Exercises for Variation databases

While searching databases pick 10 amino acid substitutions, insertions, and deletions for further analyses, if available.

Investigate your gene and find out from

SwissProt

1. What types of variants are there?

2. What is the total number of variants?

UCSC browser

3. Visualize human mRNAs in your gene

dbSNP

4. How many SNVs are there in total in your gene? And of which types.

5. How many intron variants and how many frameshift variants are there?

ClinVar

6. Are there insertions and untranslated region variants in your gene?

7. What can you say about reliability of these variant data?

Frequencies

8. Investigate frequencies for a couple of variants.

GWAS catalog

9. How many associations are there and in what traits?

10. Do the results make medical sense in regards to what is known about your gene?

COSMIC

11. How many variants cause amino acid substitutions?

12. Look at a half a dozen variants, what can you say about their tissue distribution?

13. How does the tissue distribution correlate with the primary disease of the gene?

Variant naming

15. Generate systematic names at different levels for the selected variants