Crowdsourcing Genome Wide Association Studies

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Overview

What are GWAS?

- Genome-wide Association Studies

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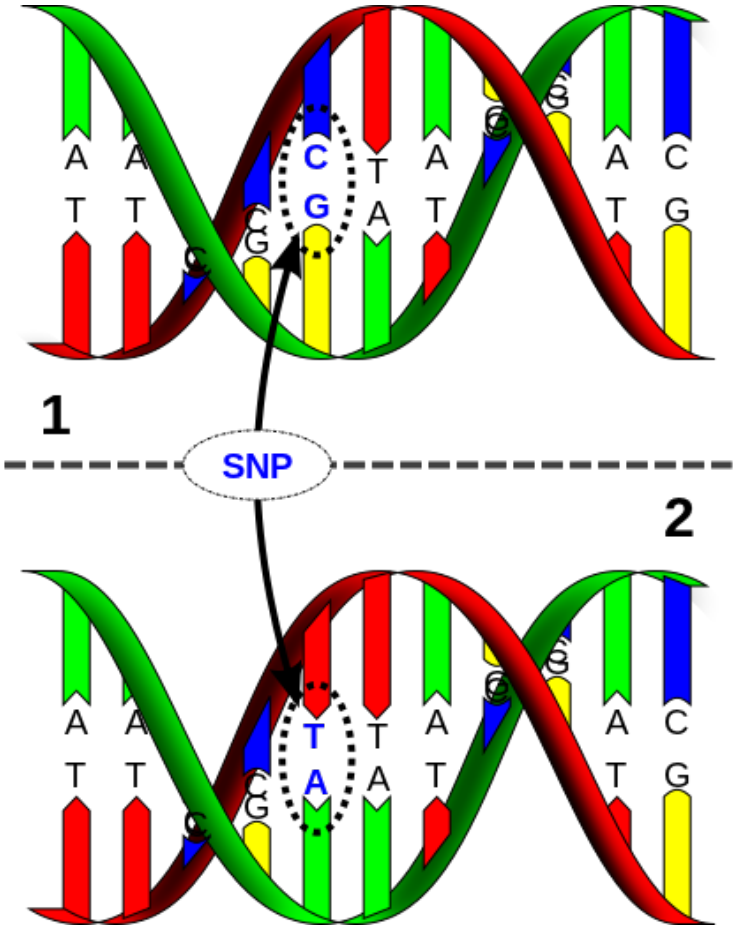
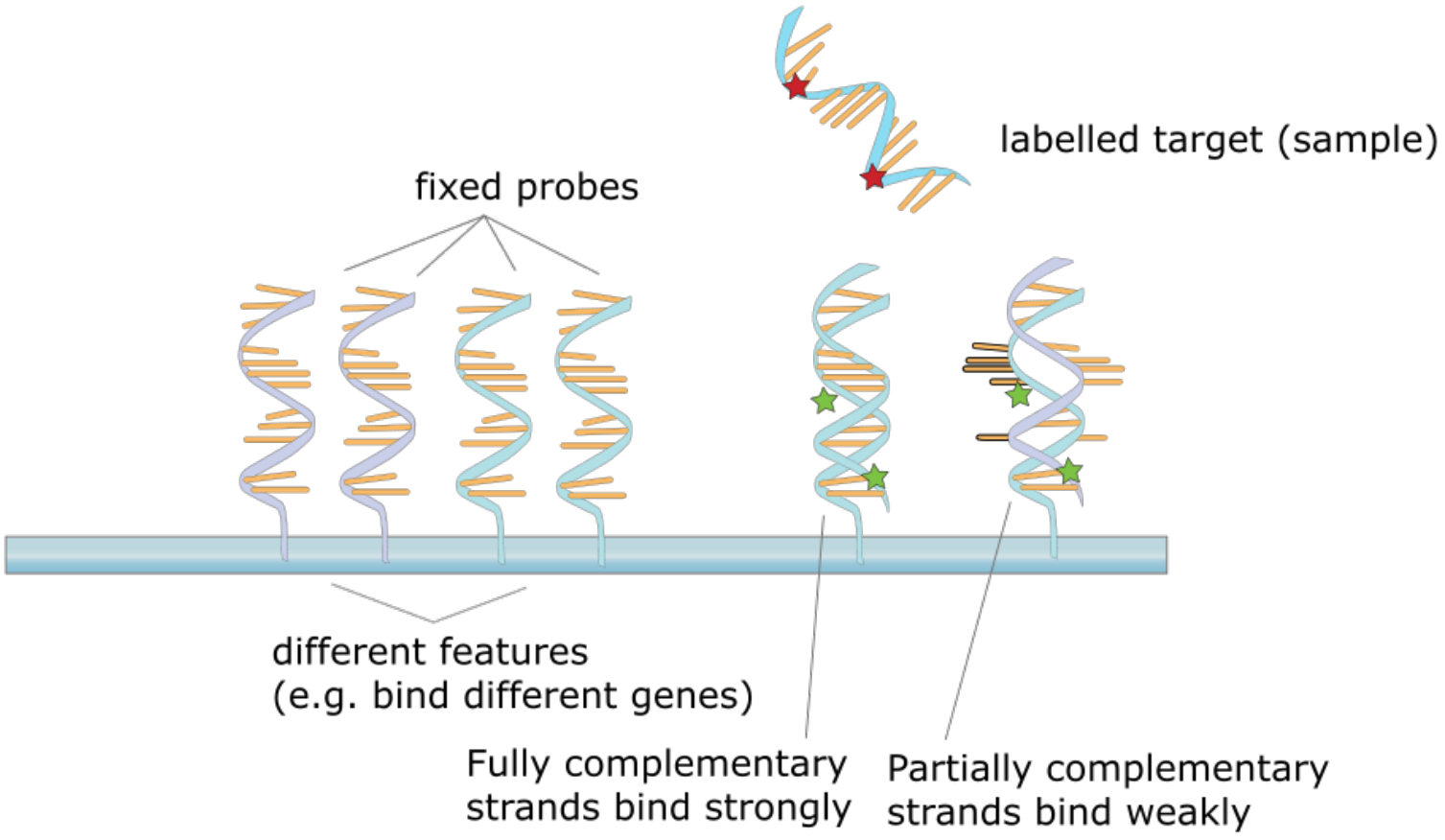
- Genome-wide Association Studies
- Link genetic variants (SNPs) to certain traits like eye or hair colour or to diseases like Diabetes, types of cancer

