Chromosome Maps

1. WHAT CHROMOSOME DID YOU CHOOSE?

Chromosome 3

2 & 3. STATE THE NUMBER OF GENES AND BASE PAIRS ON THE CHROMOSOME YOU CHOSE.

About 1900 genes

About 200 million base pairs

4. LIST ONE GENE THAT IS LOCATED ON THIS CHROMOSOME.

MLH1 gene

5. STATE THE FUNCTION OF THE GENE YOU LISTED IN #4.

Codes for colon cancer and recognize errors in the MMR pathways and then initiate the repair process for them.

Introduction to Nucleotide BLAST

6. WHAT IS THE SECOND SEQUENCE DESCRIPTION MATCH FOR YOUR QUERY SEQUENCE?

Homo sapiens CFTR (CFTR) gene, partial cds

7. WHAT DOES THE ENCODED PROTEIN DO IN THE BODY?

CFTR is Cystic Fibrosis Transmembrane Conductance Regulator. This protein controls ion and water secretion and absorption in epithelial tissues.

8. FOR WHAT DISEASE IS A MUTATED FORM OF THIS GENE RESPONSIBLE?

Cystic Fibrosis

9. ON WHAT CHROMOSOME IS THE GENE LOCATED?

7

10. Return to the original nucleotide sequence alignment descriptions. CHOOSE A SPECIES (STATE THE

SCIENTIFIC NAME) OTHER THAN HOMO SAPIENS THAT ALSO HAS A 100% IDENTITY (Per. Ident) FOR THIS SEQUENCE?

Pongo abelii

11. WHAT IS THE COMMON NAME FOR THIS SPECIES?

Orangutan

12. DOES IT SURPRISE YOU THAT THIS SPECIES ALSO HAS A 100% SIMILARITY IN IDENTITY? WHY OR WHY NOT?

This doesn't surprise me at all because homo sapiens have been linked through monkeys throughout history.

13. Return to the original nucleotide sequence alignment DESCRIPTION. Find the first match that has less than 100% similarity identity. Click on the description to answer this question.

a. WHAT IS THE GENUS AND SPECIES WITH THIS NUCLEOTIDE SEQUENCE?

Sapajus apella

b. WHAT IS THE COMMON NAME?

tufted capuchin

c. ARE THERE ANY GAPS BETWEEN THE TWO SEQUENCES (THE ONE YOU ORIGINALLY SUBMITTED AND ONE THAT HAS LESS THAN 100% QUERY COVER)?

Yes, there is one gap.

14. WHAT IS A GAP IN SEQUENCE ALIGNMENTS?

A gap shows where genetic mutations have occurred, whether it be insertion or deletion.

FOR EACH, STATE WHAT THE GENE IS (#15-18). (Again, describe the gene or gene

product, not the nucleotide sequence.)

15. NM_145556

Mus musculus TAR DNA binding protein (Tardbp), transcript variant 1, mRNA

16. NM_013444

Homo sapiens ubiquilin 2 (UBQLN2), mRNA

17. NM_001010850

Homo sapiens FUS RNA binding protein (FUS), transcript variant 2, non-coding RNA

18. KJ174530

Homo sapiens superoxide dismutase-1 (SOD-1) gene, exon 1 and partial cds

19. Search Google to answer the following: WHAT DISEASE IS ASSOCIATED WITH MUTATIONS OF THE GENES REFERENCED IN #15-#18? WHAT IS A "COMMON NAME" OF THE DISEASE?

Amyotrophic Lateral Sclerosis 10 With Or Without Frontotemporal Dementia and Motor Neuron Disease. Common name: Lou Gehrig's disease

20. BLAST is possible because of the submission of DNA sequences to GenBank. WHAT IS GENBANK?

GenBank is a database that provides open access to the most recent and comprehensive DNA sequence information in the science community.

Introduction to Protein BLAST

21. First, answer this question: WHAT IS cDNA?

cDNA has coding and noncoding sequences and is typically used for gene cloning.

22. WHAT IS THE SEQUENCE MATCH?

The sequence match is 63%. Top match is beta-globin [Homo sapiens]

23. DO YOU SEE ANY DIFFERENCES BETWEEN THE TWO AMINO ACID SEQUENCES? (Look for a space between same amino acid comparison for both sequences.)

Yes

24. IF YOU SAW DIFFERENCES, WHAT WERE THEY?

Query 780 SSSGDDSVFAHDLLPPAPPSSGGSRT 805

SSSGDDSVFAHDLLPPAPPSSGGSRT

Sbjct 781 SSSGDDSVFAHDLLPPAPPSSGGSRT 806

25. ARE THERE ANY GAPS IN THE SEQUENCE ALIGNMENT?

Yes there is one or 2 gaps at the 721 segment

26. WHAT GENE ENCODES FOR THE POLYPEPTIDE YOU WERE ANALYZING?

fibroblast growth factor receptor 3 isoform 1 precursor [Homo sapiens]

27. WHAT IS THE FUNCTION OF THIS PROTEIN?

This protein binds acidic and basic fibroblast growth hormones and plays a role in bone development and maintenance.

28. WHAT HUMAN DISEASE IS CAUSED BY A MUTATION IN THIS GENE?

craniosynostosis and skeletal dysplasia

29. WHAT IS THE CONNECTION AMONG THE FOLLOWING: NIH, NLM, NCBI, and HHS? (What do the abbreviations stand for? Who oversees what?)

All of these are run by the U.S. Department of Health and Human Services. The NIH (National Institutes of Health) oversees biomedical and public health research. The NLM (National Library of Medicine) provides access to biomedical information. The NCBI (National Center for

Biotechnology Information) is a division of NLM that develops databases and tools that store biological data.

30. WHAT WAS ONE POSITIVE THING AND ONE NEGATIVE THING YOU ENCOUNTERED WHILE DOING THIS ASSIGNMENT?

One positive thing is this made me understand database researching a little bit better and was good practice. One negative thing is some questions were a little confusing as I didn't know how or where to find some of the information asked for.