Name:

Email address:

**Assignment for Laboratory #3**

**Due 11:59pm on Feb 19th, email to** [**fcbb2homework@gmail.com**](mailto:fcbb2homework@gmail.com)**.**

In OpenCRAVAT, go to the Store tab and install the annotators for ClinVar, gnomAD, and PharmGKB. Run an OpenCRAVAT job on your input file with these 3 annotators.

1. Are there any “Pathogenic” ClinVar variants are in the sample? What are they?
2. Are there any variants with PharmGKB annotations in the sample? What are they?
3. If there are “Pathogenic” variants, do they have allele frequency in gnomAD less than 0.01? Would you expect the allele frequency to be this low? Why or why not?
4. “Dominant” disorders can be caused by a single, heterozygous variant. “Recessive” disorders require two copies of the variant, i.e. homozygous for the disease-causing allele. Are any of the diseases identified by ClinVar in this sample known to exhibit dominant inheritance? Hint: Click through to ClinVar explore the literature about the disease.
5. In this sample, do any of the PharmGKB findings carry the potential to impact the treatment of the diseases discovered by ClinVar?