Single Nucleotide Polymorphism



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

How to analyse SNPs?



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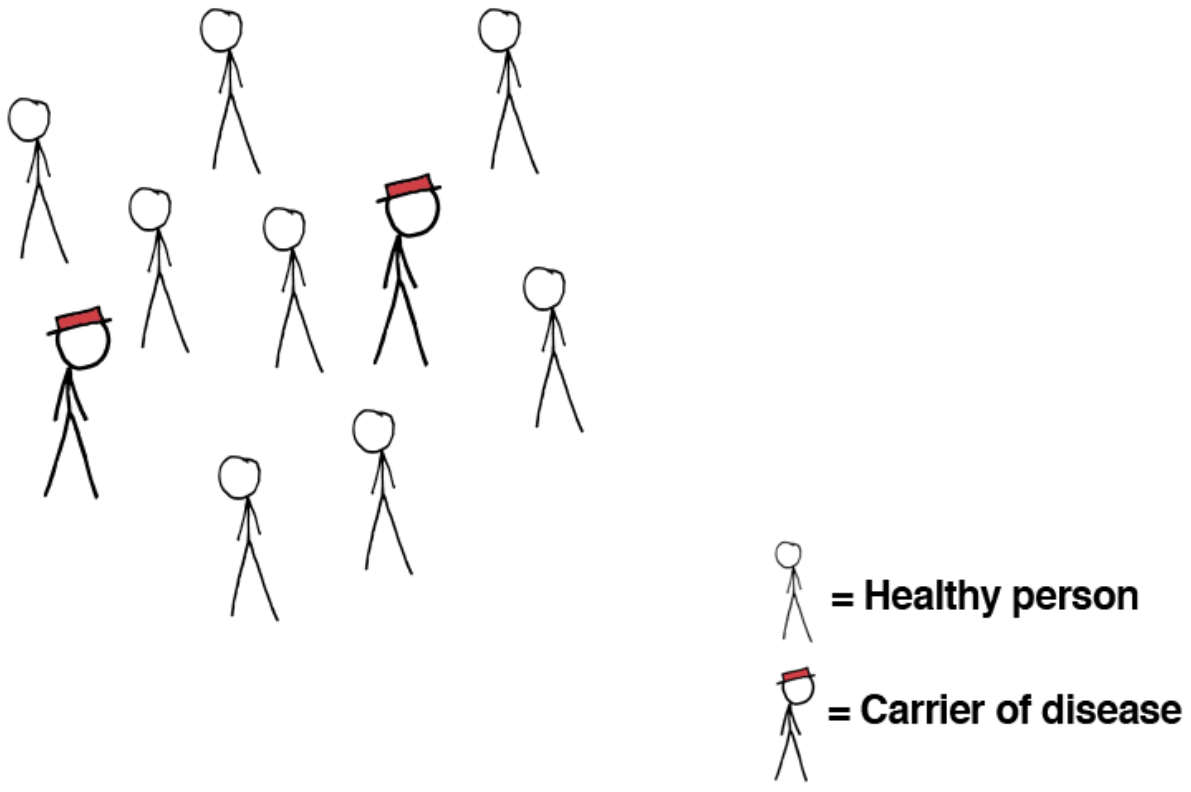
How do GWAS work?



