Crowdsourcing Genome Wide Association Studies

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Overview

What are GWAS?

- Genome-wide Association Studies

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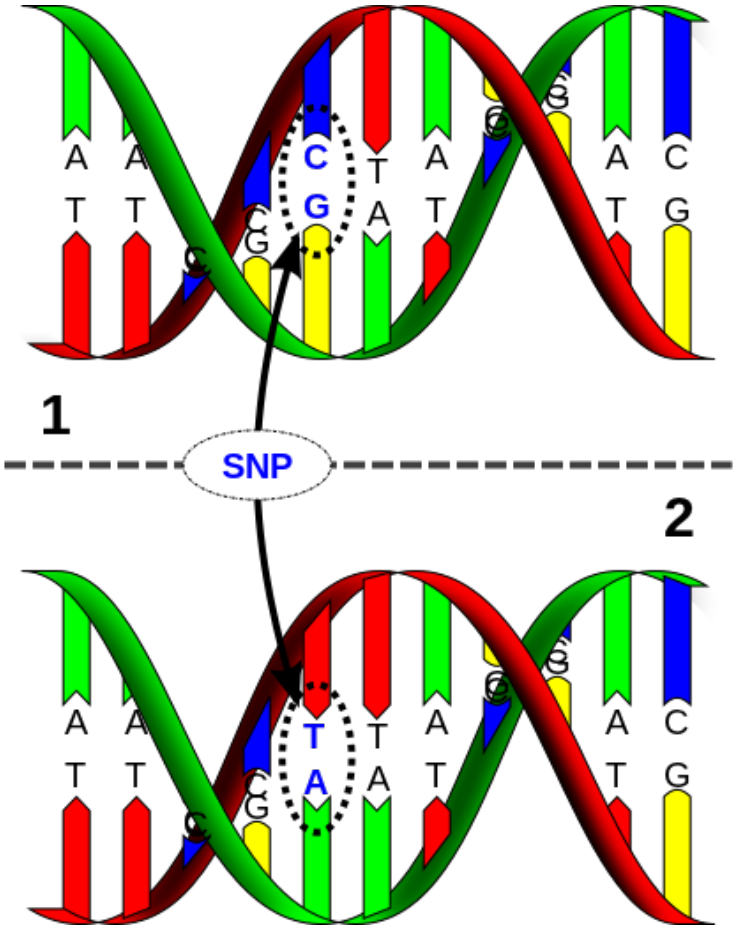
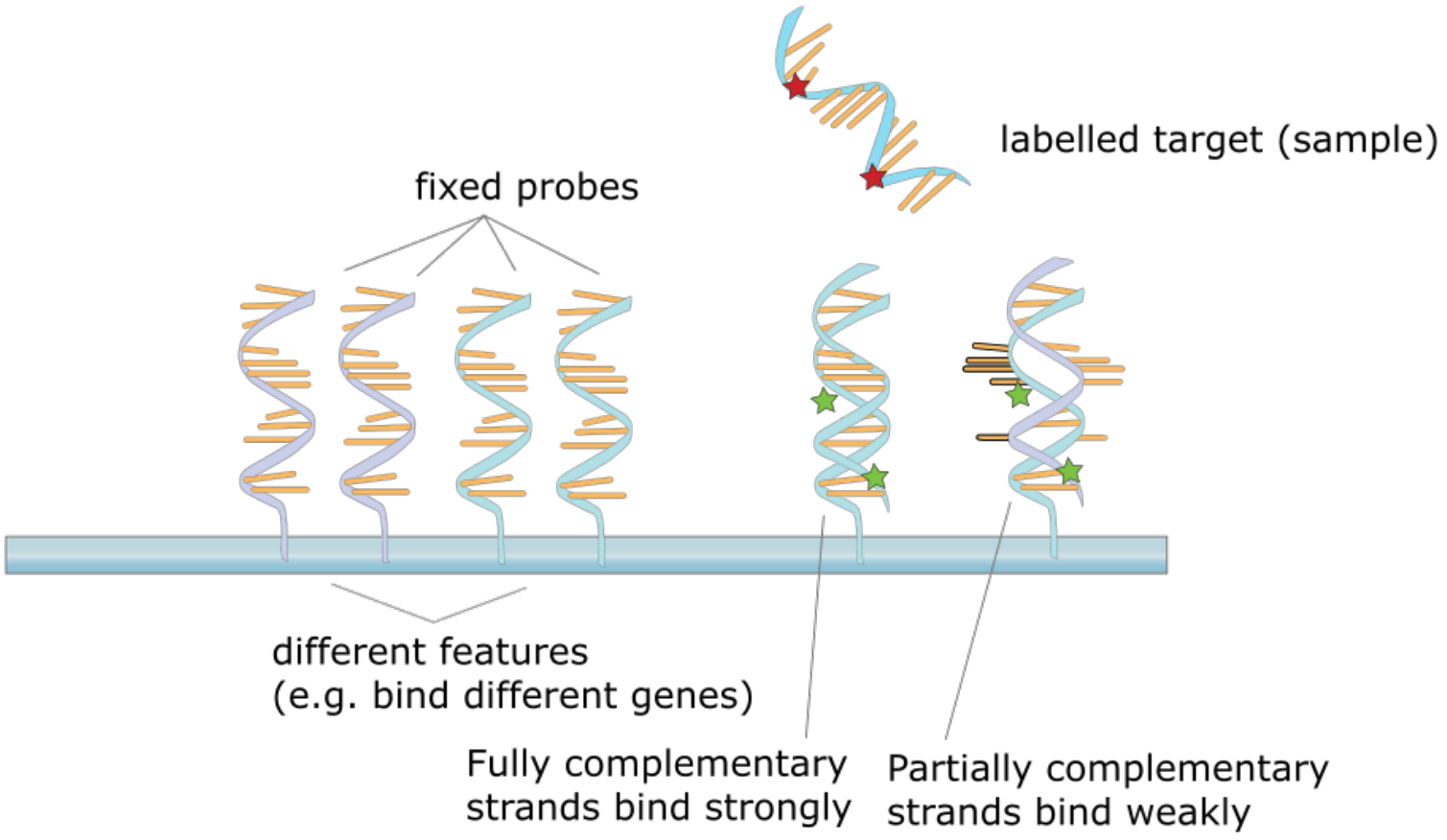
- Genome-wide Association Studies
- Link genetic variants (SNPs) to certain traits like eye or hair colour or to diseases like Diabetes, types of cancer

