Journée des doctorants

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Outline

1. Main objective of the thesis

2. R packages

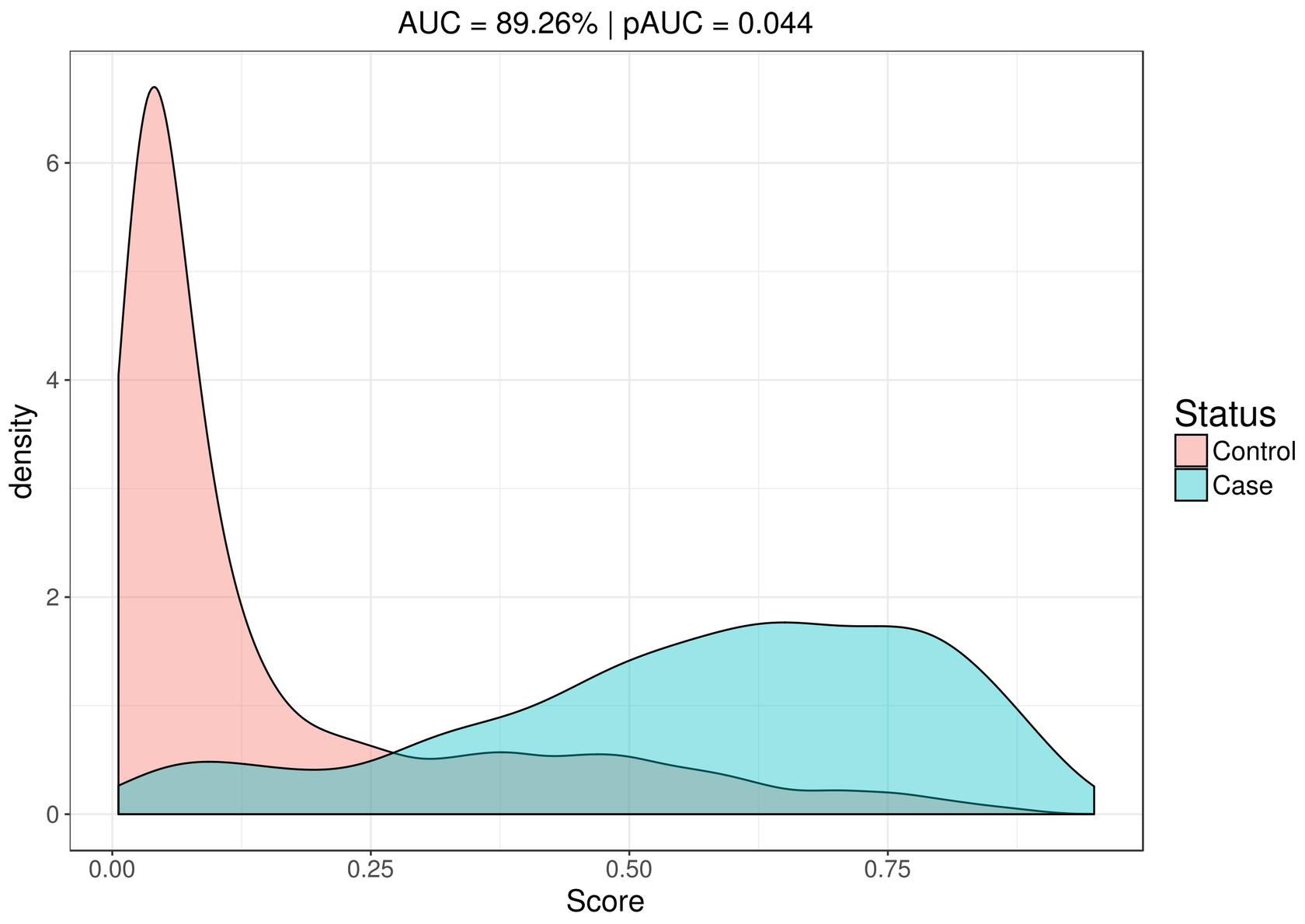
3. Ongoing paper

4. Future work

Main objective

Compute Polygenic Risk Scores (PRS)

in order to differentiate a healthy person from a diseased person



4 main difficulties

- Size of the data (dozens to hundreds of GB)
- Hundreds of thousands of correlated variables (variables with overlapping information)
- Generalization of models on different populations
- Integration of non-genetic data in the models

Big Data

Simpler solutions are easier to implement

What I want to be able to do

Data analysis on large-scale genotype matrices!

- Be fast to test many ideas quickly
    - code should be fast
    - I shouldn't have to make many conversions
    - it should be easily to combine multiple functions
- Not be restricted in my analysis
    - Basically use all I already know in R
- Work on my computer
    - I have 64 GB of RAM and 12 cores
    - Working on a server is not as easy as on my computer

Smooth and fast analysis!