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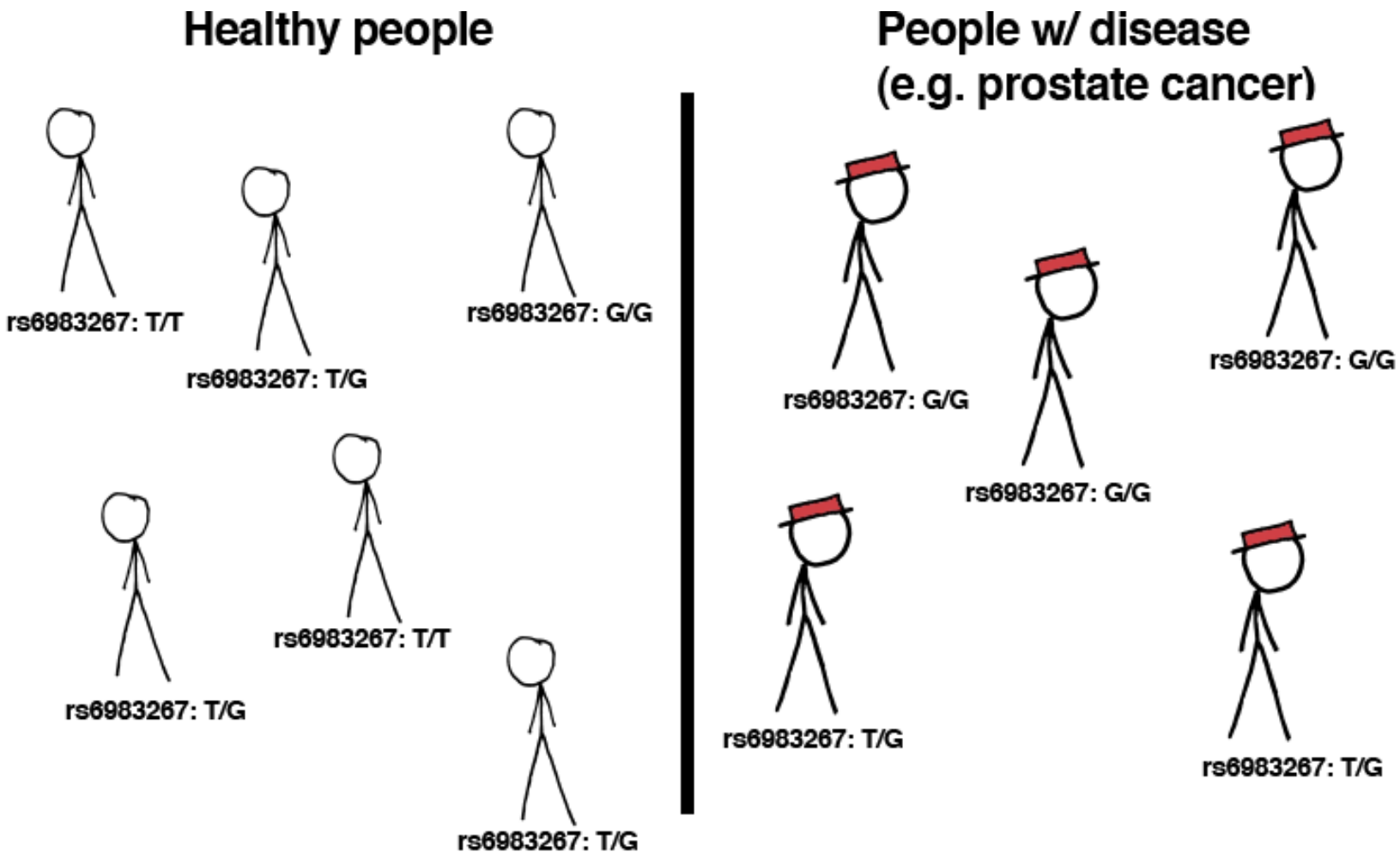