Single Nucleotide Polymorphism



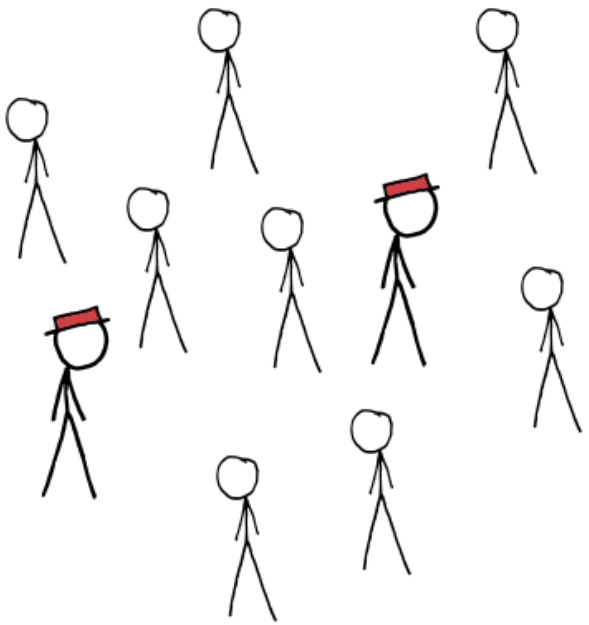
Source: http://en.wikipedia.org/wiki/File:Dna-SNP.svg

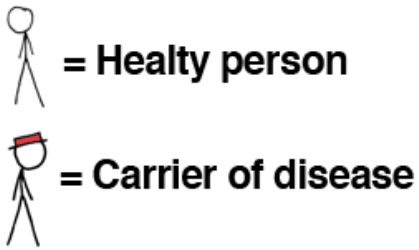
How to analyse SNPs?



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

How do GWAS work?





