

Lab sheet 3**Practicing Ensembl****Objective:**

- To know how to browse different data in Ensembl.

Using Ensembl genome browser, find detailed information about the human 5'-aminolevulinate synthase 2 (ALAS2) gene by answering the following questions:

1. What is the ENSEMBL identifier of the gene?
2. Have a look at the External References. What is the function of this gene?
3. On which chromosome is this gene located?
4. Show the graphical position of the gene on the chromosome (Region in details).
5. Retrieve the gene sequence.
6. Are there any diseases associated with variants in this gene?
7. Find two SNPs associated with the gene.
8. How many transcripts (splice variants) has ENSEMBL annotated for it?
9. Are these transcribed from the forward or from the reverse strand of the genome assembly?
10. Which transcript has a CCDS record associated with it?
11. Show how each variant differ from the other by showing the transcript diagram.
12. What is the longest transcript?
 - a. How long is the protein it encodes? Retrieve the amino acid sequence.
 - b. How many exons does it have?
 - c. Are any of the exons completely or partially translated?
 - d. Retrieve the sequence of the transcript?
 - e. Is it possible to monitor the expression of this transcript using the Illumina microarray?

Exercise:

Search for the human PPT2 gene:

- ✓ Find two diseases associated with **PPT2 gene in human.**
- ✓ On which chromosome is this gene located?
- ✓ How many completely translated exons does PPT2-249 have?
- ✓ Retrieve the sequence of PPT-248 transcript.