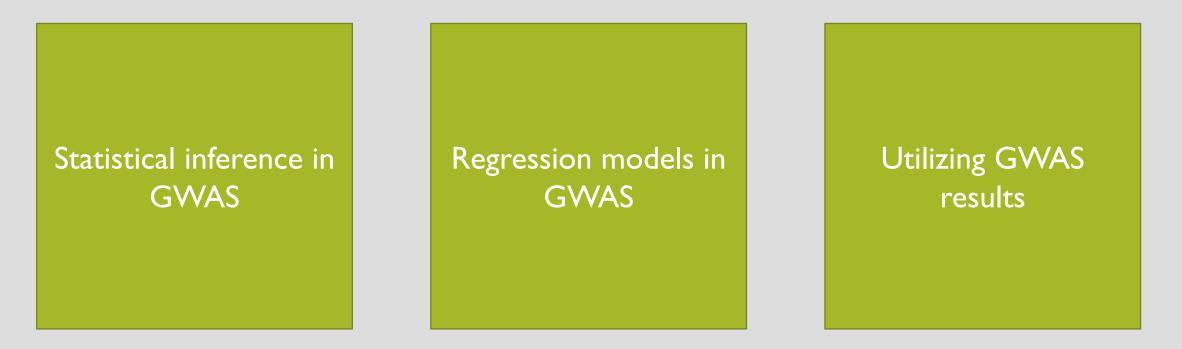
GWAS 2019

SUMMARY & EXAM

27.2.2019

THE POINT OF THE COURSE

How GWAS are carried out and what is the motivation behind each method?



Effect size, SE, P-value, power, Bayes factor, probability of association Covariates, confounders, joint model stepwise search, fine-mapping, PCA, relatedness, heritability with LMM Meta-analysis, LD-score regression, joint model from summary stats, polygenic scores, Mendelian randomization (idea)

STATISTICAL INFERENCE

- GWASI: HWE, GWAS effect size (beta and OR), SE, additive model (linear / logistic)
- GWAS2:What are P-values? What is the multiple testing problem in GWAS? How to decide significance thresholds?
- GWAS3: What is power? Which parameters it depends on and how? Why are well powered studies important?
- GWAS:4 When/how can we talk about probability of association?

GWAS REGRESSION MODELS

- GWAS 5: Relatedness and PCA, and their use in GWAS regression models?
- GWAS 6: Confounders and other covariates, such as colliders, mediators and independent covariates? How do independent covariates behave in case-control data analyzed with logistic regression?
- GWAS 7: Linkage disequilibrium? Relationship between marginal effects and causal effects? Joint model, stepwise search, fine-mapping?
- GWAS 8: Linear mixed model for heritability estimation and for GWAS? Idea of LD-score regression?

UTILIZING GWAS RESULTS

- GWAS 9: Meta-analysis, heterogeneity measures? Joint model from summary statistics? Polygenic risk scores?
- GWAS 10: Idea of genotype and summary statistics imputation? (no technical details)
- GWAS II: Idea of Mendelian randomization? (no technical details)
- GWAS 12: GWAS criticism?

EXAM

- Wed 6.3 12.15-15.00 in room D122 in Exactum, Kumpula Campus
- With pen & paper
 - you can bring to the exam a single, one-sided A4 sheet that you have prepared to summarize key points/formulas of the course
- Questions
 - Explaining concepts, drawing / interpreting figures, interpreting R output, simple numerical examples
 - No details will be asked about topics we haven't covered in exercises
 - Only general idea of imputation and Mendelian Randomization
 - No R-commands needed, algorithms (if any) can be written in pseudo-code
 - No calculus, no matrix algebra. Mathematical level: understanding formulas.