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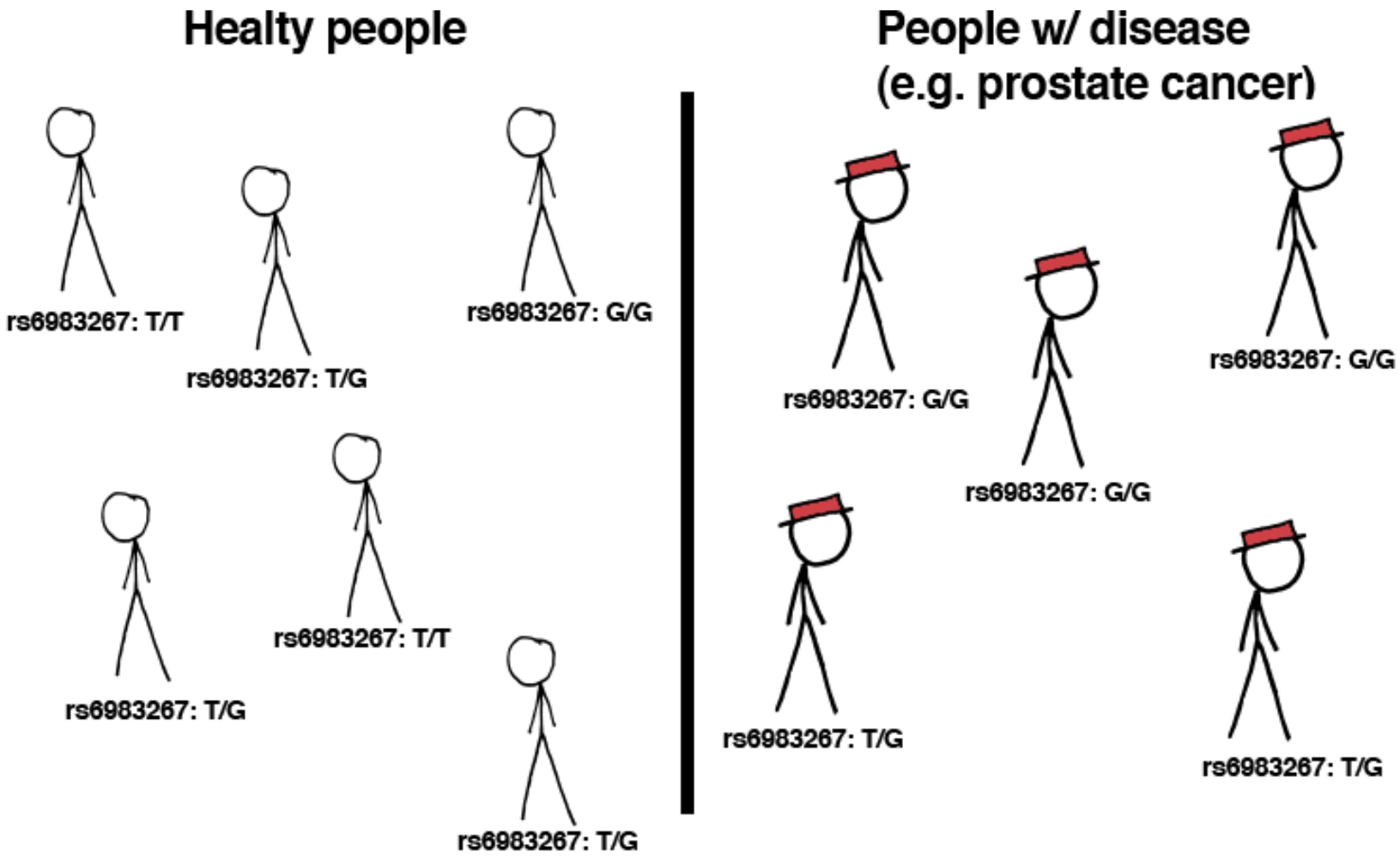