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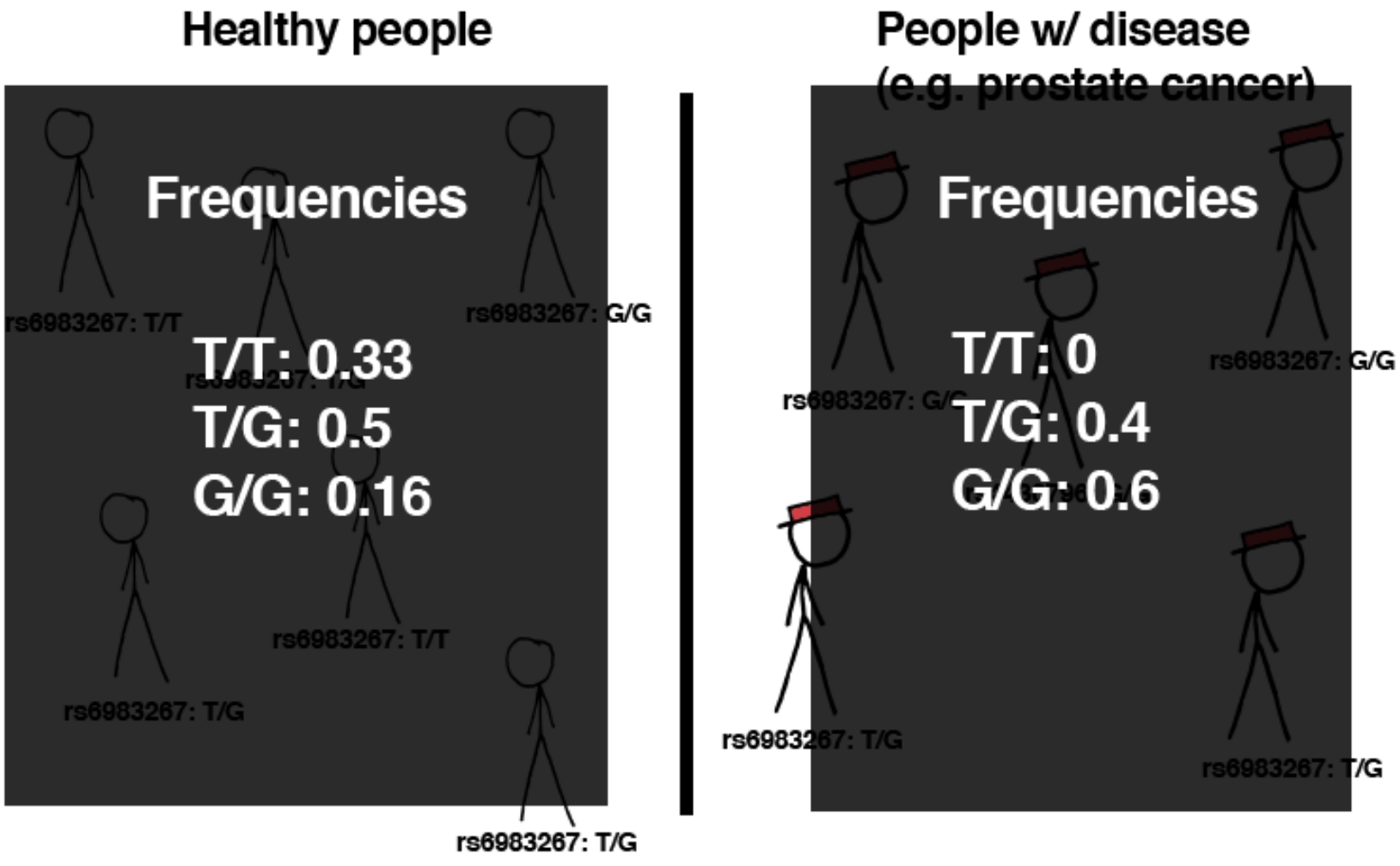

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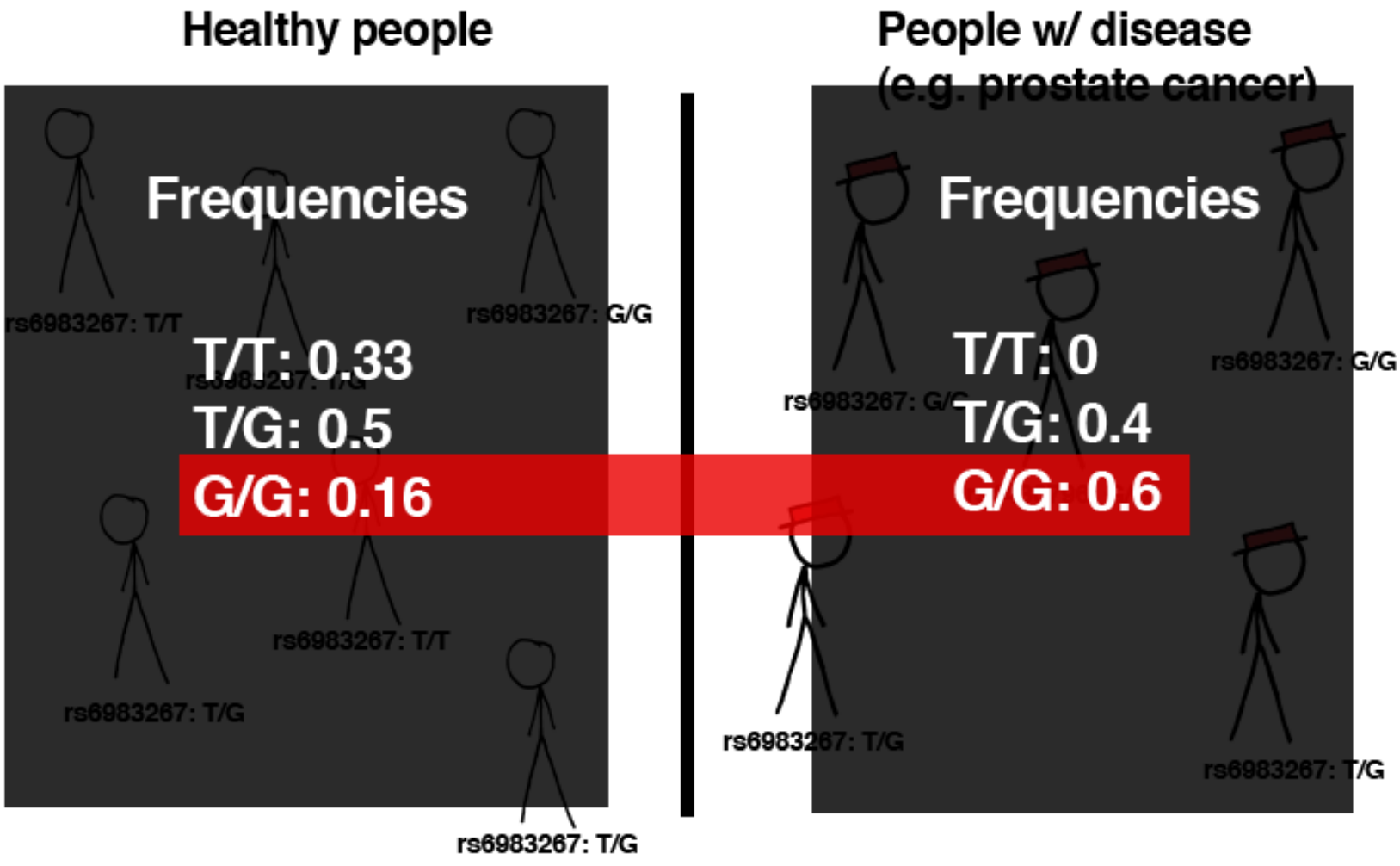

