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**Assignment for Laboratory #3**

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

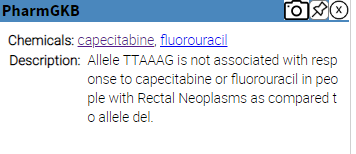
In OpenCRAVAT, go to the Store tab and install the annotators for ClinVar, gnomAD, and PharmGKB. Run an OpenCRAVAT job on your input file with these 3 annotators.

1. Are there any “Pathogenic” ClinVar variants are in the sample? What are they?

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| Chrom | Position | Ref\_Base | Alt\_Base | Disease\_Name |
| Chr1 | 114716126 | C | G | Acute myeloid leukemia|Multiple myeloma|Cutaneous|Non-small cell lung cancer |
| Chr1 | 237445489 | - | T | Long QT syndrome|Cardiovascular phenotype |
| Chr17 | 7676071 | TTTGTTTG | - | Qvarian Neoplasms |
| Chr5 | 177211067 | A | - | Inborn genetic diseases |
| Chr17 | 43092696 | CAA | - | Hereditary breast and ovarian cancer syndrome|Hereditary cancer-predisposing syndrome |
| Chr9 | 95467334 | - | A | Gorlin syndrome |
| Chr3 | 37012099 | G | A | Hereditary nonpolyposis colon cancer|Hereditary cancer-prediposing syndrome |
| Chr4 | 1001742 | T | C | Mucopolysaccharidosis type I|Hurler syndrome |
| Chr17 | 43094529 | - | A | Hereditary cancer-predisposing syndrome |

1. Are there any variants with PharmGKB annotations in the sample? What are they?

Yes, variants of [dbSNP: rs11280056](https://www.ncbi.nlm.nih.gov/snp/rs11280056) with a PharmGKB annotations



chemical is capecitabine; fluorouracil

Chemical ID is: <https://www.pharmgkb.org/chemical/PA448771>

Phenotype Category is: efficine

1. 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?

Yes, there are Mucopolysaccharidosis type I , Hurler syndrome ,

And Deficiency of acetyl-CoA acetyltransferase

Because the frequency of genetic mutations is very small, the probability of developing the disease after mutation is even smaller.

1. “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.

Hypercholesterolaemia and paroxysmal tachycardia is Dominant disorders

1. In this sample, do any of the PharmGKB findings carry the potential to impact the treatment of the diseases discovered by ClinVar?

Yes, such as in the [dbSNP: rs11280056](https://www.ncbi.nlm.nih.gov/snp/rs11280056)

So the capecitabine and fluorouracil can be applied to the treatment of the diseases of Capecitabine response -efficacy and fluorouracil response – efficacy.