

## Additional instructions when preparing your HW1

- Literature style homeworks:
  - The bioinformatics literature-style homework assignments will need to be presented on 30<sup>th</sup> November.
  - The genetics literature-style homework assignments will need to be presented on 23<sup>rd</sup> November.
  - Presentations are prepared as follows: every group members should talk approximately for an equal number of minutes (minimum 5 minutes). Thereafter there will be about 15 minutes of questions and discussion.
  - For those students who select a practical GWAS paper via disgenet for instance, guiding questions are provided in Annex 1.
- All assignments are submitted the latest on 23<sup>rd</sup> November. This is the case if you selected a literature-style homework, and if you selected a classical Q&A homework. The same due date for both. Your written assignment (slides in case of a literature style homework) is submitted in group (1 submission per group) by sending your bioinformatics homework to [kristel.vansteen@uliege.be](mailto:kristel.vansteen@uliege.be) and your genetics homework to [fdequiedt@uliege.be](mailto:fdequiedt@uliege.be)
- Everything else regarding the homework organization can be retrieved from **Lecture 1\_ HW Organization** (see course website)

Annex 1: guiding questions to practical GWAS analysis paper selected via <https://www.disgenet.org/>

1. Describe the biological/research question(s) and put them in context.
2. What is the design of the study? (markers, subjects). Is it different from the designs seen in class? If so, what was the motivation to select a different design?
3. Which quality control procedures have been put in place? Are they in line with the Travemünde criteria? If not, was there a motivation given in the paper for adopting a different criterium, or can you come up with a motivation yourself? Be critical.
4. How did one make use of the concept of LD (linkage disequilibrium)? Was it used to reduce the number of tests? Was it used after the analysis to identify causal variants? Other uses?
5. What type of association test was carried out? Single locus at the time? Haplotype-analysis was considered as well? What is the possible advantage of performing a haplotype analysis? What are the drawbacks?
6. Was there a need to correct for population stratification? What is population stratification? How did one correct for it? Are there other ways?
7. Which multiple testing criterium was used?
8. Were the genetic association results supported by a replication analysis or a validation analysis? If so, what did it involve? What are the factors causing a non-replication? May it also be the existence of gene-gene interactions?
9. What are the final conclusions of the study and how much trust can be given to them (when looking at the replication/validation results)?
10. What type of follow-up analyses do the authors advocate? Do these analyses involve multi-omics data? Why or why not?