**BINF 732 – Genomics**

**Final Exam**

**Due May 16, 2022**

**Please answer all questions. Please limit answers to one page per question.**

1. **What is polyploidy? Please give one example each of when it might be beneficial and another example when it might be harmful?**
2. **There are several tools to perform phenotype prediction from the genome sequence. These include standalone tools (SIFT, Polyphen, MutationAssessor, I-Mutant, FatHMM), and consensus tool (Condel, Pon-P, PredictSNP). Consider the amyloid beta protein (UniProtKB - P05067** [**https://www.uniprot.org/uniprot/P05067**](https://www.uniprot.org/uniprot/P05067)**). Choose 8 of the SNP variants and predict pathogenicity. Pick another gene with variants and see how well the tools make predictions. What can you say about the accuracy of standalone vs the consensus-tools? Please note that the PredictSNP can take over a week for output to be returned due to a long queue.**
3. **The term ‘junk DNA’ refers to the approximately 98% of the genome that is not coding for genes. It was coined as it was believed that this DNA did not do anything. Please comment whether or not this is true and support your answer with details.**
4. **Describe how the genome is regulated taking into the account both the regulation of expression and the structure regulation of the genome.**