Some GWAS-examples

- 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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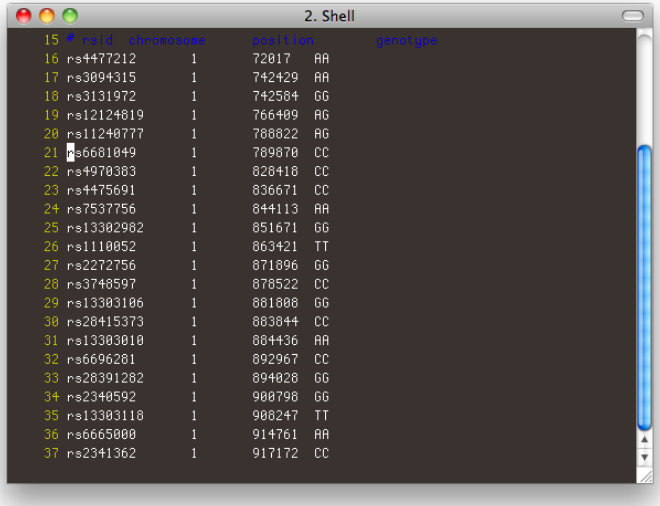
- Providers: 23andMe, deCODEme, FamilyTree DNA, ...
- You get access to the raw data!



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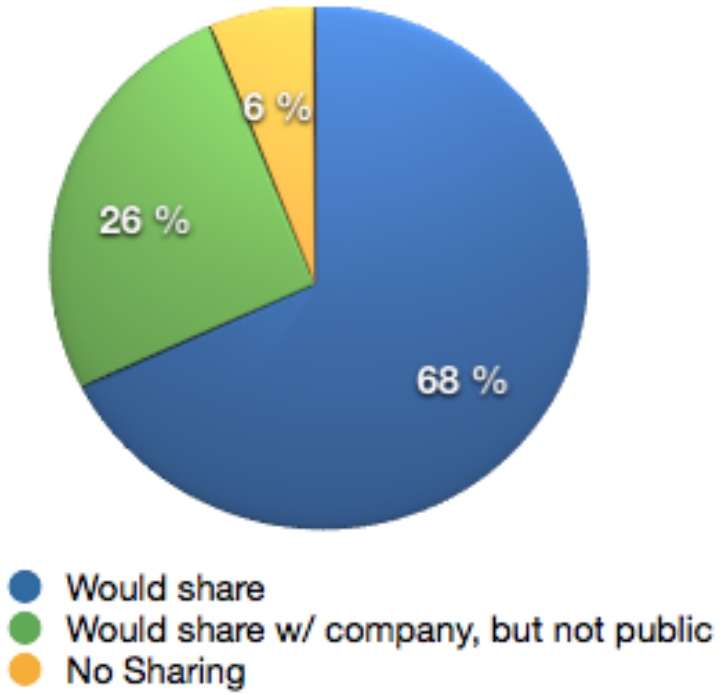
- The *Personal Genome Project*: Open data, but closed participation

Willing to share?



