Emmanuel pyle CRN: 21774 Professor Rinehart-Kim Genome assignment

Chromosome Maps

1. WHAT CHROMOSOME DID YOU CHOOSE? Chromosome 6

2 & 3. STATE THE NUMBER OF GENES AND BASE PAIRS ON THE CHROMOSOME YOU CHOSE. Approximately 1900 genes and 170 million base pairs.

4. LIST ONE GENE THAT IS LOCATED ON THIS CHROMOSOME. The IDDM1 Diabetes gene (insulin-dependent diabetes mellitus).

5. **STATE THE <u>NORMAL</u> FUNCTION OF THE GENE YOU LISTED IN #4**. To make sure insulin is in check and immune cells are not attacking the pancreatic cells. To help regulate glucose homeostasis.

6. **STATE THE POSSIBLE DISEASE(S) RELATED TO THIS GENE**. Diabetes type 1. The body attacks its own insulin and pancreatic cells.

<u>GenBank</u>

7. WHAT IS GENBANK? How we conserve the genetic material of organisms. In vitro storages for example. Genetic material for species can be withdrawn for studying or using for epidemics, like medicine development .

Introduction to BLAST

8. WHAT IS THE TOP SEQUENCE DESCRIPTION MATCH FOR YOUR QUERY SEQUENCE? Homo sapiens CFTR promoter region (LOC111674463) on chromosome 7

9. WHAT DOES THE ENCODED PROTEIN DO IN THE BODY? This gene makes the protein, cystic fibrosis transmembrane conductance regulator that functions as a channel in the membrane of cells to bring in mucus, saliva, and digestive system. Also essential for the regulation of transmembrane chloride reabsorption.

Citation

Betapudi, B., Aleem, A., & Kothadia, J. P. (2020). Cystic Fibrosis And Liver Disease. In *StatPearls*. StatPearls Publishing.

10. FOR WHAT DISEASE IS A MUTATED FORM OF THIS GENE RESPONSIBLE? Cystic fibrosis

11. ON WHAT CHROMOSOME IS THE GENE LOCATED? Chromosome 7

12. WHAT SPECIES (STATE THE SCIENTIFIC NAME) OTHER THAN *HOMO SAPIENS* ALSO HAS A 100% IDENTITY (Ident) FOR THIS SEQUENCE? Gorilla gorilla

13. WHAT IS THE COMMON NAME FOR THIS SPECIES? lowland gorilla

14. DOES IT SURPRISE YOU THAT THIS SPECIES ALSO HAS A 100% SIMILARITY IN IDENTITY? WHY OR WHY NOT? NO, because these walking animals are similar to humans and it is said that we have evolved from them. It's not surprising that this gene is 100% similar to humans that we have probably inherited it from them.

15. HOW MANY GAPS OCCUR BETWEEN THE TWO SEQUENCES (THE ONE YOU SUBMITTED AND THE FIRST ONE THAT IS NOT A HOMO SAPIENS)? There are 0 gaps out of 120.

16. WHAT IS A GAP IN SEQUENCE ALIGNMENTS? Gaps account for the insertions or deletions in the sequence of dna which will signify that there has been mutations.

17. STATE WHAT THE FOLLOWING GENE IS: NC_045512.2

Severe acute respiratory syndrome coronavirus 2 isolate Wuhan-Hu-1 (sars-cov2)

18. SCROLL DOWN THE LIST BELOW THIS SEQUENCE. THERE ARE MANY SEQUENCES THAT LOOK SIMILAR. CAN YOU DETECT WHAT IS DIFFERENT ABOUT THE OTHER SEQUENCES?

The difference is the exactness of the nucleotide sequence at the end which shows the variety of the virus dna. The ones with a smaller percentage of identity comes from the sequence of amino acids that differ.

Introduction to Swiss-Prot to Study Protein Sequences

19. WHAT IS cDNA? Complementary DNA, which is synthesized from a single-stranded RNA or mRNA from the enzyme reverse transcriptase. Usually done by some prokaryotes.

20. USING THE SAME PROGRAM YOU USED IN THE INTRODUCTION TO BLAST ABOVE, WHAT IS THE SEQUENCE MATCH? The match is the Homo sapiens hemoglobin subunit beta (HBB) gene

21. HOW MANY 5' TO 3' FRAMES DID YOU OBTAIN? BASED UPON THE LENGTH OF THE POLYPEPTIDE, WHICH FRAME IS MOST LIKELY THE CORRECT ONE? I obtained 3 of them. It must be Frame 1 that is correct.

Amino Acid Sequence Comparisons

22. DO YOU SEE ANY DIFFERENCES BETWEEN THE TWO AMINO ACID SEQUENCES? Yes there are two differences in sequence starting 361 and 781.

23. IF YOU SAW DIFFERENCES, WHAT WERE THEY?

fibroblast growth factor receptor 3, that controls growth factors in the cell. There is a substitution mutation of glycine and arginine in the 361. There is a deletion of serine in the 781 section.

24. WHAT CHROMOSOME HAS THE GENE THAT ENCODES FOR THE POLYPEPTIDE YOU WERE ANALYZING?

Chromosome 5

25. REFLECT ON ONE THING THAT YOU LEARNED FROM DOING THIS ASSIGNMENT

I learned how to compare DNA between two different organisms and or the genes on the same organism but different individuals.