Two R packages: bigstatsr and bigsnpr

Statistical tools with big matrices stored on disk

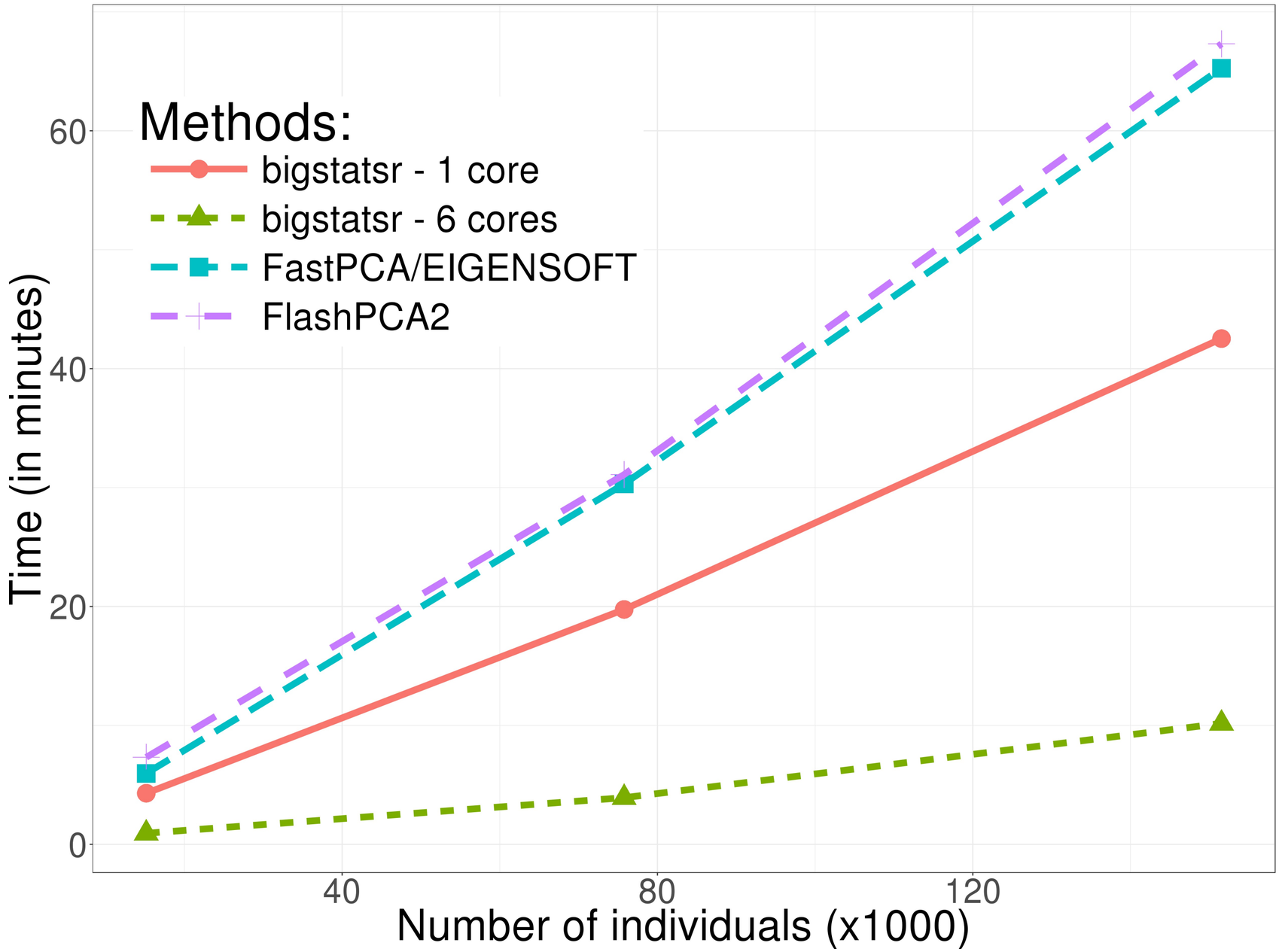
- **bigstatsr** for many types of matrix, to be used by any field of research
- **bigsnpr** for functions that are specific to the analysis of genetic data

Submitted Manuscripts

STATUS	ID	TITLE	CREATED	SUBMITTED
Pending decision	BIOINF-2017-1798.R1	Efficient analysis of large-scale genome-wide data with two R packages: bigstatsr and bigsnpr View Submission	31-Jan-2018	02-Feb-2018

Comparative performance

Computing partial SVD



Ongoing paper

Comparison of methods for computing PRS

(will be submitted by the end of April)

Recall of what we want to achieve

Predict a phenotype: pitfalls of the widely-used model

- Weights learned independently
- Correlation is taken care of heuristically
- Regularization is taken care of heuristically

A better solution?

We can use **statistical learning methods**.

For example, we can use logistic regression on all variables at once by using a clever implementation.

Future work

UK Biobank

UK Biobank

It is an extremely large dataset with

- genetic data
- clinical data
- environmental data

Prospects

- [Paper 3, before the end of 2018] training in one population to improve training and prediction in another population
- [Paper 4, in 2019, while writing the thesis] assess how can we combine the information provided by genetic data with clinical and environmental data, possibly in a non-linear way
- find a job in Machine Learning in some company

Thanks!

Presentation available at

https://privefl.github.io/thesis-docs/JDD.html

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Slides created via the R package **xaringan**.