BIOL 312
FINAL ASSIGNMENT
SPRING 2020

The final assignment is 25% of your grade. There are 9 questions to be answered. You will work individually to complete this assignment. Each student is assigned a genetic disease/trait (see BIOL312_StudentList_FinalAssignment_Spring2020 document to find your assigned disease/trait). Please answer all questions below for your assigned disease/trait.

Deadline for submission: 10 June 2020, 23:59

*****For each question, please provide the detailed name of the software/tool/database used and the relevant accession numbers.*****

1- Considering your assigned genetic disease/trait, identify a gene responsible for it. Is this a monogenic disease/trait? (2 points)

2- Please provide the following details with regards to the gene from question 1 (2 points):
- **A-** Number of exons/introns
- **B-** Size of the gene in base pairs

3- Which variant(s) in this gene are leading to your assigned disease/trait? Please list all. If more than three variants, list only three of them. For each variant, please answer the questions below (4 points):
- **A-** What type of a variant is it? Please provide all specific details possible.
- **B-** What is the frequency of this variant in two different populations?
- **C-** What is the effect of each variant on the gene/protein?
- **D-** Please provide +/-50 bp flanking sequence of each variant.

4- How many domains does the protein encoded by this gene have? List their names. Are any of the variants you listed above found in any of these domains? (3 points)

5- In three different species, compare one of the domains of this protein. How many fully-conserved/identical/semi-conserved amino acids are there? Please list all three species, provide a screenshot of the alignments and summarize the comparisons as requested for full marks. (3 points)

6- Please provide a 3D representation of the protein and state the number of subunits if multiple subunits are present using PDB. Please provide another 3D representation of the protein using SWISS-MODEL. Then provide two differences between PDB and SWISS-MODEL i.e. which different kinds of information can you access using PDB vs. SWISSMODEL about your protein. (5 points)

7- Are any animal models for studying this gene? Very briefly describe one of the animal models. (1 point)

8- Are there any variants found in this gene that are associated with *other genetic diseases/traits*? If yes, which disease(s)/trait(s) and variant(s). Please list all. If more than three variants, list only three of them and their associated disease(s)/trait(s). (3 points)

9- Provide " three publications related to your gene. Which keywords did you use for your search? (2 points)