Crowdsourcing Genome Wide Association Studies

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Overview

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

Association studies

What are GWAS?

Genome-wide Association Studies

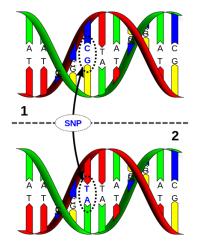
What are GWAS?

- Genome-wide Association Studies
- Link genetic variants (SNPs) to certain traits like Diabetes

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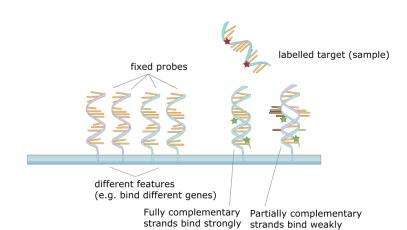
- Genome-wide Association Studies
- Link genetic variants (SNPs) to certain traits like Diabetes
- Compares groups carrying a certain SNP with groups without that SNP

Single Nucleotide Polymorphism



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

How do GWAS work?



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

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

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

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Personalised GWAS

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- You get access to the raw data!

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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

What can happen with open data?

Positive and negative consequences

Introduction

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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

Positive consequences

More knowledge about yourself

Positive consequences

- More knowledge about yourself
- Cheap, open science

Positive consequences

- More knowledge about yourself
- Cheap, open science
- Great data-source for citizen scientists

Consequences

Negative consequences

• People know more about you than you'd like

Negative consequences

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Privacy

Your boss, your insurance company...

Negative consequences

- People know more about you than you'd like
- Your boss, your insurance company...
- Personal SNPs very similar to parents and relatives

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Negative consequences

- People know more about you than you'd like
- Your boss, your insurance company...
- Personal SNPs very similar to parents and relatives
- You could be carrying a deadly disease

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- So far: 78 genotypings and 188 users

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- Open GWAS are the future of personalised medicine
- It's in the hands of users to make or break the situation
- 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

Future of openSNP

- We've won the PLoS/Mendeley Binary Battle
- Constantly improving the project
- 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.