

# Large genetic studies in biobanks: from registries screening, to interpretation of GWAS and beyond

You can choose between two types of report: a literature based report or a dataset based report. The reports should be 5 pages, including figures and you have until the 25th of November to deliver them to Stephanie by email ([stephanie.hellard@uib.no](mailto:stephanie.hellard@uib.no))

If you want to pass the course to get the diploma and claim the 5 ECTs from your University you must deliver the report.

If you don't want the 5 ECTs we can deliver a certificate of attendance but not of passing the course.

The reports will be distributed to the most adequate person depending on which subject you choose. You will get feedback on your report. We won't give grades but a "pass / not pass". If you get "not pass" you will get the opportunity to rework on your report and resubmit it.

## Literature based report.

From published literature identify one complex trait/disease, use what you have learned during the course to describe how the disorder has been studied. You will need to search the literature yourself. Report should include as much as possible of the following points, and minimum 4 of the following points:

- a description of the trait/disease selected, and its epidemiology (prevalence, what is known about the heritability)
- selection of samples (biobanks, cohorts, clinical selection, single sample, meta-analyses, etc),
- GWAS (methods, basic analyses, results)
- genetic overlaps (with other traits and Bayesian analyses),
- annotation of associations,
- pathway analyses.

Examples of traits that would have several of these analyses: schizophrenia, bipolar disorder, smoking, Parkinson disease, anorexia nervosa, diabetes type 2, Alzheimer's disease....

If you would like to choose another one, please send your suggestion to Stephanie before starting and get your choice validated (just to avoid that you choose a trait which would not have enough information to produce a good report).

## Dataset based report.

If you have your own data that you would like to analyse with some of the tools presented you can present that as a report. The reports must include

- a (short) introduction and presentation of hypothesis,
- material and methods,
- results with figures and/or tables
- (short) discussion.

These report will be marked by the different lecturer depending on field of expertise, so it is a good occasion for you to get feedback on your analysis.

Examples:

- Selection of a sample on which a GWAS has been performed, which you need to QC, present basic analyses, preliminary Q\_Q plots, regional plots.
- Polygenic risk score using an existing GWAS summary statistics, test for PRS association in your trait of interest.
- Annotation of a GWAS at the gene level, functional level or pathway level

PS. we do not have datasets to provide for the dataset based reports, you must find it yourself. It can be a dataset that has been used in your group and you want to reproduce the analyses, or it can be a new dataset or it can be one that is publicly available like the ones that Tatiana has presented during her lecture.