



Ensembl browser workshop

University of Ljubljana- 4th-11th May 2021

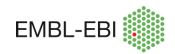
https://training.ensembl.org/events/2021/2021-05-04-Ljubljana

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!





Course Overview

Today:

14.00 - 15.30 Introduction to Ensembl

Break

15.45 - 17.00 Genes and transcripts

Thursday:

14.00 - 15.30 Variation and VEP

Break

15.45 - 17.00 Regulation

Next Tuesday:

14.00 - 15.30 BioMart

Break

15.45 - 16.30 Custom data

16.30 - 17.00 Wrap-up and Q&A

Trainers:

Emily Perry, Aleena Mushtaq

Webinar, Slides and Demo available to download:

https://training.ensembl.org/events/2021/2021-05-04-Ljubljana

Feedback survey

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.

The golden rules for efficient online training

We suggest everyone follows these few simple rules for the course to run as smoothly as possible:

- Mute all your microphones only unmute when tutor asks you to do that
- If you have questions during the course:
 - If you would like to ask a question to all the class: type your question in this shared reference document. Avoid typing in the Zoom chat as it is difficult to keep track there





When you are ready to move on from a practical/exercises please click the



yes button Raise Hand so that the tutor knows you are ready and can proceed with the course. Please remember there are more exercises than we have time for. You are not expected to complete all of them, but rather pick and choose the ones most relevant to your work and you are welcome to finish them in your own time.

Resources:

<u>www.ensembl.org</u> - 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

<u>Train Online</u> - EMBL-EBI e-learning for our databases and tools

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 and write your name.

 <Simon><How does ENSEMBL define insertion/deletion in terms of its lenght, for example when does a "deletion" becomes a structural variant (bigger chromosomal deletion)>

<Emily><If only there was an easy answer. There is no formal definition of short insertion/deletion vs SV. It is simply whatever it was submitted to the public database as. In the crazy old days before standardisation, people just picked and chose what they defined as, so some people would submit a 100bp deletion to dbSNP as a short variant, and others would submit to DGVa as an SV. I have heard the cut-off as 50bp and as 1000bp. Officially the definition is now that we know and can happily quote the deleted/inserted sequence in an short variant, but a structural variant is that we don't quote the sequence. I'm sorry I can't give you a more useful/sensible answer.>





- <Simon><Thanks Emily, your answer clarifies a lot of (my) confusion about this issue
 - <Tutor><Answer>
- 3. <Zala><Hello! I just wanted to emphasize how well designed your Coursebook is. All the "demos" and explanations are really well put together and helpful! I have no problem with completing exercises, and will definitely use the Coursebook for my future work.> <Aleena> Awesome! That's great to hear. But if you get stuck on anything, please ask away:)
- 4. <Tina><If I understood correctly, there are methylation probes included in Ensembl. Could you demonstrate how you can find the specific methylation probe and estimate the effect of hyper- or hypomethylation of associated region on gene expression?

Great course, thank you!

<Emily><I'm afraid we don't have annotation that can be traced back to specific probes. We have methylation data from RRBS and WGBS that can be turned on in the region views, but this is not mapped to any named probes. Nor do we have links between this and gene expression.>

<Tina> Thank you. The correlation between hypo- or hypermethylation status of probe linkage region and gene expression data would be interesting and useful information.

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<Tutor> Answer

- 12. <Name><Question>
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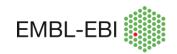




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- 28. <Name><Question>
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- 29. <Name><Question>
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Other resources

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.

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 run 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>.