Bioinformatics practice: Zoo456

Zoology Department

- 1. Go to the NCBI homepage http://www.ncbi.nih.gov/
- 2. Take a few minutes to look around the site. The goal is to familiarize yourself with a few key components of the NCBI.
 - a. What is the name of one interesting resource or database shown in the blue box on the left? What do you think is its function or purpose?

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b. What is one interesting resource listed in the Popular Resources menu on the right? What do you think is its function or purpose?

- 3. Find the search box in the center of the webpage (black box in above image). This search box uses the NCBI search engine Entrez to look for your search term (or "query") across all of the databases at the NCBI.
- 4. Type "BRCA1" into the Search box. Make sure there is no space between BRCA and 1. Click Search. BRCA1 is a tumor suppressor gene that normally prevents cancer. Mutations in this gene are associated with increased risk of hereditary breast cancer and ovarian cancer when normal function is lost. The white box to the left of each database contains the number of "hits" returned from that database (see screen shot, above). This is like searching in iTunes[®] without specifying
- 5. "feature (PC: "Control+F" Mac: "Command+F") to Find "Map Viewer." Click on the "Map Viewer" link. The resulting page categories like ringtones, podcasts, movies, TV, or songs.
 - a. Why are we searching for BRCA1?

- b. The Nucleotide database has DNA sequences that have been loaded onto the NCBI database. How many times is 'BRCA1' cited in the Nucleotide database?
- c. The PubMed database has the articles that have been published about a specific gene or disease. How many times is 'BRCA1' cited in the PubMed database?
- d. Compare the numbers you got for Questions a and c. Do these relative numbers surprise you? What does this tell you about the BRCA1 gene? Explain
- 6. Go back to the NCBI homepage by clicking the NCBI logo in the upper left corner of the screen. This search shows that there is a lot of information at the NCBI! It can be challenging to try to make sense of it all. Let's start with something more familiar.
- 7. Click the "All Resources" link from the list of resources on the left side of the screen.

- 8. Find "Map Viewer." Click on the "Tools" tab and either scroll through the alphabetical list, or use the "Findis called Map Viewer and it allows us to search the genomes of many different organisms, including humans
- 9. Open the Search menu, select Homo sapiens from the pull-down menu, and click "Go."
- 10. Now we can see the Homo sapiens (human) genome view. A genome is all of the genetic information in an organism. Each figure you see in the "genome view" represents a pair of chromosomes. Most of the chromosomes are numbered, but a few are not. The abbreviations "X" and "Y" refer to the human sex chromosomes.



- a. How many different types of chromosomes do you see?
- b. What does "MT" represent? [Note: you can click the "MT" link to find out.]
- c. With the exception of MT, the chromosomes of the human genome are in pairs. X and Y are a pair. Using this information and the information from your answer to Question 9A, how many pairs of chromosomes are in the human genome?
- 11. The Breast Cancer Susceptibility gene BRCA1 is on chromosome 17 in humans. [Click on the link below chromosome 17.] Explore some of the links and views. What do you see when you click on chromosome 17? Explore some of the links on the picture, and write down two things you found interesting, such as the description of other genes that are also found on chromosome 17.
- 12. To find the location of the BRCA1 gene, type "BRCA1" in the "Search" box at the top left of the screen, and click "Find in This View." Scroll through the Map of Chromosome 17 and locate the BRCA1 gene, which should be highlighted in pink. "BRCA1" will be found in the list of Symbols. You can also use the "Find" feature (PC: "Control+F" Mac: "Command+F"), which will highlight in yellow every mention of "BRCA1," including the BRCA1 gene. Draw a picture of chromosome 17 and show the approximate location of BRCA1 on this chromosome.
- 13. Click on the BRCA1 link. This will take you to Entrez Gene, which provides a summary of the information available at the NCBI for BRCA1. Scroll through the webpage and explore some of the information available. Scroll down the webpage to the section titled "Gene Ontology." There is a table titled "Function." List three of the functions that the BRCA1 protein performs.
- 14. To learn about all of the phenotypes associated with mutations of BRCA1, return to the top of the web page and from the "Table of Contents" on the right, select "Phenotypes." This will bring you to the portion of the web page that contains the phenotype information for BRCA1.
 - a. Based on what you've learned in class, what is a phenotype?
 - b. What phenotypes are associated with mutations in the BRCA1 gene? (You don't need to click the links.)
- 15. Return to the Table of Contents at the top of the page and click "Reference Sequences." This will take you to the portion of the webpage that contains the actual genetic sequence of the BRCA1 gene.

- 16. Reference sequences are DNA or protein sequences that scientists, doctors and genetic counselors use to study genes like BRCA1. You can download these sequences in different formats. For this exercise, click "FASTA" (which is sometimes pronounced FAST-ay).
- 17. This link takes you to the FASTA sequence for BRCA1. Scroll through the web page. This gene is very large!
 - a. What four letters make up this long sequence?
 - b. Based on what you've learned in class, what do these letters represent?
- 18. Return to the NCBI homepage by clicking on the NCBI icon on the top left of the web page.
- 19. Type BRCA1 in the Search box and select "Nucleotide" from the pull-down menu beside the Search box, to limit your search to the database containing all of the DNA and RNA (Nucleotide) sequences. Click the "Search" button.
- 20. What other organisms have BRCA1 genes? You can scroll through the list of organisms, but note that these are listed by the scientific name of the organism (Genus and species), not the common name. For example, Homo sapiens is the scientific name for humans. Also, the Top Organisms (or the organisms with the most "hits") are listed on the right. Helpful Hint: Hold your cursor over the species name to see the common name appear. Alternatively, you can perform an internet search to find the common name(s) of your organisms. List three organisms other than humans that have BRCA1 genes. Include both the scientific and common names.
- 21. Look back at your list of functions for the BRCA1 gene (question #12). Does it surprise you that so many organisms share the BRCA1 gene? Explain
- 22. What kind of information can you find at the National Center for Biotechnology Information? Summarize what you have learned today by listing three types of information found at the NCBI.

Using <u>NCBI</u> (National Center for Biotechnology Information) database for collecting information about certain gene.

Each student will work with a different gene. A. Find a gene on a chromosome using NCBI Map Viewer, then follow the below questions: (Hint: Select only related Homo gene to Sapiens). _ Chromosome # contains the largest number of gene. 1. # 2. Chromosome contains the desired gene. B. Using OMIM (Online Mendelian Inheritance in Man) and PubMed database to get information about gene. then follow the below steps: The MIM is 1. number 2. Mention function encoded the of the protein by gene

3. The PMID number of the latest paper _____ and its URL is

C. Attach the sequence of nucleotides and amino acids of assigned gene as a text file with other your files.

D. Examine the protein structure from NCBI's 3D structure viewer (Cn3D).
1. The number of models that you find is ______
2. The MMDB ID of the first model of protein is ______ and PDB ID

is_

3. Save the molecule for opening by Cn3D program and then keeping it with other your files.

#	Gene
1	ERCC2
2	Leptin
3	Resistin
4	Adiponectin
5	XRCC1
6	ERCC1
7	NER
8	BRCA1
9	HNPCC
10	MYH
11	XRCC3
12	XPD
13	XPF
14	XPG