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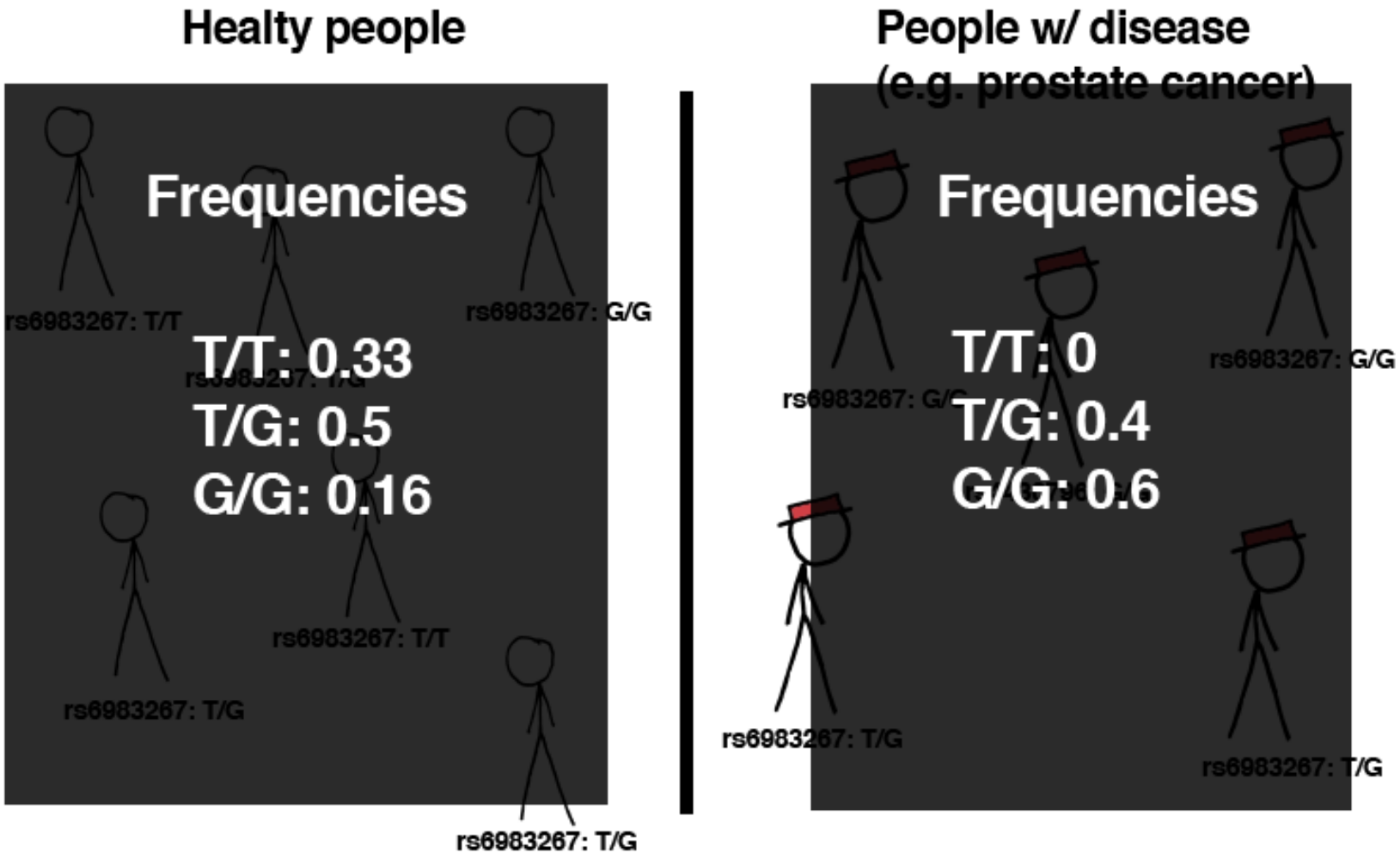

