LMU KLINIKUM
Gemeinsam. Fürsorglich. Wegweisend.

HELMHOLTZ MUNICH

KimHellmuth Lab

DR. VON HAUNERSCHES KINDERSPITAL
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Hands-on Biological Data Science with R

Final Assignment

Final Assignment
Upload Format

- The upload format of the final assignment is a **powerpoint slide deck.** It's not needed for you to upload your notebook this time.
  - Name your file in the following format **`final_assignment_SURNAME_FIRSTNAME`**
  - Make sure it can be opened before you upload.
- **Deadline: 13.01.2025 Mon, 23.59**

Final Assignment Instructions

- Using the GWAS summary statistics data from exercise 3 and the 'brfssw24_assoc_withgenotype.csv' file you will find on moodle:
  - **1.** Create a Manhattan plot
    - Highlight the plot with two different colors showing the most significant hit with its +/- 250kb region, and the hit with highest absolute effect size (beta) that is significant with its +/- 250kb region.
  - **2.** Create 2 LocusZoom plots, one for each of two hits (and their +/- 250kb).
  - **3.** Create 2 boxplots, with the genotypes on the x- and the height on the y-axis, one for each of the two hits.
- Prepare a slide with your 5 plots. Add additional slide pages with a brief Gene & SNV information for both the hits, mentioning your hits association, your phenotype in the literature with a few study samples. Don't forget to cite your sources.

- **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.

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Thanks for your attention!

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