Chromosome Maps

Assignment Goal: To use the Internet-based Genes and Disease site (NCBI) to view the assignment of genes to chromosomes.

Assignment: Access the Genes and Disease site at <u>http://www.ncbi.nlm.nih.gov/books/NBK22183/</u>

Under "Contents", select "Chromosome Map" (at the very bottom).

A karyotype will appear.

Click on a chromosome.

1. WHAT CHROMOSOME DID YOU CHOOSE? Chromosome 13

2 & 3. STATE THE NUMBER OF GENES AND BASE PAIRS ON THE CHROMOSOME YOU CHOSE. 2. 800 genes

3. <u>110 million base pairs</u>

4. LIST ONE GENE THAT IS LOCATED ON THIS CHROMOSOME. BRCA2

5. STATE THE FUNCTION OF THE GENE YOU LISTED IN #4. The BRCA2 gene helps in repairing radiation-induced breaks in double-stranded DNA.

Introduction to Nucleotide BLAST

Assignment Goal: To use the Internet-based site BLAST, Basic Local Alignment Search Tool (NCBI), to search for similarities between nucleotide sequences.

Assignment: Access the BLAST site at http://blast.ncbi.nlm.nih.gov/Blast.cgi

Click on "Nucleotide Blast"

Assume that you found this nucleotide sequence when you cloned a piece of gene in the laboratory in which you work:

aattggaage aaatgacate acageaggte agagaaaaag ggttgagegg eaggeaceea gagtagtagg tetttggeat taggagettg ageceagaeg geeetageag ggaceecage

Enter the above sequence (you may copy and paste) into the "Enter Query Sequence" box at the top of the page. Under "Program Selection" near the bottom of the page, choose "somewhat similar sequence (blastn)"

Click the "BLAST" button at the bottom of the page to run the search.

Give some time for the results of your search to show up.

You will be given significant matches for the sequence that you entered.

6. WHAT IS THE **SECOND** SEQUENCE DESCRIPTION MATCH FOR YOUR QUERY SEQUENCE? <u>Homo</u> <u>sapiens CFTR (CFTR) gene, partial cds</u>

7. WHAT DOES THE ENCODED PROTEIN DO IN THE BODY? The encoded protein functions as a chloride channel, making it unique among members of this protein family, and controls ion and water secretion and absorption in epithelial tissues. Channel activation is mediated by cycles of regulatory domain phosphorylation, ATP-binding by the nucleotide-binding domains, and ATP hydrolysis.

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

9. ON WHAT CHROMOSOME IS THE GENE LOCATED? 7q31.2

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? <u>Gorilla gorilla gorilla</u>

11. WHAT IS THE COMMON NAME FOR THIS SPECIES? Lowland gorilla

12. DOES IT SURPRISE THAT THIS SPECIES ALSO HAS A 100% SIMILARITY IN IDENTITY? WHY

OR WHY NOT? It doesn't suprise me that Gorillas have a 100% similarity in identification. They share many common things with humans making it a lot easier for scientists to study.

13.

- a. WHAT IS THE GENUS AND SPECIES WITH THIS NUCLEOTIDE SEQUENCE? Nomascus leucogenys
- b. WHAT IS THE COMMON NAME? Northern white-cheeked gibbon

c. HOW MANY GAPS OCCUR BETWEEN THE TWO SEQUENCES (THE ONE YOU ORIGINALLY SUBMITTED AND ONE THAT HAS LESS THAN 100% QUERY COVER)? . <u>O gaps</u>

14. WHAT IS A GAP IN SEQUENCE ALIGNMENTS? Is when one ore more amino acid that had been deleted in the squence.

FOR EACH, STATE WHAT THE GENE IS (#15-18). (Again, give the description of the gene or gene product, not the nucleotide sequence.)

- 15. NM_145556 : TAR DNA binding protein: Mus musculus TAR DNA binding protein (Tardbp), transcript variant 1, mRNA
- 16. NM_013444 : UBQLN2: ubiquilin 2: Homo sapiens ubiquilin 2 (UBQLN2), mRNA
- 17. NM_001010850 : FUS RNA binding protein: Homo sapiens FUS RNA binding protein (FUS), transcript variant 2, non-coding RNA
- 18. KJ174530 : Superoxide dismutase 1: Homo sapiens superoxide dismutase-1 (SOD-1) gene, exon 1 and partial cds
- 19. WHAT DISEASE IS ASSOCIATED WITH MUTATIONS OF THE GENES REFERENCED IN #15-#18?
- WHAT IS A "COMMON NAME" OF THE DISEASE? <u>Amyotrophic lateral sclerosis (ALS)</u>

20. WHAT IS GENBANK? Is data bank that contains dna sequences that are available for the public to access and reasearch.

Introduction to Protein BLAST

Assignment Goal: To use the Internet-based site BLAST, Basic Local Alignment Search Tool (NCBI), to 1) translate cDNA and 2) to search for similarities between amino acid sequences.

Assignment: Access the BLAST site at http://blast.ncbi.nlm.nih.gov/Blast.cgi

Click on "tblastx". (This is a smaller button next to "Nucleotide BLAST".

21. First, answer this question: WHAT IS cDNA? Is complementary DNA which is syntethic DNA that has been transcribed from a specific mRNA.

Enter the following cDNA sequence on "blastx":

ACATTTGCTTCTGACACAATTGTGTTCACTAGCAACCTCAAACAGACACCATGGTGCATCTGACTCC TGAGGAGAAGTCTGCCGTTACTGCCCTGTGGGGGCAAGGTGAACGTGGATGAAGTTGGTGGTGAG GCCCTGGGCAG

22. WHAT IS THE SEQUENCE MATCH? 63% beta-globin [Homo sapiens]

Assignment: While still on BLAST, choose "Protein BLAST". Check the box which says "Align two or more sequences".

