

# Hands-on Biological Data Science with R

## Final Assignment



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## Upload Format

- The upload format of the final assignment is a **powerpoint slide deck**. It's not needed for you to upload your code or R notebook this time.
  - Name your file in the following format **'final\_assignment\_SURNAME\_FIRSTNAME'**
  - Make sure it can be opened before you upload.
- **Deadline: 30.01.2026 Fri, 23.59**

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## Data

- In your final assignment, you will use a GWAS summary statistics data computed for **asthma** as the phenotype. You can access the data downloading it directly from its source, which is the UK Biobank's Asthma GWAS:
  - <https://pheweb.org/UKB-TOPMed/pheno/495>
    - We recommend that you explore the webpage after your download as well.
- Due to data size being large, to be able to process the data more easily, you can apply a p-value filtering to the data before do your operations.



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## Instructions

- Using the GWAS summary statistics data:
  - Detect the **top hit**, and **the top hit when HLA region excluded**.
  - **1.** Create a Manhattan plot
    - Highlight the plot, showing your **2 hits (HLA and non-HLA) loci** (+/- 250kb region) with two different colors.
  - **2.** Create **2 LocusZoom** plots, one for each of your two hits (and their +/- 250kb).
  - **3.** Create a QQ-plot.
- Prepare a slide with your plots. Add additional slide pages (1 per hit) with a brief Gene & SNV information for both of your hits, mentioning your hits association, your phenotype in the literature with a few study samples. Don't forget to cite your sources.
- Why would we want to look at the hits as HLA / non-HLA region? What is special about HLA region ? Read up on it.
- **Bonus:** For a bonus point on your overall grade, include a brief summary (maximum of 2 slides) on a modern method in human genetics (e.g., single-cell sequencing, ATAC-seq, PheWAS, or molQTLs) at the end of your presentation and share it with the group.

# Thanks for your attention!

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