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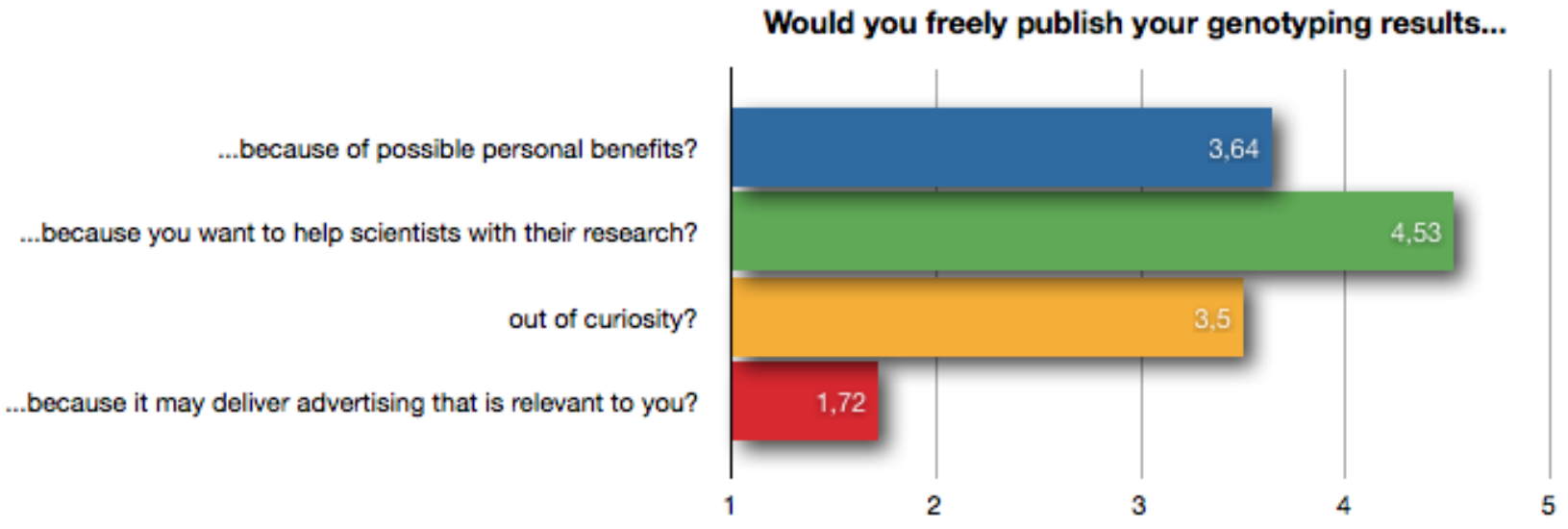

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- Positive and negative consequences

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

Positive consequences

- More knowledge about yourself

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- More knowledge about yourself
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- Great data-source for citizen scientists

Negative consequences

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- Knowledge isn't static: Future research could show new, negative (or positive) associations.
- Personal SNPs very similar to parents and relatives

Somebody Else's Problem? A case study

Me: rs1219648(A/A)

rs1219648(A/A): 1.64x risk for breast cancer
rs1219648(A/G): 1.20x risk for breast cancer
rs1219648(G/G): "normal" risk

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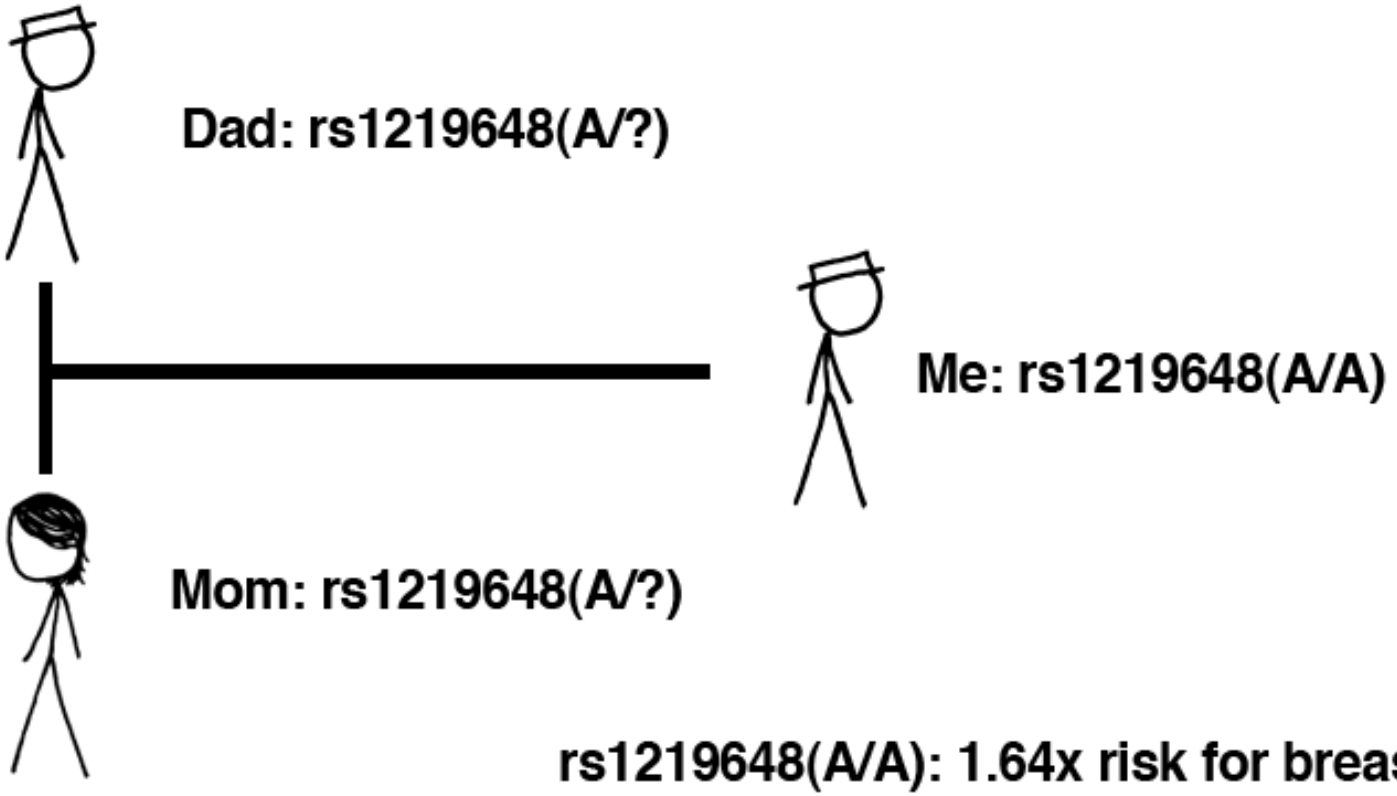