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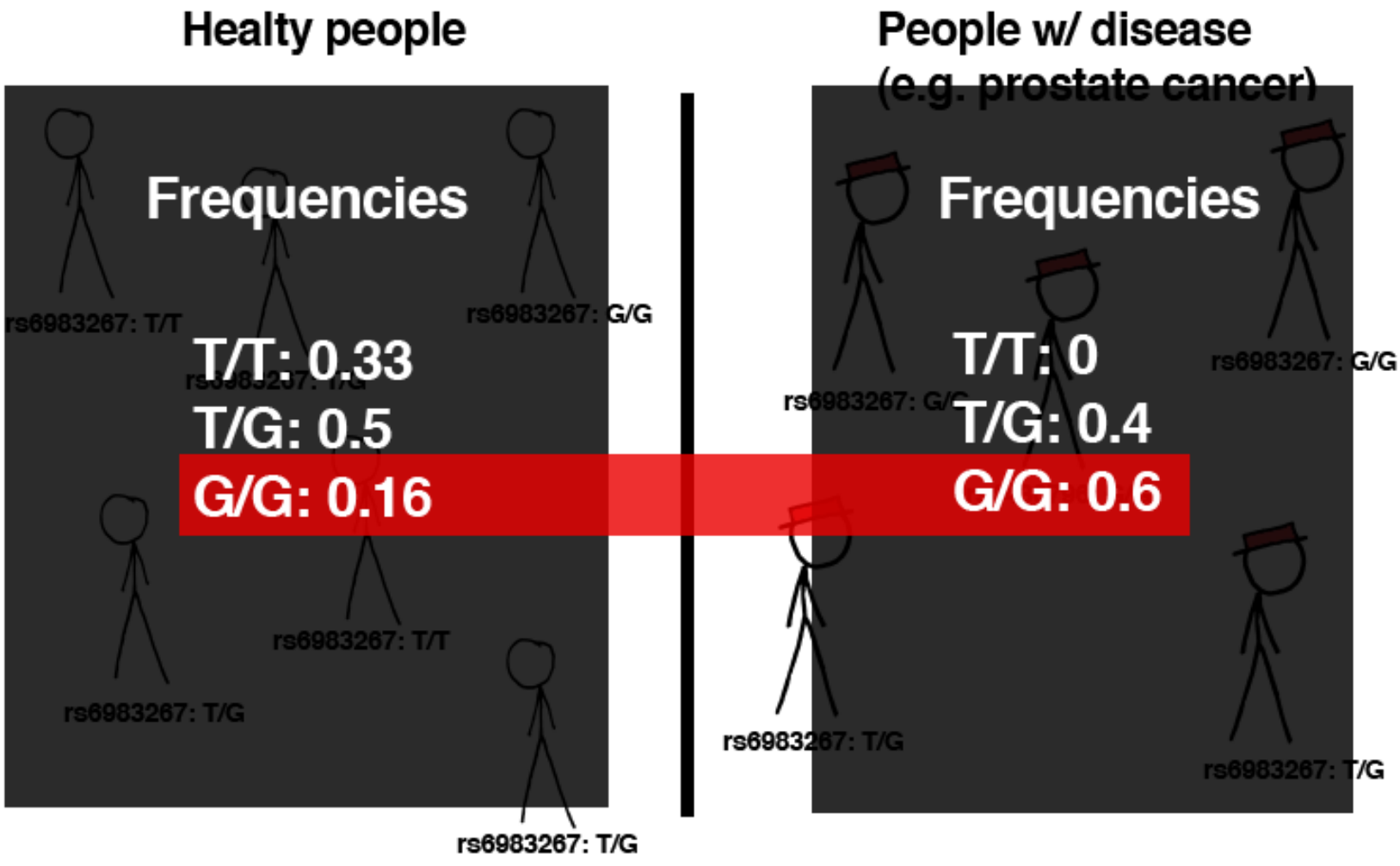

Some GWAS-examples

- Sladek *et al.* (2007) identified four gene locations linked to heightened type 2 diabetes risk

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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

Problems with GWAS

- Large enough sample size

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- Correlation != Causation

Putting GWAS to use

- Direct-To-Consumer genetic testing
- Analyse about 1 million SNPs and provide summary of disease risks & ancestry
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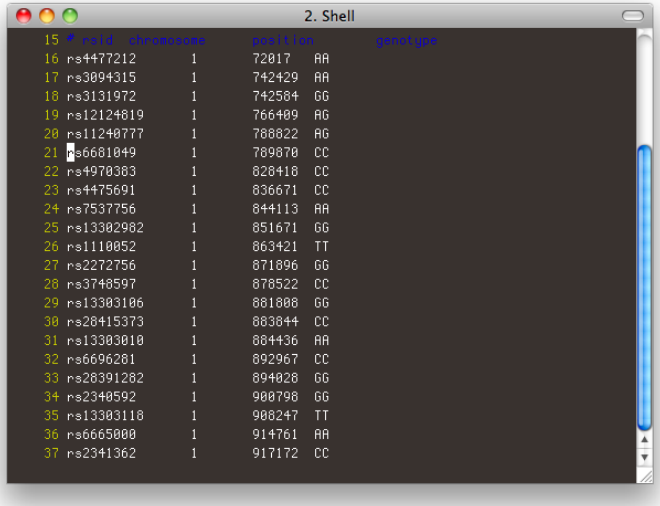
- Providers: 23andMe, deCODEme, FamilyTree DNA, ...
- You get access to the raw data!



