



# Using the Ensembl Genome Browser

Friday, 3<sup>rd</sup> November 2023

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 <u>Ensembl Helpdesk</u>.

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:

- Navigate Ensembl using the web browser platform
- Access main data types in Ensembl: genomes, genes, genetic variation, regulatory features and comparative genomics
- View genomic regions and manipulate the view to add features.
- Explore information about genes and their sequences.
- Analyse genomic variants and associated phenotypes and your own variation data using the Variant Effect Predictor (VEP).
- View homologous genes and genomic regions.
- View functional elements involved in gene regulation and their activity in different cell types
- Export gene data in bulk using BioMart.
- Visualise your own data in the Ensembl genome browser.





# **Course Overview**

### Agenda

Time (EDT)	Friday, 3 <sup>rd</sup> November 2023
09:00 - 10:00	Introduction to Ensembl
10:00 - 10:45	Exploring genomic regions
10:45 - 11:00	Coffee break
11:00 - 12:00	Genes and transcripts
12:00 - 13:00	Lunch break
13:00 - 14:30	Variation and the Variant Effect Predictor (VEP)
14:30 - 15:30	Regulation
15:30 - 15:45	Coffee break
15:45 - 16:45	BioMart
16:45 - 17:00	Wrap-up

#### **Trainers**

Louisse Paola Mirabueno

### Demo, exercises and slides available to download

https://training.ensembl.org/events/2023/2023-11-03-GU\_browser

## Feedback survey

https://forms.gle/spx3P7Dp5zMrFMup9

We would really appreciate it if you could share your thoughts with us regarding these sessions. We are interested in your opinions, how you feel the experience has benefited you and how it could be improved. If you could find a few minutes to complete a short survey at the end of the last session it would really help us in improving the training we can deliver.





## **Questions**

If you have any questions/problems that you would like to share and are applicable to the whole class please write them below. A tutor will answer your question.

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.

 Q: What does this value 1,1,1 mean for the feature called "Phenotype or Disease under VEP analysis? My email <u>rr228@georgetown.edu</u>; Name: Rabin Roy

A: These numbers refer to whether any phenotypes are associated to the variants in the *Existing variant* column. Let's say you have rs0001 and rs0002 in the *Existing variants* column and 1,0 in the *Phenotype or disease* column. The numbers mean:

1 = a phenotype is association with the variant 0 = no phenotypes are associated with the variant In our example, a phenotype is associated with rs0001 and no phenotype information is available for rs0002. I hope this makes sense!

2. Q: The same question I have for somatic status, which shows 0,1,1 for the same variant

A: Please see the answer to Q1 above.

- 3. **Q**:
  - A:
- 4. Q:
  - A:
- 5. Q:
  - A:
- 6. Q:
  - A:
- 7. **Q**:
  - A:
- 8. Q:
  - A:
- 9. 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.

# Helpful websites

www.ensembl.org - Ensembl genome browser (chordates)
grch37.ensembl.org - Ensembl archive for browsing data associated with the human
GRCh37 genome assembly

www.ensemblgenomes.org - Ensembl Genomes genome browser (non-chordates)

www.ebi.ac.uk - EMBL-EBI website

www.ebi.ac.uk/services - EMBL-EBI databases and tools

#### Train online

EMBL-EBI provides an e-learning platform called <u>Train online</u>. Train online provides free courses on Europe's most widely used data resources, created by experts at EMBL-EBI and collaborating institutes. You do not need to have any previous experience of bioinformatics to benefit from this training. We want to help you to become a confident user of our data resources; we are not trying to train you to become a bioinformatician.

### Tutorials of interest might include:

- Ensembl: Quick Tour
- Ensembl: Browsing Genomes
- Ensembl Genomes (non-chordates): Quick Tour
- Ensembl REST API

#### Webinar series

The EMBL-EBI training team also runs regular <u>webinar series</u> featuring the EBI resources. See the <u>Training pages</u> for more information. You can also catch up on any webinars that you might have missed in <u>Train online</u> or on <u>Youtube</u>.