

Using the Ensembl Genome Browser

23 October 2024

UCL

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:

- 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 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.

Course Overview

Trainers

Jorge Batista da Rocha

Slides and Demo available to download

https://training.ensembl.org/events/2024/2024-10-23-UCL_precmed_echan_browser

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.

Resources:

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 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<Tutor><Answer>

1. <Name> Is rs1801133 a missense variant in all transcripts of the *MTHFR* gene? What is the amino acid change? - text answer could use an update
2. <Tutor><Answer>
3. <Name>Why are the alleles for this variation in Ensembl given as G/A and not as C/T, as in the literature?

ENS always reports the forward direction for variation

But on the website it says Alleles G/A/C

- Multiallelic variants are possible through pop diversity, and be wary of the gene direction (forward / reverse strand)

So why we only pick the first 2 out of three alleles on the website?

Good Q! - we pick the reference allele based on the specific reference, which may not be representative of all population diversity. Pangenomics is a field aiming to address this issue.

4. <Tutor><Answer>
5. <Name> what do they mean by the amino acid change is A/V for allele A, and A/G for allele C. What is V

a. Amino acid- valine, G in this case is not guanine (nucleotide) but glycine amino acid

- 1.
6. When you search up a gene using its coordinates if the gene stretches across 50kb, do u always search 50kb forwards and backwards? Can you not just do 25kb forwards and backwards?
You could - but in this example I would use a bigger search - use at least 50kb up and down of the coordinate. Too short and you could miss your gene.

7. <Name><Question>
<Tutor><Answer>
8. <Name><Question>
<Tutor><Answer>

9. <Name><Question>
<Tutor><Answer>

10. <Name><Question>
<Tutor><Answer>

11. <Name><Question>
<Tutor><Answer>

12. <Name><Question>
<Tutor><Answer>

13. <Name><Question>
<Tutor><Answer>

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 [Train online](#). 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 [webinar series](#) featuring the EBI resources. See the [Training pages](#) for more information. You can also catch up on any webinars that you might have missed in [Train online](#) or on [Youtube](#).