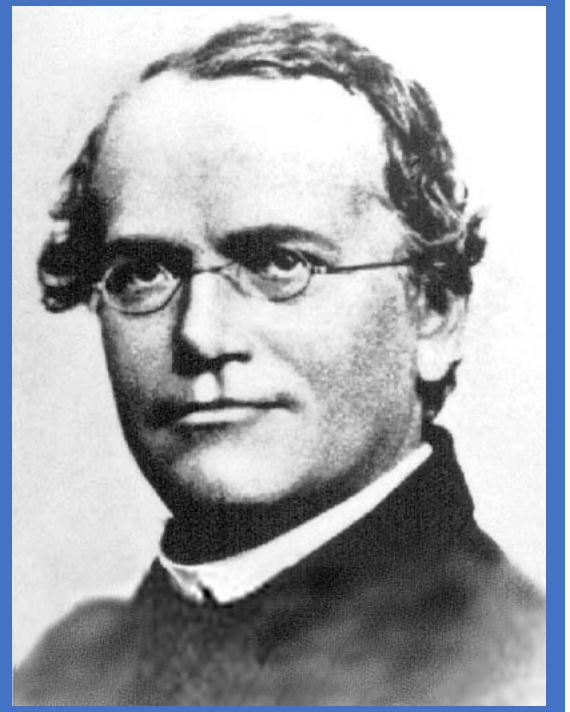
Two stories using OpenMendel

OpenMendel Workshop
ASHG Annual Meeting 2020



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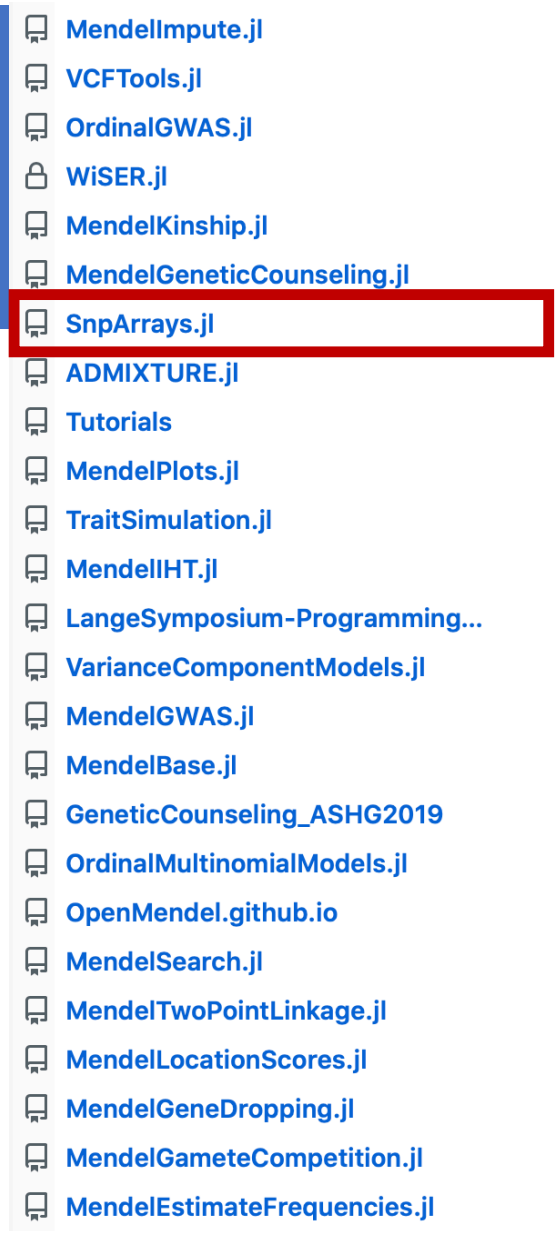
SNPArrays.jl