Copy and paste the following sequence into the "Enter Query Sequence" box: Person 1/Sequence 1:

MGAPACALALCVAVAIVAGASSESLGTEQRVVGRAAEVPGPEPGQQEQLVFGSGDAVELSCPPP GGGPMGPTVWVKDGTGLVPSERVLVGPQRLQVLNASHEDSGAYSCRQRLTQRVLCHFSVRVTD APSSGDDEDGEDEAEDTGVDTGAPYWTRPERMDKKLLAVPAANTVRFRCPAAGNPTPSISWLK NGREFRGEHRIGGIKLRHQQWSLVMESVVPSDRGNYTCVVENKFGSIRQTYTLDVLERSPHRPI LQAGLPANQTAVLGSDVEFHCKVYSDAQPHIQWLKHVEVNGSKVGPDGTPYVTVLKTAGANT TDKELEVLSLHNVTFEDAGEYTCLAGNSIGFSHHSAWLVVLPAEEELVEADEAGSVYAGILSYG VGFFLFILVVAAVTLCRLRSPPKKGLGSPTVHKISRFPLKRQVSLESNASMSSNTPLVRIARLSSGE GPTLANVSELELPADPKWELSRARLTLGKPLGEGCFGQVVMAEAIGIDKDRAAKPVTVAVKML KDDATDKDLSDLVSEMEMMKMIGKHKNIINLLGACTQGGPLYVLVEYAAKGNLREFLRARRPP GLDYSFDTCKPPEEQLTFKDLVSCAYQVARGMEYLASQKCIHRDLAARNVLVTEDNVMKIADF GLARDVHNLDYYKKTTNGRLPVKWMAPEALFDRVYTHQSDVWSFGVLLWEIFTLGGSPYPGIP VEELFKLLKEGHRMDKPANCTHDLYMIMRECWHAAPSQRPTFKQLVEDLDRVLTVTSTDEYLD LSAPFEQYSPGGQDTPSSSSGDDSVFAHDLLPPAPPSSGGSRT

Copy and paste the following sequence into the "Enter Subject Sequence" box: Person 2/Sequence 2:

MGÅPACALALCVAVAIVAGASSESLGTEQRVVGRAAEVPGPEPGQQEQLVFGSGDAVELSCPPP GGGPMGPTVWVKDGTGLVPSERVLVGPQRLQVLNASHEDSGAYSCRQRLTQRVLCHFSVRVTD APSSGDDEDGEDEAEDTGVDTGAPYWTRPERMDKKLLAVPAANTVRFRCPAAGNPTPSISWLK NGREFRGEHRIGGIKLRHQQWSLVMESVVPSDRGNYTCVVENKFGSIRQTYTLDVLERSPHRPI LQAGLPANQTAVLGSDVEFHCKVYSDAQPHIQWLKHVEVNGSKVGPDGTPYVTVLKTAGANT TDKELEVLSLHNVTFEDAGEYTCLAGNSIGFSHHSAWLVVLPAEEELVEADEAGSVYAGILSYR VGFFLFILVVAAVTLCRLRSPPKKGLGSPTVHKISRFPLKRQVSLESNASMSSNTPLVRIARLSSGE GPTLANVSELELPADPKWELSRARLTLGKPLGEGCFGQVVMAEAIGIDKDRAAKPVTVAVKML KDDATDKDLSDLVSEMEMMKMIGKHKNIINLL GACTQGGPLYVLVEYAAKGNLREFLRARRPPGLDYSFDTCKPPEEQLTFKDLVSCAYQVARGME YLASQKCIHRDLAARNVLVTEDNVMKIADFGLARDVHNLDYYKKTTNGRLPVKWMAPEALF DRVYTHQSDVWSFGVLLWEIFTLGGSPYPGIPVEELFKLLKEGHRMDKPANCTHDLYMIMREC WHAAPSQRPTFKQLVEDLDRVLTVTSTDEYLDLSAPFEQYSPGGQDTPSSSSSGDDSVFAHDLLP PAPPSSGGSRT

Click on the "BLAST" box.

After you receive results, choose the "Description" box for more information about the alignment.

23. DO YOU SEE ANY DIFFERENCES BETWEEN THE TWO AMINO ACID SEQUENCES? YES

24. IF YOU SAW DIFFERENCES, WHAT WERE THEY? On the line 721-780 An S is missing compared to the Subject.

25. ARE THERE ANY GAPS IN THE SEQUENCE ALIGNMENT? Yes.

26. WHAT GENE ENCODES FOR THE POLYPEPTIDE YOU WERE ANALYZING? <u>fibroblast growth factor receptor 3</u>

isoform 1 precursor

27. WHAT IS THE FUNCTION OF THIS PROTEIN? <u>The extracellular portion of the protein interacts with fibroblast growth</u> <u>factors, setting in motion a cascade of downstream signals, ultimately influencing mitogenesis and differentiation. This particular family member binds</u> <u>acidic and basic fibroblast growth hormone and plays a role in bone development and maintenance.</u>

28. WHAT HUMAN DISEASE IS CAUSED BY A MUTATION IN THIS GENE? Craniosynostosis and multiple types of skeletal dysplasia.

29. WHAT IS THE CONNECTION AMONG THE FOLLOWING: NIH, NLM, NCBI, and HHS? <u>They are all</u> organization responsible for provide collect research information about biomedical and genetic information.

30. REFLECT ON ONE THING THAT YOU LEARNED FROM DOING THIS ASSIGNMENT. <u>I learned how to</u> compare Human DNA sequences to other species.