 | -6 | -6 | -4 |
| M | -2 | -1 | -4 | -5 | -7 | -2 | -4 | -4 | -4 | 1 | 3 | 0 | 9 | -1 | -4 | -3 | -1 | -6 | -5 | 1 |
| F | -5 | -6 | -5 | -8 | -7 | -7 | -8 | -6 | -3 | 0 | 0 | -7 | -1 | 8 | -6 | -4 | -5 | -1 | 4 | -3 |
| P | 1 | -1 | -2 | -3 | -4 | -1 | -2 | -2 | -1 | -4 | -4 | -3 | -4 | -6 | 7 | 0 | -1 | -7 | -7 | -3 |
| S | 1 | -1 | 1 | -1 | -1 | -2 | -1 | 0 | -2 | -3 | -4 | -1 | -3 | -4 | 0 | 4 | 2 | -3 | -4 | -2 |
| T | 1 | -3 | 0 | -2 | -4 | -2 | -2 | -2 | -3 | 0 | -3 | -1 | -1 | -5 | -1 | 2 | 5 | -7 | -4 | 0 |
| W | -7 | 1 | -5 | -9 | -9 | -7 | -9 | -9 | -4 | -7 | -3 | -6 | -6 | -1 | -7 | -3 | -7 | 12 | -2 | -9 |
| Y | -4 | -6 | -2 | -6 | -1 | -6 | -5 | -7 | -1 | -3 | -3 | -6 | -5 | 4 | -7 | -4 | -4 | -2 | 9 | -4 |
| V | 0 | -4 | -3 | -4 | -3 | -3 | -3 | -3 | -3 | 3 | 0 | -4 | 1 | -3 | -3 | -2 | 0 | -9 | -4 | 5 |


3) Identify the parent for this child with sickle cell anemia **[15 marks] Val His Leu Thr Pro Glu Glu –** normal protein form is present as **Val His Leu Thr Pro Val Glu-** mutated form.
Find the pair who could be the parent of this child.

Pair 1
Father
3`CACGTGGACTGAGGACACCTCTTC 5`
5`GTGCACCTGACTCCTGTGGAGAAG 3`
Mother
3`CACGTGGACTGAGGACACCACTTC 5`
5`GTGCACCTGACTCCTGTGGTGAAG 3`

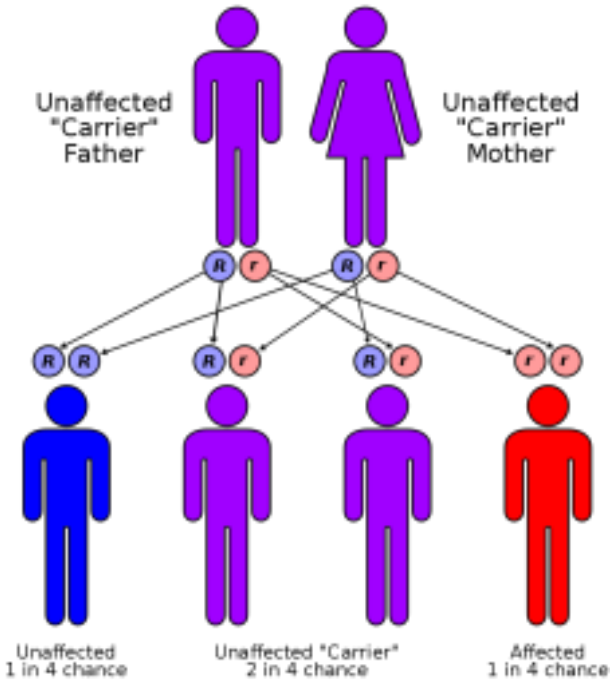
Pair 2
Father
3`CACGTGGACTGAGGACACCACTTC 5`
5`GTGCACCTGACTCCTGTGGTGAAG 3`
Mother

3`CACGTGGACTGAGGACACCACTTC 5`
5`GTGCACCTGACTCCTGTGGTGAAG 3`

Pair 3
Father
3`CACGTGGACTGAGGACACCTCTTC 5`
5`GTGCACCTGACTCCTGTGGAGAAG 3`
Mother
3`CACGTGGACTGAGGACACCTCTTC 5`
5`GTGCACCTGACTCCTGTGGAGAAG 3`

Pair 4
Father
3`CACGTGGACTGAGGACACCTCTTC 5`
5`GTGCACCTGACTCCTGTGGAGAAG 3`
Mother
3`CACGTGGACTGAGGACACCACTTC 5`
5`GTGCACCTGACTCCTGTGGTGAAG 3`
Take additional hint from the following

figure.




**Second letter**

| First letter | U | C | A | G | Third letter |
|---|---|---|---|---|---|
| **U** | UUU, UUC } Phe<br>UUA, UUG } Leu | UCU, UCC, UCA, UCG } Ser | UAU, UAC } Tyr<br>**UAA Stop**<br>**UAG Stop** | UGU, UGC } Cys<br>**UGA Stop**<br>UGG Trp | U C A G |
| **C** | CUU, CUC, CUA, CUG } Leu | CCU, CCC, CCA, CCG } Pro | CAU, CAC } His<br>CAA, CAG } Gln | CGU, CGC, CGA, CGG } Arg | U C A G |
| **A** | AUU, AUC, AUA } Ile<br>**AUG Met** | ACU, ACC, ACA, ACG } Thr | AAU, AAC } Asn<br>AAA, AAG } Lys | AGU, AGC } Ser<br>AGA, AGG } Arg | U C A G |
| **G** | GUU, GUC, GUA, GUG } Val | GCU, GCC, GCA, GCG } Ala | GAU, GAC } Asp<br>GAA, GAG } Glu | GGU, GGC, GGA, GGG } Gly | U C A G |