Crowdsourcing Genome Wide Association Studies

Bastian Greshake and Philipp Bayer

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Overview

- Introduction
 - Association studies
 - Open GWAS
- Privacy
 - Privacy implications
 - Consequences
- Oiscussion
 - Outlook

What are GWAS?

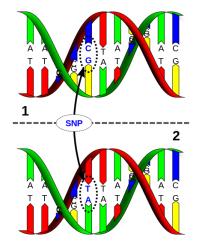
Genome-wide Association Studies

What are GWAS?

Association studies

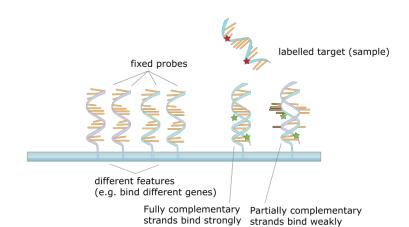
- Genome-wide Association Studies
- Link genetic variants (SNPs) to certain traits like eye or hair colour or to diseases like Diabetes, types of cancer

Single Nucleotide Polymorphism



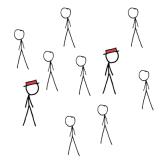
Source: http://en.wikipedia.org/wiki/File:Dna-SNP.svg

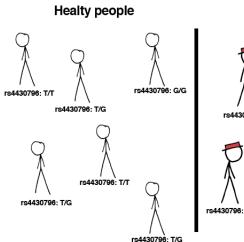
How to find SNPs?

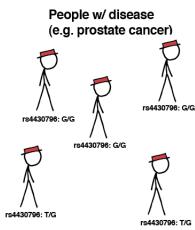


Source: http://en.wikipedia.org/wiki/File:NA_hybrid.svg

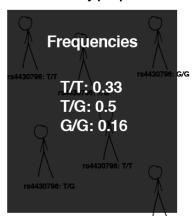
How do GWAS work?







Healty people

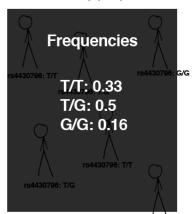


rs4430796: T/G

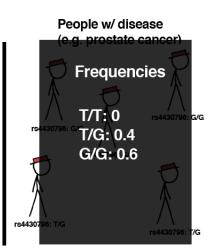
People w/ disease e.g. prostate cancer) **Frequencies** T/T: 0 rs4430796; G/G T/G: 0.4 G/G: 0.6 rs4430796; T/G

How do GWAS work?

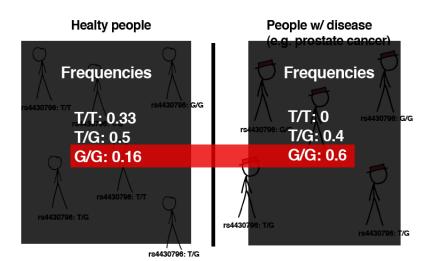
Healty people



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How do GWAS work?



Association studies

Some GWAS-examples

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- Kogan et al. (2011) linked rs53576 (G:G) to pro-social behaviour
- The Wellcome Trust Case Control Consortium (2007) linked 24 locations to 7 major diseases

Personalised GWAS

Association studies

 23andme: \$200 for a genotyping, 1.5 million SNPs + annotation

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- You get access to the raw data!

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Open GWAS

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- DTC-Companies like 23andme keep their data locked up
- At least 100.000 datasets!
- No way for scientists to access the data
- Some customers uploaded their data to the net

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- Possibly extremely bad consequences

What can happen with open data?

- Positive and negative consequences
- Possibly extremely bad consequences
- Up to you to decide whether you want to open your data

• More knowledge about yourself

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- Great data-source for citizen scientists

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- Personal SNPs very similar to parents and relatives
- You could be carrying a deadly disease
- Future research could have negative results

Open GWAS

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- So far: 78 genotypings and 188 users

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- Chance to take science into our own hands

Future of openSNP

• We've won the PLoS/Mendeley Binary Battle

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- Constantly improving the project

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- Trying to get funds for free genotypings

The end

Thanks for listening. Any questions?

References

Kogan, et al. (2011): Thin-slicing study of the oxytocin receptor (OXTR) gene and the evaluation and expression of the prosocial disposition. Proceedings of the National Academy of Sciences

Sladek et al. (2007): "A genome-wide association study identifies novel risk loci for type 2 diabetes". Nature 445 (7130): 881-5.

The Wellcome Trust Case Control Consortium (2007): Genome-wide association study of 14,000 cases of seven common diseases and 3,000 shared controls. Nature 447: 661-678.