Lab sheet#3

<u>Practicing ENSEMBL</u>

Objective:

- To know how to brows different data in ENSEMBL.

<u>Using ENSEMBL genome browser, search for the human TAC1 gene to answer the following questions:</u>

1. What is the ENSEMBL identifier of the gene?

2. On which chromosome is this gene located?

3. Show the graphical position of the gene on the chromosome (region in details).

4. How many transcripts (splice variants) has ENSEMBL annotated for it?

5. Are these transcribed from the forward or from the reverse strand of the genome assembly?

6. Which transcript has a CCDS record associated with it?

7. Show how each variant differ from the other by showing the transcript diagram.

8. What is the longest transcript, and how long is the protein it encodes? Retrieve the amino acid sequence.

9. How many exons does it have?

10. Are any of the exons completely or partially translated?

11. Retrieve the sequence of the transcript.

12. Is it possible to monitor the expression of TAC1-201 using the Illumina microarray?

13. What is the function of TAC1?

14. Are there any diseases associated with variants in this gene?

15. Retrieve the TAC1 gene sequence.

16. Find two SNPs associated with the gene.

✓ **Exercise :**

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Search for the human PPT2 gene:

  ✓ On which chromosome is this gene located?
  ✓ How many transcripts has ENSEMBL annotated for it?

  ✓ How many completely translated exons does PPT2-249 have?
  ✓ Retrieve the sequence of PPT2-248 transcript.
  ✓ Find two diseases associated with **PPT2 gene variants in human.**

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