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?

Statistical inference in GWAS	Regression models in GWAS	Utilizing GWAS results
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

- GWAS1: 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 11: 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.