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- 59 % of them share phenotypic information with 23andMe

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- Finding new associations for Parkinsons disease

Data sharing

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- The *Personal Genome Project*: Open data, but closed participation

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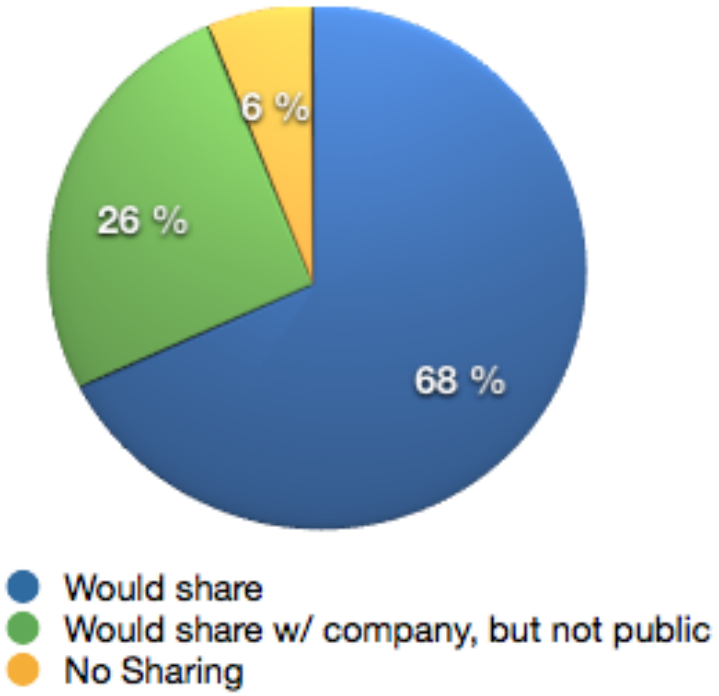

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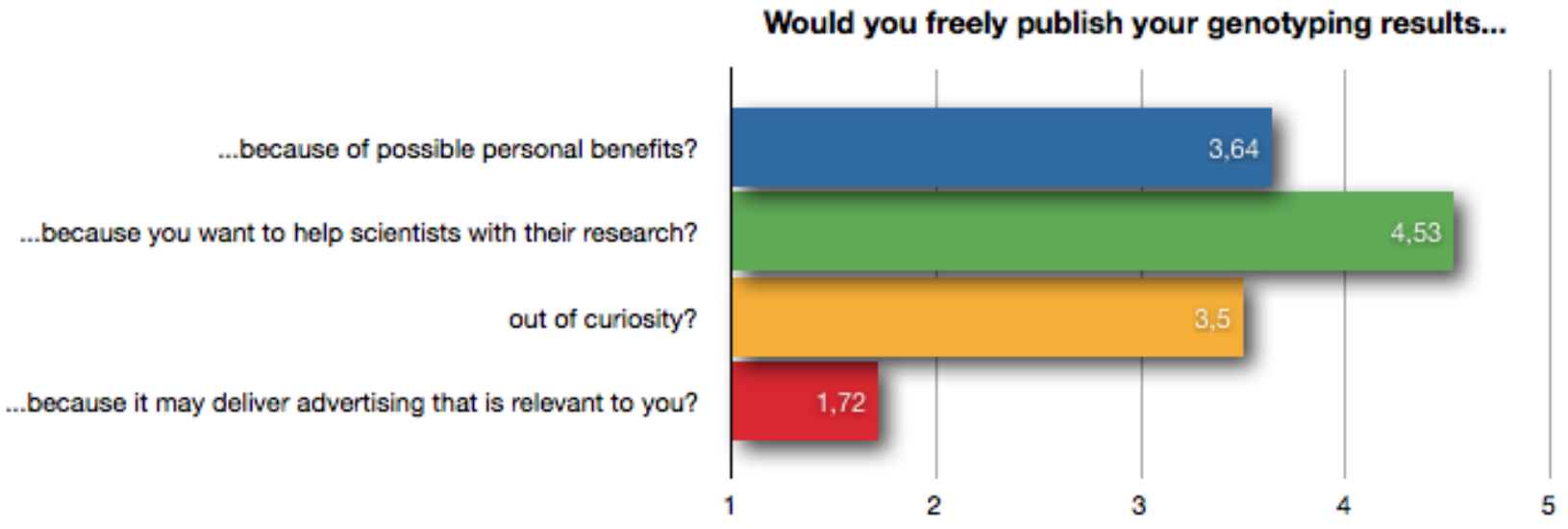

