Ensembl Browser Workshop





Ensembl Browser Workshop

VEPTC 2024 Monday, 14th to Tuesday, 16th October 2024

helpdesk@ensembl.org

Welcome to the *Living Document* for this Ensembl Browser workshop. Everyone who is registered for the course has access to edit this document. Please feel free to use this document to ask questions to the Ensembl team throughout the workshop. If you wish to ask questions privately, please do not hesitate to contact the Ensembl Helpdesk.

The *Living Document* is a great way of capturing the knowledge exchanged during the course and saving it for future use by yourselves and those who can't attend this course. Remember - you don't have to contribute, but any additions will be welcomed!

The course learning outcomes are:

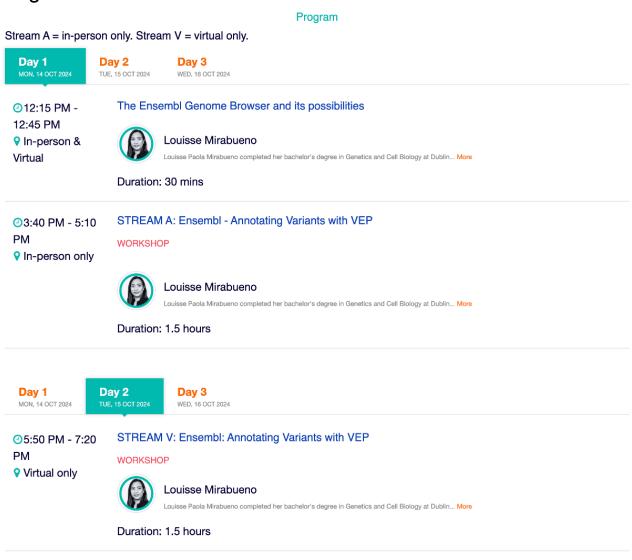
- Access the main data types through the Ensembl web browser platform: genomes, genes, genetic variation and regulatory features.
- Explore information about genes and transcripts, including their sequences and variants found within the genomic region
- Analyse genomic variants, their associated phenotypes, and annotate your own variation data using the Ensembl Variant Effect Predictor (VEP).





Course Overview

Programme



Trainer

Louisse Paola Mirabueno

Demo, exercises and slides available to download

at https://training.ensembl.org/events/2024/2024-10-14-VEPTC browser





Questions

If you have any questions/problems that you would like to share and apply to the whole class please add them below.

Write your **question** after the last one you can see in this document (you can add any screenshots if you think it might help. We will **answer** your question underneath.

- 1. Q: This question is regarding the first of the VEP exercises. The answer mentions that two of the three variants are predicted to have missense consequences of pathogenicity (PolyPhen score), however, I have found only one variant is associated with this (for coordinate 117531038). I followed the steps in the video answer, but my result was different. Do you know why this would be?
 - A:
- 2. Q:
 - A:
- 3. Q:
 - A:
- 4. Q:
 - A:
- 5. **Q**:
 - A:
- 6. Q:
 - A:
- 7. Q:
 - A:
- 8. **Q**:
 - A:
- 9. Q:
 - A:
- 10. Q:
 - A:
- 11. Q:
 - A:
- 12. Q:
 - A:





Resources

This section of the *Living Document* provides additional resources that might be useful to you in developing skills and knowledge in the course topic area.

For any updates, job opportunities and open course announcements, please visit the <u>Ensembl</u> <u>blog</u>. If you are interested in organising an Ensembl training course at your institute, please visit the <u>Ensembl training website</u> or email the Ensembl Helpdesk at <u>helpdesk@ensembl.org</u>.

Ensembl sites

- Ensembl (vertebrates)
- <u>Ensembl Genomes</u> (non-vertebrates)
- Ensembl Rapid Release
- Ensembl GRCh37
- Ensembl COVID-19

Ensembl variation

- General information
- Tutorial: Explore variants within genes
- Ensembl VEP documentation

Ensembl training

For more in-depth Ensembl training, you can find links to online tutorials, walkthroughs and exercises below:

- <u>Ensembl Training</u> (walk-throughs and exercises)
- Ensembl: Quick Tour
- Ensembl: Browsing Genomes
- Ensembl Genomes (non-chordates): Quick Tour
- Ensembl REST API

EMBL-EBI training

- European Bioinformatics Institute (EMBL-EBI) website
- EMBL-EBI resources and tools
- The <u>EMBL-EBI Training portal</u> provides courses related to genomics and bioinformatics, including free <u>on-demand online training</u>, data resource tutorials and recorded webinars (available on the <u>training portal</u> or <u>YouTube</u>).





Ensembl in social media

- <u>LinkedIn</u>
- X (formerly Twitter)
- Facebook