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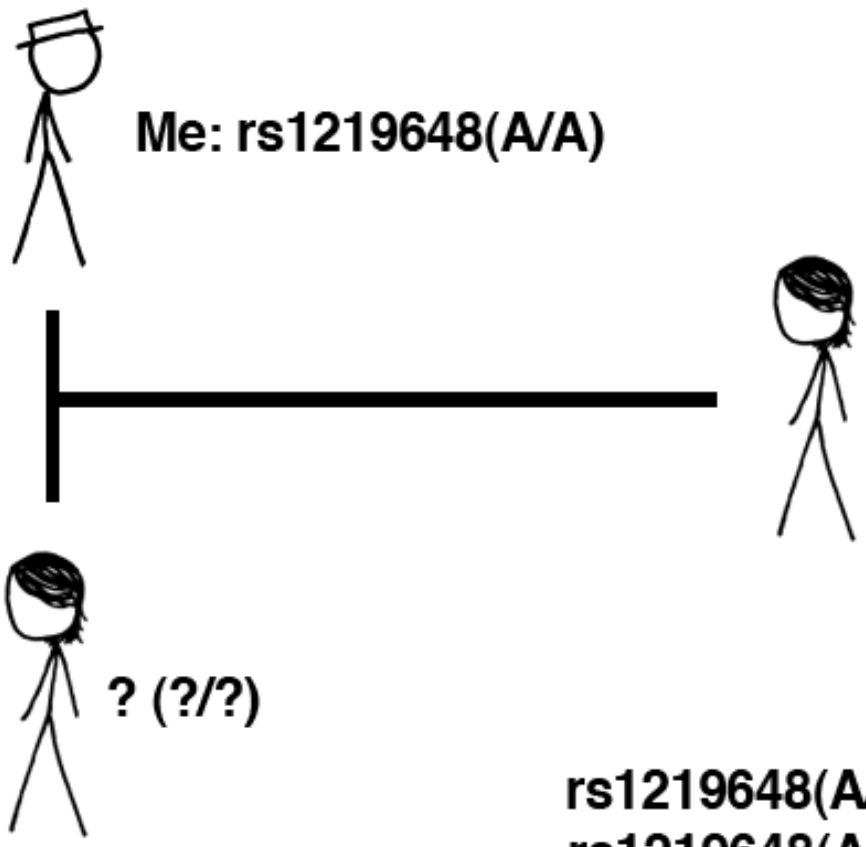

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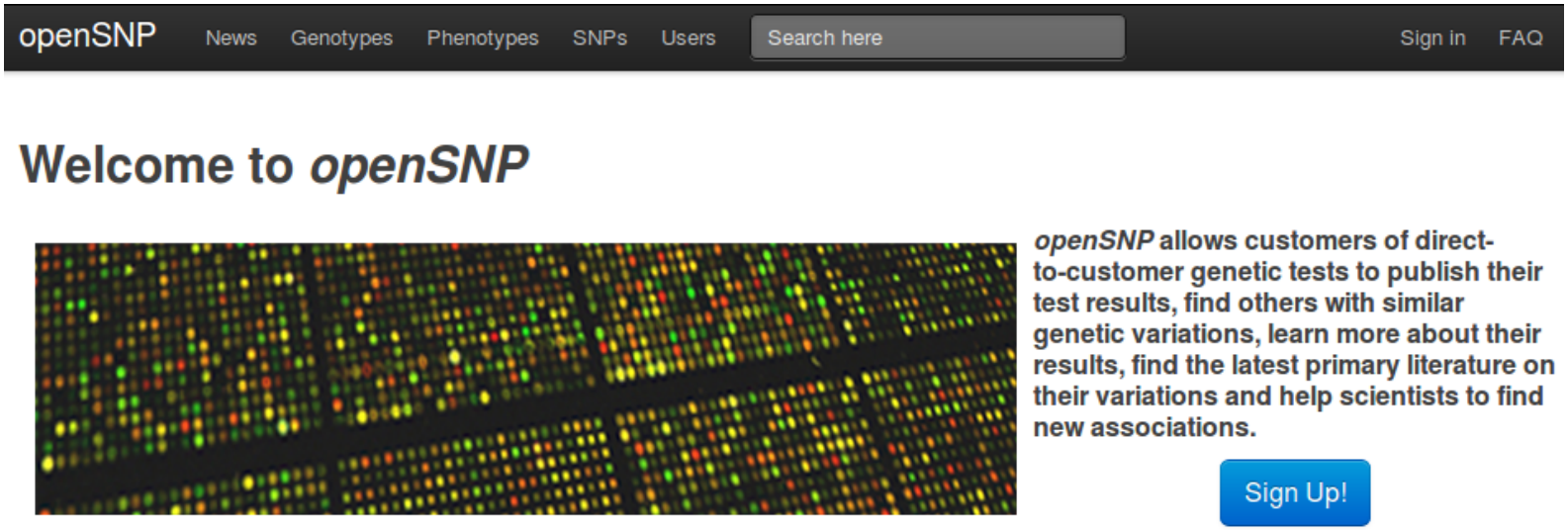
  - Germany: Gendiagnostikgesetz (GenDG, 2010)