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- Positive and negative consequences
- Possibly extremely bad consequences
- Up to you to decide whether you want to open your data

Positive consequences

- More knowledge about yourself

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- Great data-source for citizen scientists

Negative consequences

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- 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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- It's in the hands of users to make or break the situation
- Chance to take science into our own hands

Future of openSNP

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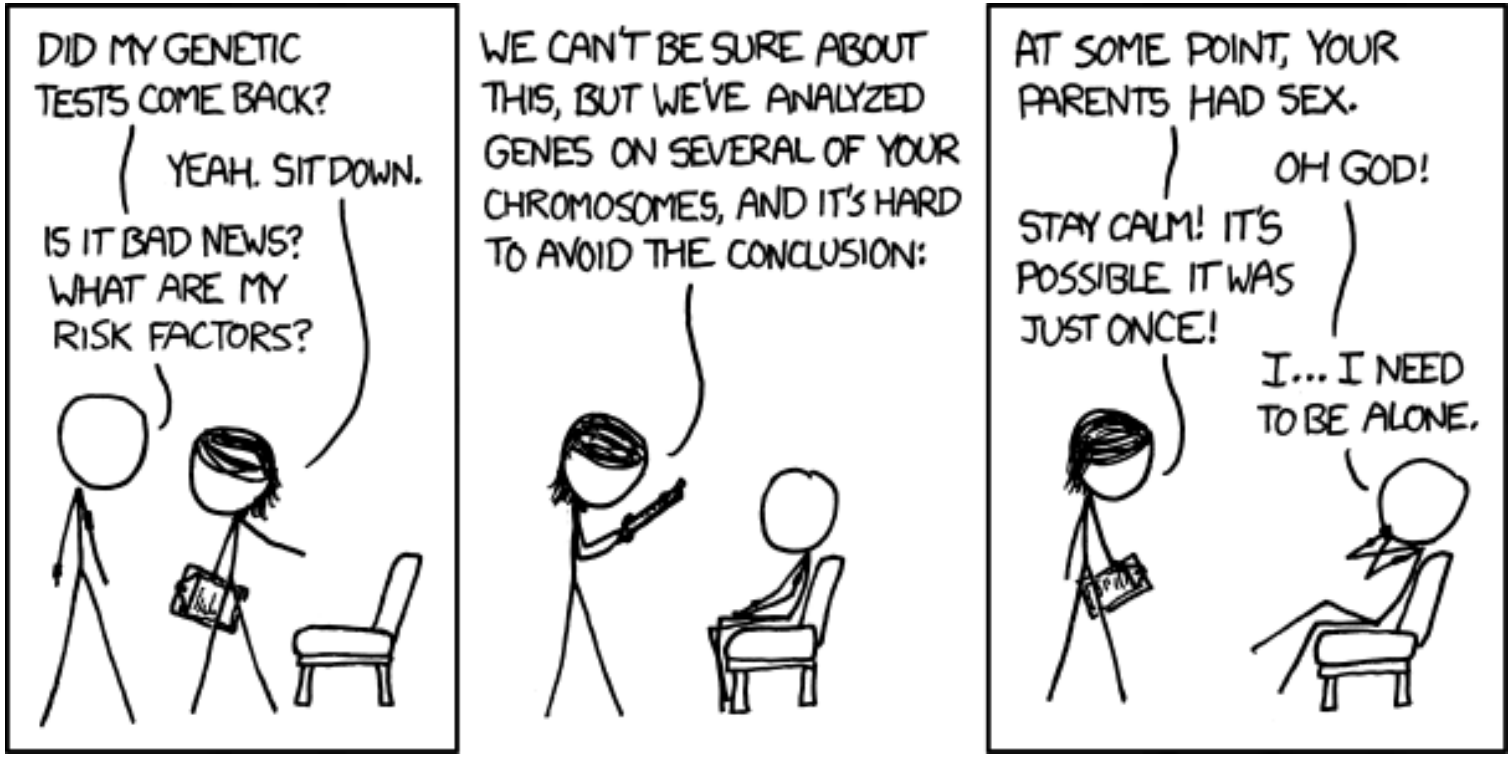
- Trying to get funds for free genotypings

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.