- GRM estimation incorporating ANCESTRY

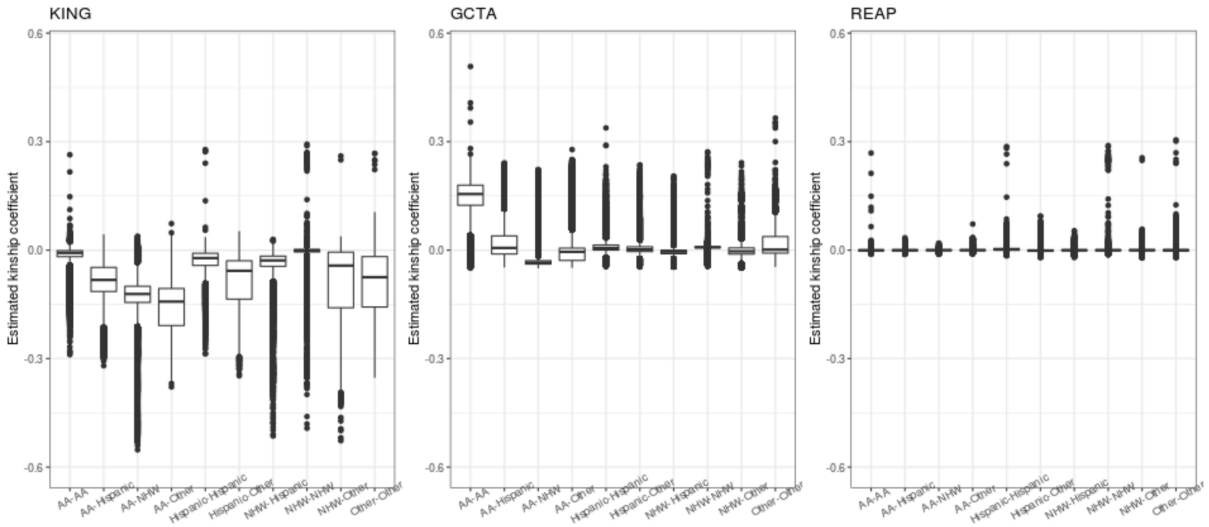
- After diabetes diagnosis, whether patients develop different types of vascular complications is due to genetics? If so, how much is due to genetics?
- We used the Action to Control Cardiovascular Risk in Diabetes (ACCORD) trial data to answer this question.
- Estimating genetic relationship matrix is the first step.



SnpArrays.jl

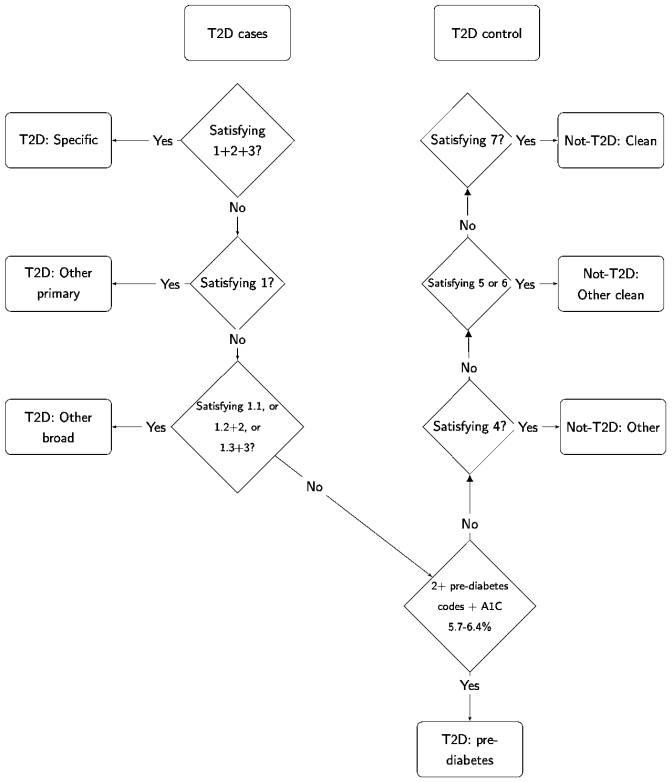
- GRM estimation incorporating ANCESTRY

REAP: http://faculty.washington.edu/tathornt/software/REAP/index.html
GCTA: https://cnsgenomics.com/software/gcta/
KING: http://people.virginia.edu/~wc9c/KING/manual.html
SnpArrays.jl: https://github.com/OpenMendel/SnpArrays.jl



- KING GRM is robust but was not positive semi-definite for down stream analysis.
- GCTA GRM removed >5000 individuals out of 6291 individuals.
- REAP took more than **6-7 hours.**
- *SnpArrays.jl* implementation took **around 6 minutes.**
- Filtering: *SnpArrays.jl* GRM 53 individuals were removed.
- ***Watch Dr. Hua Zhou's SnpArrays.jl presentation for more details.***

Ordinal Multi-categorical GWAS



- Disease phenotyping can be complex using data from EHR, e.g. T2D definition (left).
- It can generate multiple categories depending on the criterion used.
- There is ***NO*** ordinal multi-categorical GWAS tool that is available for biobank scale data.
- The current version of the US Department of Veterans Affairs' Million Veteran Program (MVP) contains genome-wide association study (GWAS) data > 800,000 veterans.
- Other large-scale biobanks include UK Biobank (> 500,000 individuals), BioVU (> 250,000), Kadoorie (> 500,000).

OrdinalGWAS.jl

- *Motivating Example: Hypertension GWAS in UKB*

- Hypertension GWAS on a UK Biobank sample (~185,000 individuals and 460,000 non imputed SNPs) took **181 minutes (~6 hours) on a standard PC**
- When outcome reduces to 2 categories, *OrdinalGWAS* is case-control GWAS
- ***Plink1.9*** **took 1999 minutes (~33 hours) for 2 categories, i.e., case-control, analysis on this dataset**
- *https://github.com/OpenMendel/OrdinalGWAS.jl*
- ***Watch Chris German's Ordinal GWAS section for details***