Exercises for Systematics

Choose one of the following human genes for your analysis: *ZAP70, JAK3, MSH6, NLM1, RAG1, ELANE, AIRE, SH2D1A, COMT, BLNK, F9, MLH1*

Everybody investigates a different gene. Work on the same gene/protein throughout the course.

If some data is missing for your gene use *ADA* as backup.

Send your finished assignments to [mauno.vihinen@med.lu.se](mailto:mauno.vihinen@med.lu.se)

HGNC and VGNC

1. Find alias names and symbols.

2. Is there a mouse homologue?

MANE

3. Find MANE transcript for your gene.

RefSeq

4. How many transcripts are there and how do they differ?

UniProt

5. How many variants are there in SwissProt and of which types?

EC

6. What is the EC number for the protein and what does it mean?

HGVS

7. How are frameshift variations systematically named?