# Crowdsourcing Genome Wide Association Studies

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28.12.2011

#### Overview

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

## What are GWAS?

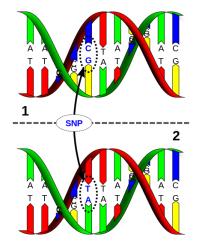
Genome-wide Association Studies

#### What are GWAS?

Association studies

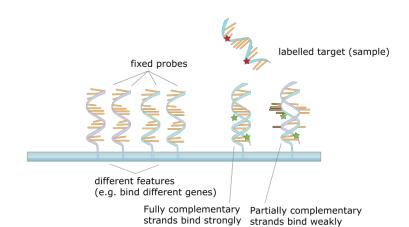
- Genome-wide Association Studies
- Link genetic variants (SNPs) to certain traits like eye or hair colour or to diseases like Diabetes, types of cancer

#### Single Nucleotide Polymorphism



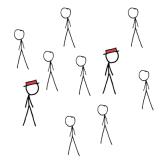
Source: http://en.wikipedia.org/wiki/File:Dna-SNP.svg

#### How to find SNPs?



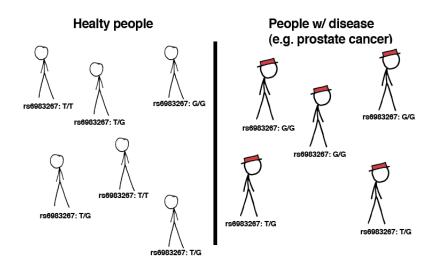
Source: http://en.wikipedia.org/wiki/File:NA\_hybrid.svg

#### How do GWAS work?



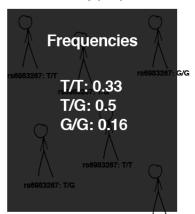
Introduction

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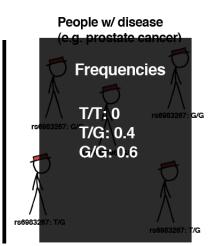


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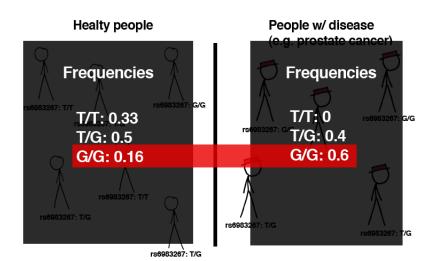
#### Healty people



rs6983267: T/G



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- The Wellcome Trust Case Control Consortium (2007) linked 24 locations to 7 major diseases

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- You get access to the raw data!

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- Some customers uploaded their data to the net

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- 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

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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

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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.