For those who still want to share: Open GWAS



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- We've created openSNP.org
- open source repository for CC0-genotypings from 23andme, deCODEme and others

... continued

- Allows users to annotate with phenotypes (hair colour, nicotine dependence...)

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- So far: 81 genotypings and 207 users

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- Chance to take science into our own hands

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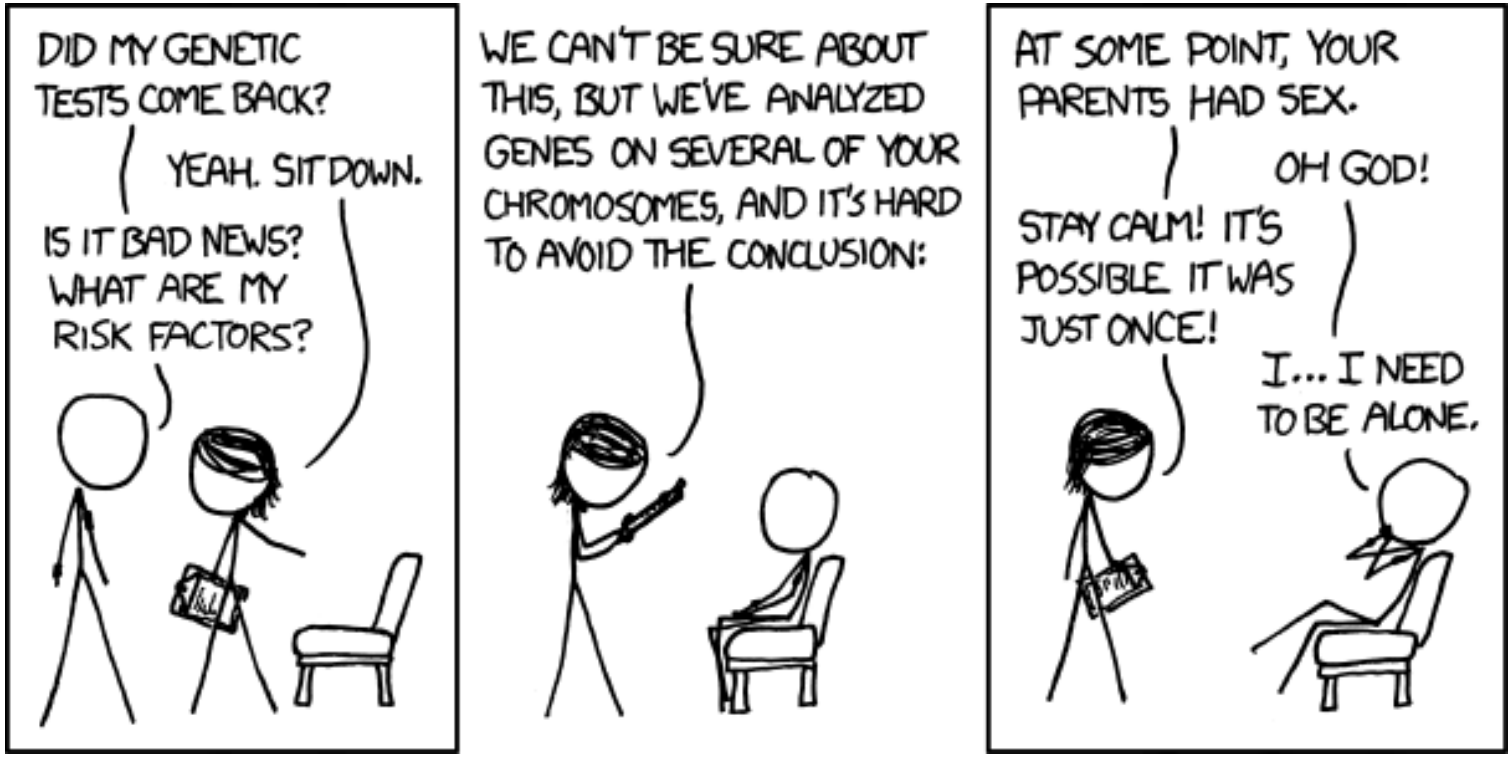
- Constantly improving the project (and are happy if somebody wants to help)

The end



Thanks for listening. Any questions?

References

Do *et al.* (2011) Web-Based Genome-Wide Association Study Identifies Two Novel Loci and a Substantial Genetic Component for Parkinson's Disease. PLoS Genetics 7(6): e1002141. doi:10.1371/journal.pgen.1002141

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